Behavioural Phenotype of Smith-Magenis Syndrome (SMS)

Individual characteristics and parental and school staff's experiences

by

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Summary

Challenging behaviours constitute a complex concept, and the term is used to describe several types of behaviours, in kindergartens, in schools, at home, and in society in general. Challenging behaviours often have a negative impact on the person's learning performance, but they also prove challenging for the learning environment. Persons with intellectual disabilities and other developmental disabilities are at elevated risk for developing challenging behaviours. Autism Spectrum Disorders (ASD) is referred to in the Diagnostic and Statistical Manual V as a dyad of impairments, including difficulties in social interactions and social communications, and restricted and repetitive behaviours, interests, and activities. Interest in studying ASD in genetic disorders has increased, and research has shown a higher prevalence of ASD in some genetic disorders than in the general population. The focus so far has been on the prevalence of and phenomenology in different syndromes, and further studies are required to tell us more about the differences in ASD phenomenology between ASD in genetic syndromes and idiopathic autism. Smith-Magenis syndrome (SMS) is a rare, complex genetic syndrome caused by an interstitial deletion of chromosome 17p11.2 or a mutation on the retinoic acid induced 1 (RAI1) gene. The disorder is characterised by intellectual disability, multiple congenital anomalies, obesity, neurobehavioural abnormalities, and a disrupted circadian sleep-wake pattern. Children and adults with SMS appear to have a unique phenotype that is especially challenging for both parents and professionals. These phenotypical characteristics or traits include sleep disturbances, self-injurious and maladaptive behaviours, stereotypies, and sensory integration disorders. This study seeks to bring new knowledge regarding the behaviours displayed by persons with SMS, in both their educational environments and their homes, by addressing teacher and parental perceptions/beliefs based on their experiences with these children. The aim of this project is to explore challenging

behaviours in a rare disorder to understand how and why the behaviours occur. A broader understanding of challenging behaviours is necessary to be able to manage or change the behaviours.
To answer the research questions exploring challenging behaviours in a rare disorder, a combination of approaches characterised as a multimethod design was used. This PhD thesis includes qualitative, quantitative and Q methodologies. In this project, the parents of 36 persons with SMS aged between 1 1/2 and 50 years old participated. A total of 18 were from Norway, 13 came from Sweden, and 5 were from Denmark.

The first aim of this PhD thesis was to explore the characteristics of SMS in the Scandinavian population, and the relations between the different specific characteristics (Article I and II). I found important information valuable to the staff responsible for the adaptation for persons with SMS in schools. In Article I, I found that more than 50% scored above the cutoff on the Social Communication Questionnaire (SCQ), and 96% scored in the mild-moderate to severe range on the Social Responsiveness Scale (SRS). A high level of autism spectrum symptoms is important to be aware of when planning learning for persons with SMS. The two most interesting findings in these two articles are the reversed gender ratio of autism spectrum disorder in SMS (Article I) and the decrease in behavioural and emotional problems with age (Article II). This decrease might also indicate that the work that schools are doing related to challenging behaviours is working towards decreasing these behaviours. Another finding in Article II is a negative relation between the Vineland Adaptive Behavior Scales (VABS) Activities of Daily Living (ADL) sub-scale and challenging behaviours, indicating that those with the most challenging behaviours had the poorest ADL skills.

The second aim was to investigate the experiences of and how school staff managed and handled the challenging behaviours of persons with SMS (Article IV). Two distinct viewpoints were found: 1) managing challenging aggressive and self-injurious behaviours in schools and 2) struggling with intense, non-physical challenging behaviours in schools. The school staff in viewpoint 1 experiences a range of challenging behaviours, both aggressive behaviours and acting out behaviours. The staff handles these behaviours and has a positive attitude towards both its work and the students. Staff has experienced that the students become angry at school, and they hit, scream, kick and engage in self-injurious behaviours, but the school staff still experiences it as positive and challenging in a good way to work with these students. School staff in viewpoint 2 experiences that the students are challenging to work with, especially because of the intensity of their behaviours, but the staff is positive towards its work and the students. However, the staff experiences demanding situations, especially if alone with the students. The view of staff loading on this viewpoint was that they experienced that these students have more non-physical behavioural problems, such as being very intense, craving attention and pushing buttons. The rationale of this aim was to investigate the school staff's beliefs regarding challenging behaviours and the types of challenging behaviours that students with SMS display in schools.

Regarding this aim, I also focused on how the school environment can adapt to meet the students' needs (Article V). The following four viewpoints were revealed regarding what the school staff needed in support to manage challenging behaviours: 1) in control, 2) struggling, 3) strugglers relying on parents and 4) support dependent. In the first viewpoint, the school staff received guidance and information regarding SMS and is handling its work well. The staff members enjoy their work and feel safe, although the students display challenging behaviours. In the second viewpoint, the school staff experiences are that it is difficult to work with students with SMS because of the challenging behaviours, and the staff struggles because it must do things slightly differently than with other students. The staff members also believe that it is difficult to inform parents of challenging behaviours because of the parents' feelings, but they are working well with the parents. In this group, they also lack support from the leadership at the schools and from other colleagues. In the third viewpoint, the school staff receives information and guidance from the parents and not as much from the school leadership and guidance from other than the parents (pedagogical centres, for example). In the fourth and last viewpoint, the school staff receives guidance and training regarding SMS and has support from the school leadership and colleagues. The staff members have not received much information regarding the disorder from the parents and are not working much with the parents.

The last aim of this thesis concerned the parents' experiences of having a child with SMS, with a focus on the challenging behaviours (Article III). Four themes emerged in this study: behavioural challenges displayed, parents' strategies for managing the challenging behaviours, parents' experiences of their own competence and parents' experiences of professionals' competence and understanding regarding children with SMS and their behavioural challenges. I found that parents of children with SMS experience being exposed to severe challenging behaviours from their children. The parents believe that they experience more misunderstandings with professionals and that the challenging behaviours increase because there are some specific characteristics of SMS that professionals are not aware of or do not consider in their support services.

The use of a multimethod design and data from different sources has provided the opportunity to explore challenging behaviours in SMS from different perspectives. It has been useful, and new knowledge has been found by exploring the same topic with several methods and across different informants (school staff and parents) and settings (home and school).

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PART I

1 Introduction

1.1 Challenging behaviours

Challenging behaviours constitute a complex concept, and the term is used to describe several types of behaviours, in kindergartens, in schools, at home, and in society in general. There are different definitions of challenging behaviours, but most of them have in common that they describe behaviours that break or go against social norms and the rules of society (Ogden, 1993, 2009). One of the best known definitions of challenging behaviours comes from Emerson (2001 p. 3): "Culturally abnormal behaviour(s) of such an intensity, frequency or duration that the physical safety of the person or others is likely to be placed in serious jeopardy, or behaviours which is likely to seriously limit use of, or result in the person being denied access to, ordinary community facilities." Challenging behaviours are often categorised along different dimensions, such as topography, frequency and intensity (Lloyd & Kennedy, 2014). Another way to categorise challenging behaviours in is by cause, function or motivation by the consequences (harm to self or others, or material damage) or the way other react to the challenging behaviours (von Tetzchner, 2003).

There seem to be two different fields within the research and practise regarding challenging behaviours: one field concerns challenging behaviours in persons without intellectual disabilities (ID), and the other field concerns challenging behaviours in persons with ID. The differences in describing challenging behaviours in persons with or without ID are further presented in Chapter 2 within the contextual framework.

Challenging behaviours often have a negative impact on a person's learning performance, but they also prove a challenge for the learning environment (Roland, Øverland, & Byrkjedal-Sørby, 2016). Challenging behaviours can be categorised from mild to profound or into different psychiatric disorders, such as conduct disorder or oppositional defiant disorder (Ogden, 2009). Behaviours often included when describing challenging behaviours are self-injurious behaviours, aggression, stereotyped behaviours, and destruction of property (McClintock, Hall, & Oliver, 2003). Persons with ID and other developmental disabilities have an increased risk of developing challenging behaviours (McIntyre, 2008). Challenging behaviours influence a person's quality of life and can lead to isolation, and little involvement in the community's social life (Lucyshyn, Horner, Dunlap, Albin, & Ben, 2002). Challenging behaviours in schools place great demands on the staff's capacities, such as their competency, motivation, and values (Roland et al., 2016). Working with challenging behaviours in schools demands special skills in the team, and there appears to be a general lack of resources in cases regarding students with challenging behaviours (Roland et al., 2016).

1.2 Autism spectrum disorder (ASD) and gender differences

ASD is referred to in the Diagnostic and Statistical Manual V (DSM V) as a dyad of impairments, including difficulties in social interactions and social communications, and restricted and repetitive behaviours, interests, and activities (American Psychiatric Association, 2013).

Lately, interest in studying ASD in genetic disorders has increased, and research has shown a higher prevalence of ASD in some genetic disorders than in the general population (Richards, Jones, Groves, Moss, & Oliver, 2015). The focus so far has been on the prevalence and phenomenology in different syndromes, and further studies are required to tell us more about the differences in ASD phenomenology between ASD in genetic syndromes and idiopathic autism.

A substantial amount of research has shown a higher rate of autistic type problems in males, compared to females. The 4:1 male to female ratio is one of the most consistent findings in ASD research (Halladay et al., 2015; Kirkovski, Enticott, & Fitzgerald, 2013; Lai, Lombardo, & Baron Cohen, 2014), and a gender difference has been part of the description of ASD since the first characterisation of the disorder.

Most of the current data suggest that the male bias is more likely to be due to female protective factors rather than male-specific risk factors, but comprehensive molecular explanations are lacking for both (Lai, Lombardo, Auyeung, Bhismadev, & Baron-Cohen, 2014; Werling, Parikshak, & Geschwind, 2016). Gender ratios in ASD differ substantially from study to study. Among individuals with ASD and normal cognitive functioning, gender differences as high as 9:1 have been reported (Brugha et al., 2011). A newer systematic review and meta-analysis from Loomes et al. (Loomes, Hull, & Mandy, 2017) found a male-to-female ratio closer to 3:1 than 4:1. According to Loomes et al. (Loomes et al., 2017), the main reasons for this change were both how ASD was diagnosed and the population that was used to investigate the male-to-female ratio in ASD in different studies. They (Loomes et al., 2017) found that studies screening the general population for ASD had a lower male-to-female ratio than studies investigating populations with pre-existing diagnoses. In cohorts with ASD in combination with intellectual disabilities, the ratio varies between 2:1 and 7:1 (Jacquemont et al., 2014; Lai, Lombardo, & Baron Cohen, 2014). Loomes et al. (2017) also found a lower male-to-female ratio in their meta-analysis in the subgroup of studies including participants with lower IQs. Epidemiological studies have described the degree of intellectual disability and the ascertainment approach as major explanations for the varying ratios that were reported (Fombonne, 2009).

Gender differences in profiles of autistic symptoms have a limited research basis (Hartley & Sikora, 2009). Several studies (Carter et al., 2007; Hartley & Sikora, 2009; Lai et al., 2011) have found that males have more restricted and repetitive behaviours than females. Some studies have found that females have more impairment in social reciprocity and communication than males, but these findings have not been consistent (Lai et al., 2011); other studies have found that females with ASD have better sociability skills than males with ASD (Head, McGillivray, & Stokes, 2014).

1.3 Smith-Magenis syndrome

Smith-Magenis syndrome (SMS) is a rare, complex genetic syndrome caused by an interstitial deletion of chromosome 17p11.2, (A. C Smith et al., 1986) or a mutation on the retinoic acid induced 1 (RAI1) gene (Slager, Newton, Vlangos, Finucane, & Elsea, 2003). The disorder is characterised by intellectual disability, multiple congenital anomalies, obesity, neurobehavioural abnormalities, and a disrupted circadian sleep-wake pattern (Williams, Zies, Mullegama, Grotewiel, & Elsea, 2012). Skeletal findings include short stature and scoliosis (Madduri et al., 2006). The incidence of SMS is estimated to be 1:25 000 births (Greenberg et al., 1991). Delayed diagnosis is common, although greater recognition of the syndrome over the last decade and achievements in genetic technology have led to earlier diagnosis (Gropman, Duncan, & Smith, 2006; Prescott, 2013).

Children and adults with SMS appear to have unique phenotypes that are especially challenging for both parents and professionals. These phenotypical characteristics or traits include: sleep disturbances, self-injurious and maladaptive behaviours, stereotypies, and sensory integration disorders (De Leersnyder et al., 2001; Gropman et al., 2006;

Madduri et al., 2006; Martin, Wolters, & Smith, 2006). Challenging behaviours are believed to increase with age and intellectual level (Neira-Fresneda & Potocki, 2015). Sleep disturbances are present in 88% of SMS patients characterised by difficulty falling asleep, frequent nocturnal awakenings, early sleep offset and daytime sleepiness with a need for daytime naps. The sleep disturbances have been associated with an unusual, inverted circadian melatonin rhythm (De Leersnyder et al., 2001; Gropman et al., 2006; Gropman, Elsea, Duncan, & Smith, 2007; A. C. Smith, Dykens, & Greenberg, 1998b). The challenging behaviours are assumed to be correlated with sleep disturbances in SMS (Neira-Fresneda & Potocki, 2015). Early expressive speech delays with and without hearing loss and mild to severe cognitive deficits have been observed (Madduri et al., 2006; Martin et al., 2006; Udwin, Webber, & Horn, 2001). Autism spectrum disorders have also been identified in almost 90% of the investigated populations with SMS (Laje et al., 2010; Martin et al., 2006). The phenotype is closely linked to the genotype in SMS, but genes other than RAI1 might account for the variable features in SMS (Girirajan et al., 2006).

The prognosis of the SMS patient is closely linked to the behavioural manifestations (Poisson et al., 2015), and research examining the relations between behavioural manifestations and other characteristics is needed.

This study seeks to bring new knowledge regarding the behaviours displayed by persons with SMS, both in their educational environments and their homes, by addressing teacher and parental perceptions/beliefs based on their experiences with these children. According to Neira-Fresneda (2015) both educational and behavioural interventions for students with SMS are extremely challenging. A good teacher-student match has been reported to be of importance (Haas-Givler & Finucane, 2014).

This PhD thesis is related to how these behavioural challenges can be managed in the pedagogical system, including how support staff relates to parents. SMS is a relative newly described disorder, and research has mostly been concerned with symptoms and genetics (Edelman et al., 2007; Elsea & Girirajan, 2008; Gropman et al., 2007). To determine how to manage these behavioural challenges, I have investigated how medical, psychiatric and other symptoms relate to behavioural challenges in schools and how both parents and school staff handle these challenges. There is a lack of guidelines regarding how to manage and treat the challenging behaviours in SMS, and these behaviours often appear in school, resulting in poor school performance (Poisson et al., 2015).

1.4 Definition of concepts

Intellectual disability (ID) is a concept used in this thesis. The medical literature in particular still uses the term "mental retardation", but in the educational field, the term "intellectual disability" is preferred. The term refers to a heterogeneous group of people who must meet three criteria for the diagnosis of ID: 1) deficit in intellectual functioning, at least two standard deviations below the mean on IQ tests, 2) deficit in adaptive functioning, and 3) the deficit in function having started during the developmental period, before turning 18 years old (Dykens, 2000). ID is often divided into four groups depending on the level of intellectual functioning: mild ID with IQ from 50 to 69, moderate ID with IQ from 35 to 49, severe ID with IQ from 20 to 34, and profound ID with IQ less than 20 (Eknes, 2001; ICD-11 International Classification of Diseases for Mortality and Morbidity Statistics, 2018).

Challenging behaviours is a concept used in this thesis. The term is used to describes all types of challenging behaviours, including but not restricted to, problem behaviours, emotional and behavioural problems, conduct disorders, oppositional defiant disorders, aggression and self-injury (Asmervik, Ogden, & Rygvold, 2004; Bru, Idsøe, & Øverland, 2016; Emerson, 2001; Holden, 2009; Weare & Nind, 2011).

Adaptive behaviour in this thesis refer to the how the concept is defined in Vineland Adaptive Behaviour Scales (VABS) (Sparrow, Balla, & Cicchetti, 2005). The authors of VABS define the concept as "the performance of daily activities for personal and social sufficiency." (Sparrow et al., 2005 p. 6). The scales are building its definition of adaptive behaviour on the American Association of Mental Deficiency's (AAMD) and the Individuals with Disabilities Educational Improvement Act's (IDEA) descriptions. These descriptions identifies three domains; conceptual (language, reading, writing and money), practical (daily living and occupational skills) and social (interpersonal, obeying laws, etc) (Sparrow et al., 2005).

Communication in this thesis refer to a broad and general definition of the ability and possibility express needs and wants and exchange information (Light & McNaughton, 2014). The concept communication is mostly used in reference to the different subscales of the questionnaires used in this thesis, in those cases, communication is defined according to the questionnaires and explained either in the description of the questionnaire or when discussed.

Person with SMS in this thesis refers to persons with SMS of all ages and is used for the most part in this thesis. In the sections where I refer to parents, the term *child* or *children* is used since parents refer to their children as children regardless of age. In the sections where I refer to school staff, the term *student* is used. This term refers to persons with

SMS of school age, from approximately 6 years old to approximately 20 years old (end of high school).

Parent refers to all types of primary caretakers such as foster parents and stepparents.

1.5 Research aims

The aim of this project is to explore challenging behaviours in a rare disorder to understand more regarding the behaviours. A broader understanding of challenging behaviours is necessary to be able to manage or change the behaviours. In this project, I search for relations between communication, challenging behaviours, and autism spectrum disorders in a Scandinavian population with Smith-Magenis syndrome. These relations could be targeted for specific interventions at home and in schools at a later point. I use data from parents and the schools to explore the challenging behaviours. The information and results will hopefully provide school staff and other professionals with a better understanding of how to adapt the environments that persons with SMS encounter and how to cooperate with these families.

1.5.1 Research questions

1. How does communication, challenging behaviours, and autism spectrum disorders vary within a Scandinavian sample of SMS? (Articles I and II)
    - Is there a relationship between the specific characteristics?
    - Are there any age or gender differences in the specific characteristics?
2. What experiences do the school staff of a person with SMS have (with tantrum, aggression, behaviour problems, teaching/class leadership, bullying, etc.)? (Articles IV and V)

  - How does the staff manage this work? (especially the challenging behaviours)
  - What does the school staff need to handle their job?
  - How does school staff cooperate with the parents of a student with SMS?
3. What experiences do the parents of a person with SMS have (especially with challenging behaviours)? (Article III)
  - How do the parents of a person with SMS view their competencies and skills considering the behavioural problems associated with this syndrome?
  - What are the parents` experiences with intervention attempts?

1.6 List of articles

This PhD thesis includes five articles. The first two investigates specific somatic and behavioural characteristics in a Scandinavian sample of SMS and explore covariation between them. Findings from these two articles were used to form the basis of the last three articles. The third article is about how the parents experienced the challenging behaviours of their children with SMS and how they perceived the work by school staff and other professionals working with their children. The last two articles concern how the school staff experienced working in the schools with a student with SMS, including cooperation with parents. Figure 1-1 illustrates how the articles relate to each other.

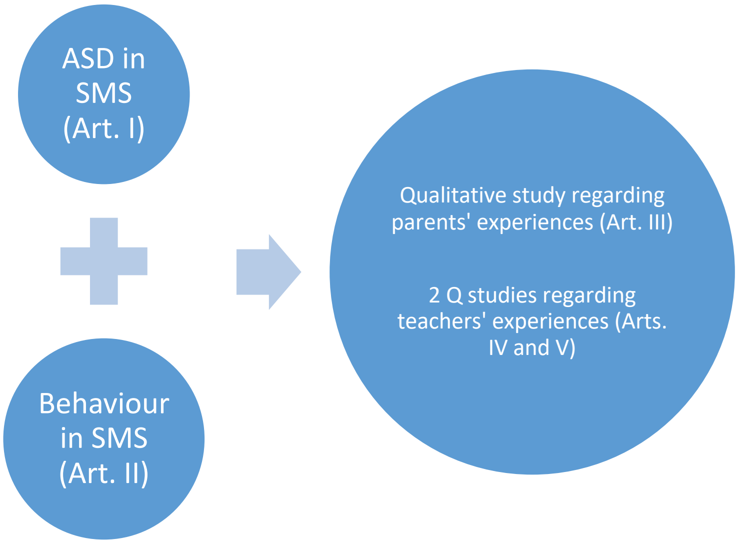

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Figure 1-1. The relations among the five articles in this PhD thesis.

1.6.1 Article I

Nag, H. E., Nordgren, A., Anderlid, B. M., & Naerland, T. (2018). Reversed gender ratio of autism spectrum disorder in Smith-Magenis syndrome. Mol Autism, 9, 1. doi:10.1186/s13229-017-0184-2.

1.6.2 Article II

Nag, H. E., & Nærland, T. (2020). Age-related changes in behavioural and emotional problems in Smith–Magenis syndrome measured with the Developmental Behavior Checklist. Journal of Intellectual Disabilities. doi:10.1177/1744629519901056

1.6.3 Article III

Nag, H. E., Hoxmark, L. B., & Nærland, T. (2019). Parental experiences with behavioural problems in Smith-Magenis syndrome: the need for syndrome-specific competence. Journal of Intellectual Disabilities.

1.6.4 Article IV

Nag, H. E., Øverland, K., & Nærland, T. (Accepted in International Journal of Disability, Development and Education). School staff's experiences and coping related to the challenging behaviours of children with Smith-Magenis Syndrome in schools: A Q methodological study.

1.6.5 Article V

Nag, H. E., Øverland, K., & Nærland, T. (In process). School Staff's Experiences with Smith-Magenis syndrome in Schools – What Do They Need and How Do They Handle the Behaviours? A Q methodological study.

1.7 Structure of the PhD thesis

This thesis has two main parts. The first part includes six chapters. Chapter 1 presents the topic of the PhD project -- challenging behaviours, autism spectrum disorders and Smith-Magenis syndrome -- and the research aims. Chapter 2 presents the contextual framework and theoretical framework. Chapter 3 offers an overview of the research methods utilised in this project. Qualitative, quantitative and Q methodologies are presented in this chapter. In this chapter, a review of ethical consideration is also presented. Chapter 4 presents the main results of the five articles in this PhD project. In Chapter 6 the results of the five articles are discussed in relation to each other. Part 2 includes the five articles included in this PhD thesis and the appendices.

2 Contextual Framework and Theoretical Framework

2.1 Contextual framework

The studies in this PhD thesis were conducted in Norway, Sweden and Denmark. The three countries have a similar educational system, including kindergartens. They all have a free public-school system (and all three have access to private schools). All three countries have individual rights for special education, although the special educational systems are slightly different among the three countries. In all three countries, students with the need for special education support have access to mainstream classrooms, support in mainstream classrooms, special education classrooms and special education schools (Education, 2018). The studies regarding the school staff in this thesis were both performed in Norway; therefore, I present the special education situation in Norway more thoroughly.

2.1.1 Special education situation in Norway

Norway has a comprehensive special educational system with individual rights (The Education Act, 1998). This act also include special education for adults (The Education Act, 1998). Adults with needs to develop or maintain basic skills have the right to such training (The Education Act, 1998 § 4A-2). The Ombudsperson for Children delivered a report in 2017 regarding the special education system in Norway, and the conclusion was that many students in special education do not receive a reasonable education like they have the right to. Lack of expectations, bad quality of the education and teachers lacking necessary competence were some of the findings of this report (Barneombudet, 2017). In 2018 came another report regarding special education in kindergartens and schools in Norway. One of the conclusions was that the staff providing special education services lack pedagogical or educational competence. Approximately half of the population receiving special education in schools receive it from an assistant, not a teacher. Children in kindergartens often receive special education from adults with a lack of formal special educational competence. At the same time, we do know that the competence of teachers is crucial for children's learning and development (Nordahl & et. al, 2018). In the fall of 2019, there is a plan for a new report to the Storting, including some suggestions for large changes in the special educational system. This report will focus on early intervention and inclusion (NOU 2019:23, 2019).

2.1.2 Laws and regulation regarding challenging behaviours in schools

The Education Act in Norway (The Education Act, 1998) states that "education shall be adapted to the abilities and aptitudes of the individual student, apprentice and training candidate" (§1-3). § 9a states, "All students attending primary and secondary schools are entitled to a good physical and psychosocial environment conducive to health, well-being and learning" (The Education Act, 1998). The right to special education is stated in § 5.1: "Students who either do not or are unable to benefit satisfactorily from ordinary teaching have the right to special education" (The Education Act, 1998). None of these sections (or any others) in the Education Act refer to challenging behaviours in schools. Students in Norway also have the right to attend the nearest or neighbourhood school (The Education Act, 1998 § 8-1), although there exist special education classrooms and special education schools. The Learning Poster in the Quality Framework of Norwegian schools also emphasises this point by stating that the schools shall "give all students and apprentices/trainees equal opportunities to develop their abilities and talents individually and in cooperation with others" and "ensure[s] that the physical and psychosocial working and learning environments promote health, well-being and learning" (Kunnskapsdepartementet, 2015 p. 2). The use of restrictive practises regarding persons with intellectual disabilities in Norway are strictly regulated (Helse- og omsorgstjenesteloven, 2011). This law does not regulate activities in schools (Utdanningsdirektoratet, 2017). Restrictive practises are only allowed in schools in self-defence, meaning to prevent danger to one's self or others and prevention of severe damage (FNs Barnekonvensjon, 1991; Straffeloven, 2005). Otherwise, the use of restrictive practises is not allowed in Norwegian schools, even though there has been decision to use restrictive practises for students outside of schools (such as respite care). There is one exception to this decision, if there is a decision about the use of restrictive practises outside the school, the same restrictive practises can be used in the school if performed by health personnel and not school personnel (Utdanningsdirektoratet, 2017). A report from 2018 indicated an increase in violence (from students towards school staff and other students) in schools (in Oslo) (Utdanningsetaten Oslo kommune, 2018), and there are especially high levels of violence by students in special educational settings (Utdanningsetaten Oslo kommune, 2018). A Danish study found in 2012 that special education teachers and child care workers are among the top 10 professions reporting being exposed to work-related threats and violence (Borg, 2012). There are no requirements to perform any behavioural assessments or develop behavioural intervention plans in Norwegian schools.

Another important element regarding challenging behaviours in schools is the school staff's duty to actively attempt to ensure that all students have safe and good psychosocial environments at school (The Education Act, 1998 § 9a-4). This requirement also includes a duty to evaluate any interventions that the schools put in place (The Education Act, 1998 § 9a-4).

2.1.3 Challenging behaviours in persons with and without intellectual disability

Somehow, the field of challenging behaviours, especially in schools, seems to be divided into two fields: one field concerning students with ID and the other concerning students without ID. The difference in the two fields likely comes from the start of the education of special education teachers at the beginning of the 1900s (Fasting, 2016). Then, the education of special education teachers was provided at specialised schools devoted to students with different disabilities. In Norway, schools for five different disabilities existed: visual impairments, hearing impairments, speech problems, intellectual disabilities, and behaviour problems (Institutt for Spesialpedagogikk). These five fields of study are still the five fields from which one can choose at the master's level at the University of Oslo, Department of Special Needs Education (Institutt for Spesialpedagogikk). The field of intellectual disabilities and the field of challenging behaviours are two distinct fields. In the field of challenging behaviours, persons with ID are almost not mentioned at all, especially not persons with moderate or profound ID (Bru et al., 2016). More interesting is that persons with ID are not even defined out of the field; they are simply not mentioned. In describing the field of ID, challenging behaviours are almost never mentioned (Asmervik et al., 2004; Befring & Tangen, 2008; Rygvold & Ogden, 2008); it seems as though challenging behaviours in persons with ID constitute a subfield within the field of ID (Emerson, 2001; Holden, 2009; Oliver, Berg, Moss, Arron, & Burbidge, 2011). It is not necessarily easy to determine whether persons with ID are defined in or outside of books or articles (Asmervik et al., 2004; Befring & Tangen, 2008; Roland et al., 2016; Rygvold & Ogden, 2008; Ttofi & Farrington, 2011; Weare & Nind, 2011). Behavioural and emotional disorders are among the most common psychiatric disorders in children (Mørch, 2003). Research has also found that persons with ID have a higher prevalence of psychiatric disorders than the general population (Einfeld, Ellis, & Emerson, 2011).

Nevertheless, challenging behaviours in persons with ID will often be regarded as a part of the disorder and not as an additional/comorbid psychiatric disorder (Eknes, 2001).

2.2 Theoretical framework

Behaviours challenge people in all different arenas in various ways. The behaviours are also influenced by how different people act and react (Tetzcner, 2003). How school staff think about the cause or function of the behaviours influences its own emotional understanding and how it acts and reacts (Grieve, 2009). Exploring challenging behaviours is the main topic of this thesis, including exploring both parents' and school staff's experiences with these behaviours. In the following section, Bronfenbrenner's bioecological theory (Bronfenbrenner, 1977, 1979, 1995) and Sameroff's transactional model of development (Sameroff, 2009) are presented since they concerns how development and environment influence each other. Then, Lazarus' model (Lazarus, 1999; Lazarus & Folkman, 1984) of stress and coping is presented since it can enlighten how the school staff, parents and persons with SMS manage the challenging behaviours that occur most often with SMS. Finally, a theory regarding proactive and reactive aggression is presented (Card & Little, 2006).

2.2.1 Developmental psychology

Knowledge about regular developmental principles is important when studying development that diverts from regular development (Tetzchner, 2012): "An ideal, complete, formal scientific theory is a set of interconnected statements -- definitions, axioms, postulates, hypothetical constructs, intervening variables, laws, hypothesis and so on." (Miller, 1993 p. 3). In psychology, few theories become formal theories, and none in developmental psychology have so far (Miller, 1993). The critical part of developmental theory is the focus on change over time

(Miller, 1993). Developmental psychology revolves around how individuals change socially, mentally and behaviourally, the processes underlying the change and the conditions that have implications for the changes (Tetzchner, 2012). A developmental theory helps us to organise information and gives meaning to the facts of development (Miller, 1993).The tasks of developmental theories are: 1) to describe the changes in behaviours over time; 2) to describe the changes in relations among different behaviours; and 3) to explain the development that has been described (Miller, 1993). There are many different theories, but two common complimentary perspectives are usually part of them all. Most developmental theories seek the conditions that lead to the characteristics that are common to all human beings. The second perspective is on the conditions that make human beings unique individuals (Tetzchner, 2012). Some characteristics are common for almost all human beings, such as talking and walking. Some characteristics are more differentiated between human beings, such as temperament and intelligence (Tetzchner, 2012). Bioecological theory and the transactional theory of development are further explored in this thesis, with an emphasis on Bronfenbrenner's bioecological theory. Developmental theories are usually constructed by relating theory and data in four basic ways: models, deductive theories, functional theories and inductive theories (Miller, 1993). Few developmental theorists have emphasised how children can influence their environments; they have mostly been concerned with how the environment influences development (Miller, 1993). Bronfenbrenner included both sociohistorical changes and the physical and social environment in his theory (Bronfenbrenner, 1977). Sameroff's transactional model of development incorporated how nature and nurture interact in development (Sameroff, 2010).

2.2.2 Bronfenbrenner's bioecological theory

Bronfenbrenner proposed a model of how children's development is influenced by different environments (from the micro to macro levels) with which the individual comes in contact, either directly or indirectly (Bronfenbrenner, 1977). The role of the theory is to create a connection and communication between the different levels/layers in the model and to develop an understanding across levels. Although the theory's reciprocal cause and effect explanations make it difficult to identify dependent and independents variables, Bronfenbrenner's bioecological theory catches the complexity of cooperation and provides a hieratical understanding of development (Klefbeck & Ogden, 1995). It considers that, through everyday activities and interactions, the individual is mutually influenced by his or her own characteristics and the diverse contexts (environments) that they encounter (Rose & Tudge, 2016). Bronfenbrenner described (in one of his first models) four different layers or environmental structures that influence the development of the individual. The model consists of concentric circles surrounding the individual (see Figure 2-1). The first circle is the microsystem, which is the individual's primary environment and the arenas in which the individual participates, such as the family, kindergarten and school. Later, Bronfenbrenner also included the importance of significant others in the microsystem, with their distinctive characteristics (Bronfenbrenner, 2005). The microsystem consists of three elements: activities, roles and relations (Bronfenbrenner, 1979). The second circle is the mesosystem. This circle represents the relations and communication between the different settings in the microsystem. This circle includes communication and cooperation between home and school/kindergarten, home and respite care and school/kindergarten and respite care among others. A crucial part of the mesosystem consists of ecological transitions, such as the transition from home to kindergarten, from kindergarten to school, and so on. The third circle is the exosystem. This circle concerns is the social institutions in the society. Social institutions could be pedagogical centres, habilitation centres, the county and so on. The fourth and last circle is the macrosystem. This circle includes the society, culture and value system (Bronfenbrenner, 1977). Laws and regulations are part of the macrosystem. The model was elaborated to a model of gene-environment interactions in human development, to put a greater emphasis on the individual's development in a social context (Bronfenbrenner & Ceci, 1994). The model considers the variance of reality and the connections that exist from the micro to macro levels (Klefbeck & Ogden, 1995). Bronfenbrenner also added a fifth circle: the chronosystem. The chronosystem includes experiences in life, both environmental influences and transitions (Bronfenbrenner, 1995; Bronfenbrenner & Ceci, 1994).

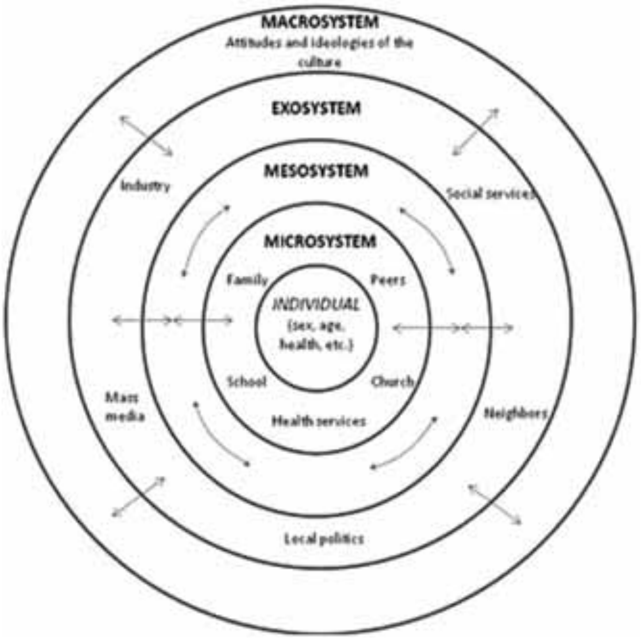


Figure 2-1. The ecological theory of human development. This figure illustrates the second revision to ecological theory (Bronfenbrenner, 1977).

Bronfenbrenner continuously developed his theory until he died in 2005 (Tudge, Mokrova, Hatfield, & Karnik, 2009). One of the developments in the theory included the PPCT concepts. PPTC stands for Process, Person, Context and Time (Tudge et al., 2009). *Process* refers to the interaction between the individual and the environment over time

(Bronfenbrenner & Morris, 1998). *Person* refers to both the genetic and biological aspects of the individual, as well as the personal characteristics that a person brings to the social situation. At a later point of developing his theory, Bronfenbrenner also emphasised the individual's role in changing his or her context, both in a passive and a more active way (Bronfenbrenner & Morris, 1998). The *context* involves the four (five) systems described earlier (Bronfenbrenner & Morris, 1998). *Time* is also a crucial concept in Bronfenbrenner's theory. An individual's development occurs over time, and the contexts also change over time (Bronfenbrenner & Morris, 1998). For a visual presentation of the PPCT model, see Figure 2-2.

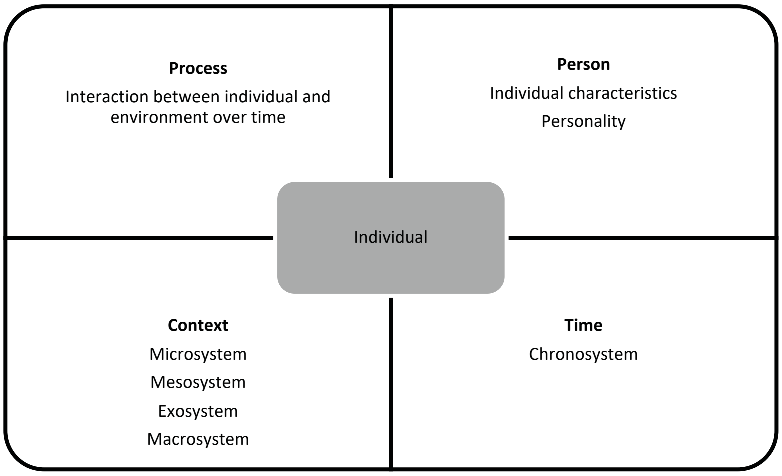


Figure 2-2. The bioecological theory of human development. The PPTC Model.

2.2.2.1 Social network

One crucial part of bioecological theory consists of social networks (Johannessen, Kokkersvold, & Vedeler, 2010). In the process of developing from a child through youth to adulthood, socialisation is important. Social networks plays an important role in how one handles the transitions (Johannessen et al., 2010). Social networks can be divided into formal and informal networks (Klefbeck & Ogden, 1995). The informal networks, also called primary networks, consist of people close to a person, such as family, friends, colleagues. The formal, secondary network consists of professionals (Klefbeck & Ogden, 1995). For persons with disabilities, such as SMS, the formal network can play a more crucial role both earlier and to a greater extent than for others.

All of the elements of Bronfenbrenner's theory are important to acknowledge, but in a situation in which an individual has a disability, it is especially important to include the individual's characteristics. A person with SMS has some specific characteristics, such as delayed language acquisition, challenging and self-injury behaviours and sleep issues, which influence both the development and the environment. How these characteristics and challenges are met and handled in the micro, meso, exo and macrosystems influences the experiences of the person with SMS and his or her family.

In Norway, we have an act related to municipal health and care services (Helse- og omsorgstjenesteloven, 2011). The purpose of this law includes objectives to prevent, treat and facilitate coping with the disability (§ 1-1) and to ensure coordination and that the services offered are adapted to the individual's needs (§ 1-1, 5) (Helse- og omsorgstjenesteloven, 2011). Families in which one person has SMS are often in contact with many different care takers and institutions, such as respite care, personal assistants, hospitals, behaviour teams, habilitation centres, other resource centres, etc. The need for cooperation and coordination of all of these institutions is important to both the family and the individual, as well as everyone involved, including kindergarten and school staff. According to Bronfenbrenner there must be communication between levels. He claimed that micro and macro must become meso. A model of how Bronfenbrenner's bioecological model would resemble for a person with SMS is presented in Figure 2-3.

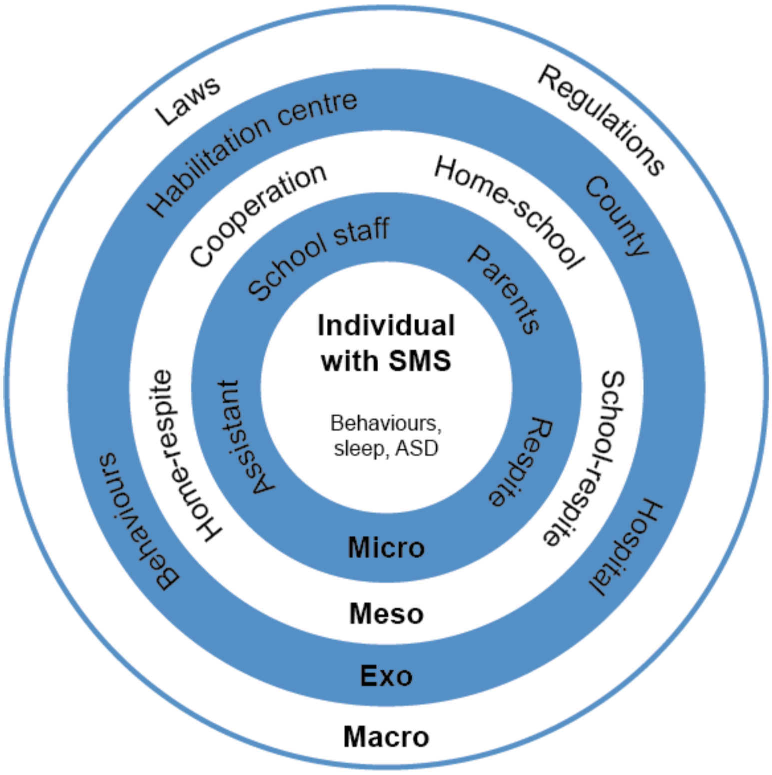


Figure 2-3. How the ecological model could appear for a person with SMS.

2.2.3 The transactional model for development

Sameroff's model of development came as a response to the historic changing of view between nature and nurture. One of his models includes transactions among phenotype, environtype, and genotype (Sameroff, 2009). Sameroff´s transactional model for development is a bidirectional model that includes both processes in the individual and processes in the individual's context over time (Sameroff, 2009). Both the environment and the individual change over time and as a response to the experiences from interactions (Sameroff, 2009). Sameroff's transactional model for development can be used to describe the relationship of parents and children, and school staff and students. It can also describe the development of behavioural challenges in schools (Sameroff, 2009). The students' behaviours influence the school staff's reactions, again influencing the students' behaviours in return (Sameroff, 2009).

In the transactional model, the development of a child is a product of the dynamic interactions of the child and his or her social settings. The model emphasises the bidirectional, interdependent effects of both the child and the environment (Sameroff, 2009).
The background of the development of the model came from the questions of why some children with medical anomalies grew up and did not have the expected difficulties in cognitive and emotional abilities and how at-risk parents could bring up well-functioning children (Sameroff, 2009). One of the first models of how the child and the environment (such as parents) have bidirectional influences on each other are presented below (Figure 2-4).

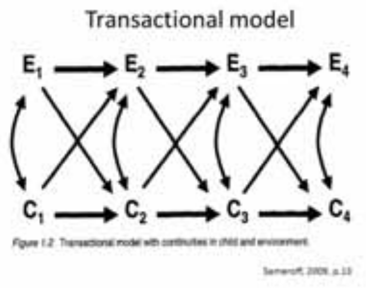


Figure 2-4: Transactional model (Sameroff, 2009).

Among the key concepts in the model are time and that both the child and environment change over time. The child influences the environment, and then the changed environment influences the child (Sameroff, 2009).

Another model presented by Sameroff in 2009 (Sameroff, 2009), included both the concept of *environtype,* which consists of all of the subsystems that transact with the child. Bronfenbrenner (1979) developed a thorough description of all of the subsystems that interact with children, including *phenotype,* which is the child itself, and *genotype*, which is the source of the biological organisation. A visual presentation of the model is presented below (Figure 2-5).

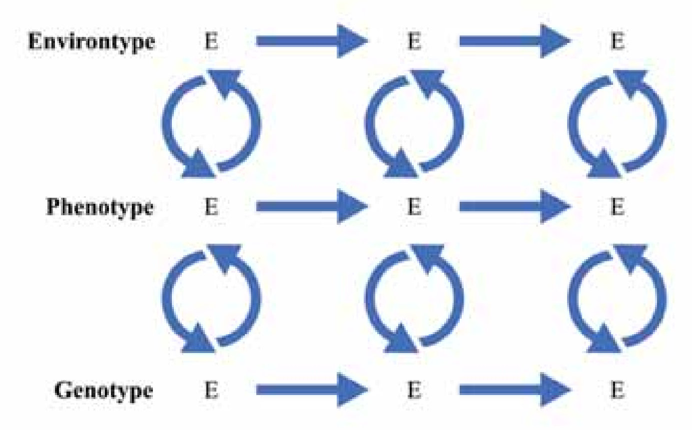


Figure 2-5: Environtype, phenotype and genotype. Interaction in an individual's development

The core symptoms of SMS seem to be linked to the lack of two functioning copies (haploinsufficiency) of the RAI1 gene (Poisson et al., 2015). This *genotype* of persons with SMS is responsible for the core symptoms or traits of SMS (Poisson et al., 2015), which are referred to as the *phenotype* (Girirajan et al., 2006). The genotype influences the environment and all subsystems with which the child directly or indirectly interacts, which is the *environtype* (Sameroff, 2009). A person's behaviours can be a product of the genotype, the phenotype and the environtype (Sameroff, 2009).

2.2.4 Stress and coping

Challenging behaviours in schools place great demands on the staff's capacities, such as their competency, motivation, and values (Roland et al., 2016). Working with challenging behaviours in schools demands special skills on the team, and it seems as if there is a general lack of resources in cases regarding students with challenging behaviours (Roland et al., 2016). Skåland (2016) found in his PhD dissertation that lack of information regarding the diagnosis and how to handle challenging behaviours changed how the school staff handled the situation. Working with students with challenging behaviours could impact both stress and coping for the school staff. Regarding caregivers of persons with SMS and stress, research has indicated that there is a relationship between the amount of challenging behaviours and the level of stress (Fidler, Hodapp, & Dykens, 2000; Hodapp, Fidler, & Smith, 1998). How school staff working with students with SMS in schools manage and handle challenging behaviours is one of the main topics in this project. How parents experience challenging behaviours is another. The results of both topics are analysed utilising Lazarus' transactional model for stress and coping (Lazarus & Folkman, 1984).

2.2.4.1 Transactional model for stress and coping

Stress is an expected part of life, but what is different is how one copes with the stress (Lazarus & Folkman, 1984). Both coping and stress can be viewed as relationships between a person and the environment (Folkman, Lazarus, Gruen, & DeLongis, 1986). Coping refers to a person's efforts to manage the demands from the transaction between person and environment. How stressful the person-environment relationship will become is based on the appraisal of the situation (Lazarus & Folkman, 1984). Coping is defined as "constantly changing cognitive and behavioural efforts to manage specific external and/or internal demands that are appraised as taxing or exceeding the resources of the person" (Lazarus & Folkman, 1984 p. 141). Stress emphasises the relationship between the person and the environment appraised by the person as either taxing or exceeding his or her resources and threatening his or her well-being (Lazarus & Folkman, 1984).

Lazarus emphasised the *appraisal* process in regard to coping and emotions (Lazarus, 1991). *Appraising* has been defined "as a set of cognitive actions, a process performed by an individual who may or may not have been conscious of doing it." (Lazarus, 1999 p. 75). He split appraisal into *primary appraisal* and *secondary appraisal.* The terms primary and secondary do not refer to an order of experiences. In *primary appraisal,* one evaluates whether what happens affect one, either in a positive or negative way and now or in the future? Primary appraisal can be divided into 1) irrelevant, 2) benign-positive, or 3) stressful. The first category, irrelevant, does not have any implications for a person's well-being. The benign-positive category occurs if the encounter is interpreted as positive and characterised by pleasurable emotions, such as joy and happiness. The third category, stressful, can include harm/loss, threat and challenge. These three types of stress appraisals can occur at the same time and are not necessarily mutually exclusive. Harm/loss often occurs after an event but can also include the threat of future negative implications. Threat can be distinguished from harm/loss by its possibilities for coping. Challenge and threat also have things in common, but the emotions by which they are characterised differ. Threat is often exemplified with emotions such as fear, anxiety and anger. Challenge is often characterised by more pleasurable emotions, such as eagerness and excitement (Lazarus & Folkman, 1984).

Secondary appraisal is an evaluation of what, if anything, can be done with a situation (Lazarus & Folkman, 1984). Secondary appraisal is very important in stressful situations. Secondary appraisal is a complex evaluation process consisting of two expectancies (Lazarus & Folkman, 1984). Lazarus and Folkman based their secondary appraisal process on

Bandura (1977), who referred to expectancies as *outcome expectancies* and *efficacy expectations. Outcome expectations* are the evaluation of the behaviours that lead to the outcome that one wants. *Efficacy expectations* are a person's conviction of the possibility of executing the behaviours required to produce outcomes (Bandura, 1977). In addition, the process includes secondary appraisal and evaluation of the consequences to use a particular strategy vis-à-vis other demands in the specific situation (Lazarus & Folkman, 1984). Primary appraisal and secondary appraisal interact with each other in shaping the degree of stress and the strength and quality of the emotional reactions. The way in which a person appraises an encounter influences the coping strategy and the person's emotional reaction (Lazarus & Folkman, 1984).

Coping can be viewed as having two forms: 1) dealing with the problem and 2) regulating the emotions (Folkman et al., 1986; Lazarus, 1999). In *problem-focused coping* (dealing with the problem), one attempts to change the situation by seeking knowledge and information to use when choosing solutions (Drageset, 2014; Lazarus & Folkman, 1984). In *emotion-focused coping* (regulating the emotions), one often experiences a situation as unchangeable and attempts to manage the emotional stress in the situation more than change the situation (Drageset, 2014; Lazarus & Folkman, 1984).

This coping theory is relevant in relation to working with a person with SMS or having a child with SMS. How well professionals and parents manage and handle challenging behaviours depends on the appraisal process (Lazarus & Folkman, 1984). In particular, where they in the primary appraisal perceive the encounters as a challenge or a threat could be important. The availability of resources and knowledge can play an important role in the secondary appraisal process(Lazarus & Folkman, 1984). Stress can also be an issue for persons with SMS themselves. Anxiety has been described in this disorder (Gropman et al., 2006). High levels of anxiety have been associated with stress in persons with autism

(Gillott & Standen, 2007). Anticipation and change were especially correlated with high levels of anxiety (Gillott & Standen, 2007).

One of the main factors influencing psychological stress, according to Lazarus (1984), is *beliefs*. *Beliefs* shape the understanding of meaning and determines what is factual. Two sets of beliefs are relevant to appraisal: beliefs that have to do with *personal control* and beliefs that have to do with *existential concerns* (Lazarus & Folkman, 1984). Beliefs of *personal control* concern feelings of mastery and confidence, both in general ways of thinking and in situation-specific expectations (Lazarus & Folkman, 1984). Beliefs related to *existential concerns* are those that help people create meaning out of life, such as believing in God, fate or some other order in the universe (Lazarus & Folkman, 1984). It is clear that beliefs about control play a major role in determining the degree to which a person feels threatened or challenged in a stressful encounter, and this belief will colour the appraisal.

There are also situational factors that influence appraisal. Lazarus emphasised six such factors: novelty, predictability, event uncertainty. temporal factors, ambiguity and timing of stressful events in relation to the life cycle (Lazarus & Folkman, 1984). These factors create potentials for threat, harm or challenge. All of these situational and personal factors are interdependent, and their significance for stress and coping derives from the operations of cognitive processes that give weight to one in the context of the other (Lazarus & Folkman, 1984).

The feelings of mastery and confidence of school staff and the parents of children with SMS can be influenced by the level of knowledge that they have, regarding both the disorder itself and how to manage challenging behaviours. The experience of support, either from the community for the parents or from colleagues and leaders for the school staff, can also influence these people's beliefs about control and their appraisals of situations. For persons with SMS, both mastery and confidence can play crucial roles. Personal control might be the most important part for persons with SMS due to their need to know what is happening and being prepared.

2.2.5 Proactive and reactive aggression

Historically, especially in the field of challenging behaviours in persons without ID, aggression has been differentiated between the lines of proactive and reactive aggression (Card & Little, 2006; Dodge & Coie, 1987). *Proactive aggression* refers to deliberate acts directed towards obtaining desired goals, and there is a link between aggressive behaviours and desired outcomes (Card & Little, 2006; Farmer & Aman, 2009). *Reactive aggression* refers to angry responses to perceived offenses or frustrations, often emotionally dysregulated. Reactive aggression is not focused on personal gain (Card & Little, 2006; Farmer & Aman, 2009). These two types of aggression were first described in the 1960s (Kempes, Matthys, de Vries, & van Engeland, 2005). The descriptions arose from two different theories: frustration-aggression (reactive aggression) and social learning theory (proactive aggression) (Kempes et al., 2005). In the beginning, these theories were considered opposite and competing theories of aggression, but later, it was realised that the theories referred to different aspects of aggression (Kempes et al., 2005). These constructs of aggressive behaviours have not been thoroughly researched in the field of persons with ID (Farmer & Aman, 2009). How these two descriptions of aggression relate to the challenging behaviours in SMS have, to my knowledge, not been researched in any extensive manner. Whether challenging behaviours are perceived either as proactive or reactive could have an impact on how the school staff manages these behaviours. One of the subscales on the DBC could be used to assess aggressive behaviours (Farmer & Aman, 2009).

3 Previous Research

3.1 Research regarding challenging behaviours in schools and kindergarten

Classroom disruption and students' challenging behaviours have become among the largest issues in classrooms today (Alter, Walker, & Landers, 2013; Bru, 2009). In earlier international investigations (PISA 2001, 2004), Norway often came out on the top of lists regarding challenging behaviours in school. This situation has changed in a positive direction over the last two PISA investigations, and Norway has had results similar to those of the other Nordic countries and OECD countries regarding challenging behaviours in school (PISA 2012, 2015). In a report regarding violence in the schools in Oslo, there was noted an increase in violence towards both school staff and other students (Utdanningsetaten Oslo kommune, 2018). Norway also came out on the top of a list of how many resources are spent on schools (Eriksen, 2008). Some studies have shown that teachers perceive what are defined as mildly challenging behaviours, such as off-task behaviours and verbal disruptions, as the most frequent and problematic in schools (Alter et al., 2013; Rosenberg & Jackman, 2003). Challenging behaviours in schools place great demands on the staff's capacities, such as their competency, motivation, and values (Roland et al., 2016). Working with challenging behaviours in schools demands special skills in the team, and according to research, there is a general lack of resources in cases regarding students with challenging behaviours (Kelly, Carey, McCarthy, & Coyle, 2007; Lindsay, Proulx, Thomson, & Scott, 2013; Roland et al., 2016).

In a qualitative study of teachers teaching students with ADHD, one of the major accusations was lack of support from the school leadership (Harazni & Alkaissi, 2016). In general education, one of the suggestions for preventing challenging behaviours in school is to provide explicit and engaging academic instruction (Alter et al., 2013).

3.2 Research regarding SMS

Research regarding SMS seems to be scarce. A broad search in MEDLINE and PubMed using "Smith-Magenis syndrome" as a search string returned fewer than 400 hits. This search also included articles only mentioning SMS and not only research regarding SMS. Most of the research thus far has concentrated on describing the syndrome and its symptoms (Gropman et al., 2007; Martin et al., 2006; Poisson et al., 2015). Fewer studies have been concerned with the relations between symptoms (Poisson et al., 2015). In 2004, Haas-Givler (2004) performed a thorough search for articles concerning the educational needs of children or adults with SMS but did not find any. Two studies used the school as a place of observation to observe the nature of social preferences and interactions in SMS (Wilde, Silva, & Oliver, 2013), as well as the gene-environment interaction (Taylor & Oliver, 2008). None of these research studies focused on the educational needs or the school staff per se.

Future research is needed to identify both genetic and environmental modifiers regarding the broad phenotypic spectrum of SMS (Chen, Mullegama, Alaimo, & Elsea, 2015). Additionally, a lack of consistent data to assess the usefulness of interventions regarding behavioural problems in SMS was documented (Poisson et al., 2015). The research performed in this thesis is therefore a step to fill this gap in the research regarding SMS.

Variable levels of cognitive impairment have been documented through a variety of psychometric tests that assess adaptive behaviours, intelligence quotient (IQ), and speech and language development (Madduri et al., 2006; Martin et al., 2006; Udwin et al., 2001). Administering formal psychometric assessments might be difficult with persons with SMS due to maladaptive behaviours, sleep disturbances, and difficulties in language skills (especially expressive language)(M. R. Smith, Hildenbrand, & Smith, 2009). Both behavioural problems and the need for assistance in many situations last into adulthood (Udwin et al., 2001).

This disorder has a great impact on families' lives, especially aspects such as challenging behaviours, communication problems, autism spectrum disorder, and sleep problems. Hodapp et al. (1998) found that parents of children with SMS reported greater pessimism and parent and family problems than families coping with Down syndrome. Another study of caregivers for children with SMS showed that the caregivers have increased amounts of distress in the form of depression, anxiety, and sleep problems (Foster, Kozachek, Stern, & Elsea, 2010). There have also been findings that support that the level of stress among caregivers is related to the level of challenging behaviours that the child with SMS displays (Fidler et al., 2000; Hodapp et al., 1998).

There has been a lack of research regarding SMS and behavioural challenges and educational needs in learning institutions (Haas-Givler & Finucane, 2014). Only a couple of studies have been conducted in the school setting (Taylor & Oliver, 2008; Wilde et al., 2013). Behavioural challenges in learning institutions have been thoroughly researched, but not much of the research has been related to persons with intellectual disabilities and behavioural challenges (Tetzcner, 2003). Exploring challenging behaviours in SMS both inside and outside of the school setting is important to learn more about SMS and how to handle behavioural challenges of persons with SMS.

4 Methodology

To answer the research questions exploring challenging behaviours in a rare disorder, a combination of approaches characterised as multimethod design was used. According to Esteves and Pastor (2004), when a study comprises two or more rigorously and complete methods in one project, it could be described as a multimethod design. This PhD thesis includes quantitative, qualitative and Q methodologies. Articles I and II use quantitative methods, Article III uses qualitative methods, and Articles IV and V use Q methodology. The main aim of this study was to explore and describe the challenging behaviour of persons with SMS. A Multimethod design was therefore chosen as it may be beneficial in explorative research regarding subjects and themes where there have not been done a lot of research before (Fetters, Curry, & Creswell, 2013; Schoonenboom & Johnson, 2017). In this chapter, I first present the measurements and data analysis regarding the three different methods (quantitative, qualitative and Q methods). Then, the study population, trustworthiness and ethical aspects regarding all three methods are presented.

4.1 Quantitative methods

Articles I and II utilised quantitative methods, mainly standardised questionnaires. The gold standard for diagnosing ASD is either the Autism Diagnostic Observation Schedule (ADOS) or the Autism Diagnostic Interview-Revised (ADI-R) (de Bildt et al., 2004). Both these measures require interviews or direct observation. The goal in this thesis was not to diagnose ASD, but to identify autism spectrum symptomatology. Therefore, due to both time and travel restrictions, the SRS and the SCQ were used, even though they have less sensitivity and specificity than the ADOS and the ADI-R (Oosterling et al., 2010). Both SRS and SCQ are commonly used in research regarding ASD symptomatology (Moody et al., 2017). I have used informant report for all measures, with caregivers completing the questionnaires. Even though some of the persons with SMS do not have ID, based on the judgement of the caregivers and myself, none of the persons with SMS would be able to self-report on standardised questionnaires.

4.1.1 Measurements

The *Vineland Adaptive Behavior Scale II (VABS II)* (Sparrow, Balla, & Cicchetti, 1984; Sparrow et al., 2005) is a semi-structured interview or rating form for parents or caregivers to assess the everyday behavioural functioning of children and adults from birth throughout life. In this study, it is the parent/caregiver rating form that was used. The scales yield standard scores (mean =100, one standard deviation (SD) =15) in the domains of communication, daily living skills, socialisation and motor function, as well as a total sum score on adaptive behaviours. Each domain contains several subdomains, the subdomains yield v-scale scores (mean =15, one standard deviation (SD) = 3). Motor function can only be assessed in children younger than six years of age. In this study, the Norwegian, Swedish and Danish versions of the scales based on Scandinavian normative data was used. The VABS II is a standardised and validated tool. Many studies have confirmed its reliability and validity, making it one of the most widely used tools for assessing adaptive behaviours (Sparrow & Cicchetti, 1985). Regarding the reliability coefficients in the internal-consistency reliability studies more than half of the items were 0.90 or greater, less than six were below 0.80 (Sparrow et al., 2005). Studies have found a strong evidence for applicability of VABS in the population with ID (de Bildt, Kraijer, Sytema, & Minderaa, 2005).This tool has also been used for SMS several times (Madduri et al., 2006; Martin et al., 2006).

The Developmental Behaviour Checklist (DBC) (Einfeld & Tonge, 1992, 2002) is a questionnaire completed by parents or other primary carers or teachers, reporting problems over a six month period. It is an instrument for assessment of a broad range of behavioural and emotional problems of persons with developmental and intellectual disabilities (Dekker, Nunn, & Koot, 2002; Einfeld & Tonge, 1995). The Norwegian version of the questionnaire have recently been evaluated (Halvorsen & Helverschou, 2020). The DBC have a confirmed reliability and validity (Dekker et al., 2002; Einfeld & Tonge, 1995). Both the inter-rater and test-retest reliabilities had high levels of agreement (0.80 and 0.83) (Einfeld & Tonge, 2002). When assessing the content validity, experienced psychiatrist reviewed the answers provided by an informant. The psychiatrist reviewed the answers based on a psychiatric interview. Only three out of 96 items were scores differently by the psychiatrist and the informant (Einfeld & Tonge, 2002). The DBC has been proved useful in research of prevalence of psychopathology in the population of persons with ID (Einfeld & Tonge, 1995). Each behavioural description is scored with a 0, 1, or 2 rating where 0 = “not true as far as you know”, 1 = “somewhat or sometimes true”, and 2 = “very true or often true”. Five versions of the checklist are available: the Parent/Carer version (DBC-P), Teacher version (DBC-T), Adult version (DBC-A), Short-form (DBC-P24), and Monitoring chart (DBC-M). In this study, the DBC-P was used. The DBC-P consist of five subscales and a Total Problem score. The subscales are disruptive/antisocial, self-absorbed, communication disturbance, anxiety and social relating. Both the Total Problem score and the subscales are converted to percentiles (Einfeld & Tonge, 2002). The Total Problem score also have a clinical cut off of 46 (Einfeld & Tonge, 2002).

The *Social Responsiveness Scale (SRS)* is a 65-item, quantitative, parent-reported or adult self-reported measure that assesses social impairment associated with autism spectrum disorders (Constantino JN & CP, 2005). The SRS requires 15 to 20 minutes to complete. The instrument inquiries about the specific and observable elements of reciprocal social behaviours (39 items), social use of language (6 items), and behaviours characteristic of children with autism and other PDDs (20 items), and it generates a singular scale score in addition to five subscale scores (Constantino & Gruber, 2005). The five subscales are; social awareness, social cognition, social communication, social motivation and autistic mannerisms (Constantino & Gruber, 2005). Scores on both the SRS Total and all subscales are converted to T-scores. On the total score, a T-score between 60 and 75 are considered to be mild to moderate, and a T-score of 76 or higher are considered to be in the severe range (Constantino & Gruber, 2005). Studies have found this instrument valid and reliable for measuring autistic traits (Bölte, Poustka, & Constantino, 2008; Constantino et al., 2003). The Cronbach's alpha on the Total Raw score are all in the acceptable range (<0.90) (Constantino & Gruber, 2005). The interrater agreement were also in the acceptable range (0.75 - 0.92) (Constantino & Gruber, 2005). The SRS has also deemed feasible for research studies of autism spectrum conditions (Constantino et al., 2003).

The *Social Communication Questionnaire (SCQ*) is a standardised screening tool for the evaluation of communication forms and social function in children or adults to exclude autism or an autism spectrum disorder (ASD) (Rutter, Bailey, & Lord, 2003). The questionnaire is used from the age of four. It contains 40 items, which are answered with "Yes" (=1) or "No" (=0), and it comes in two versions. The SCQ-Current covers the individual's behaviours during the most recent three months, while the SCQ-Lifetime is based on the individual's entire developmental history. Both versions yield a single total score, with a score of 15 or greater regarded as an indicator of possible ASD (cutoff). A review of different assessment of rating scales for ASD found the SCQ performing well on psychometric properties, and is useful as a screening instrument (Norris & Lecavalier, 2010). SCQ and the Autism Diagnostic Interview (ADI) are significantly correlated on both total (r .71) and the subscales (r between .31 - .67) (Rutter et al., 2003). The Cronbach's alpha within different age groups ranged from 0.84 – 0.93, and across diagnostics groups 0.81 – 0.92 (Rutter et al., 2003). In this study, the SCQ-Lifetime questionnaire was used (Schjølberg & Tambs, 2005). The tool is used in research about different genetic disorders (Hall, Lightbody, Hirt, & al., 2010; Magyar, Pandolfi, & Dill, 2012) and has been used in studies in which persons with SMS were part of the study (Laje et al., 2010).

Study-specific questionnaire:
I designed a form to retrieve specific diagnostic information from caregivers. This form includes information about the use of any medications (now and earlier), other medical complications, and sensory losses (especially sight and hearing problems).

4.1.2 Data analysis

The quantitative data were compiled for statistical analysis using the Statistical Package for the Social Sciences (SPSS), version 25 and 26 (IBM). Due to the varying distribution of data in the variables in question both parametric and non-parametric tests were used in this thesis. Shapiro-Wilk analyses was performed on DBC across age groups, VABS, SRS, SCQ and ID. Only ID had a significant deviation from normality ($p < 0.001$). I therefor used the non-parametric Mann-Whitney independent sample test when investigating group differences in ID (Field, 2014). In Article I descriptive statistics were derived, and the differences between male and female on the total scores and subscores obtained from the SRS and the SCQ were analysed using the independent samples t-test (Field, 2014). The ratio was calculated as the number of females above the cutoff on the SCQ total score divided by the number of males above the cutoff. Effect sizes (Cohen's d) were calculated using Social Science Statistics' online resources (Social Science Statistics, 2017). Fisher's two-sided exact test (Field, 2014) was used to test the proportions of males and females above the SCQ cutoff and in the different SRS classifications. Multiple regression analyses were conducted to assess the impact of “gender”, “DBC” and “VABS II” standard scores’ on the “total SCQ score” and on the “SRS raw score”. Due to a relatively small sample, we were only powered to detect medium to high effect sizes. The results need to be interpreted with caution since we have high number of covariates and comparisons in the analysis of this modest sample. The normality of the residuals was checked using the visual inspection of P plots (Field, 2014). Due to the combination of dichotomous and continuous predictor variables, I report the standardised coefficients (β), in addition to unstandardised B.

In Article II, both total scores and subscores were obtained from the DBC, VABS and SCQ.
Pearson’s r was calculated using bivariate correlation analysis (Field, 2014). The participants were divided into three age groups (< 9 years, 9-18 years and > 19 years) to investigate whether the behavioural and emotional problems measured with DBC change with age. The t-test was performed to investigate the relationships of DBC (including subscales) with gender, VABS and SCQ results. Multiple regression analyses were conducted to assess the impacts of ID grade, gender, age, VABS (including the communication subscale), SRS and SCQ on DBC Total. Our analysis was powered to detect medium to high effect sizes. The results need to be interpreted with caution.

The p-value used in this thesis is 0.5. Due to modest sample sizes, the p-levels are not corrected for multiple tests, but effect sizes are also reported for improved interpretation of the comparisons conducted. Also due to this restriction, conclusions are drawn with great care.

4.2 Qualitative methods

In Article III, I utilise a phenomenological approach to investigate the parents' lived experiences (Creswell, 2013). Phenomenology is an approach especially suitable to investigate peoples' lived experiences (Creswell, 2013). Phenomenology may be viewed as both a philosophical approach, a methodology and a method (Creswell, 2013; Moran, 2000). Edmund Husserl is considered as the fountainhead of phenomenology (Groenewald, 2004). Phenomenology is concerned with the experience of the individual, and based on a paradigm of personal knowledge and subjectivity (Lester, 1999). Phenomenology as a research method was chosen to investigate the parents' experiences living with a child with SMS, because they are close to the children. Their personal knowledge may add important information about how to help these children. I decided to use written responses to open-ended questions and in-depth interviews to supplement the written responses and grasp the parents' subjectivity. In phenomenological research, significant statements are emphasised, and these statements are divided into themes organised based on the description of the parents' experience of the phenomenon (Creswell, 2013). After descriptions and themes have been obtained, the researcher can revisit the participants again to clarify or validate the data (Creswell, 2013). An important step in phenomenological studies is to set aside one's own personal experience by writing it down and bracketing it (Creswell, 2013). Bracketing means that the researcher identifies personal experiences (beliefs, feelings and perceptions) with the phenomenon and sets it aside to be able to focus on the experiences of the participants (Creswell, 2013). Edmund Husserl, the founder of phenomenology, called this practise *epoché* and developed it as a method to be better able to describe the phenomenon exactly as experienced (Husserl, 1954). In this study, the researcher's own experiences and preconceptions were written down at the beginning of the project. This information was then revisited during the analysis process to reduce the influence of researcher bias on the results in a substantial way. My own preconceptions in regard to this study stem from my experience with meeting both families and professionals, as well as from attendance to conferences and reading of literature over several years. I also have considerable clinical experience with other rare disorders apart from SMS, so my preconceptions are based in the range of issues facing persons with rare disorders and their families. Especially regarding how the parents experienced the challenging behaviours, I expected them to emphasise mostly on challenges with the lack of sleep, the aggressive behaviour, and especially aggressive behaviour outside of their home. I also believed they would emphasise that they had met a lot of professionals without knowledge about SMS, and therefore had endured a lot of interventions not tailored to their child. These preconceptions were revisited throughout the analysis process, in order to check that I didn't just follow my own preconceptions. I was thoroughly checking the data, to ensure that the data guided the analyses, and tried my best to have a reflected attitude about my own preconceptions. I also studied the various methodologies and methods, used in this study, and how to reduce biases.

4.2.1 Data collection

Open-ended questions were provided to the parents to answer in writing through a questionnaire. The questions were formulated on the basis of literature research and the researcher's experience with the population:

- Does your child have behaviour challenges? If yes, please describe them.
- What do you think is the cause of the challenging behaviours?
- How do you or did you feel about your own competence handling your child's challenging behaviours?
- What type of support and/or guidance did you receive for handling your child's challenging behaviours?

- What type of support and/or guidance would you have preferred for handling your child's challenging behaviours?
- What types of interventions have been tried for your child's challenging behaviours, and how well did they work?

After an analysis of the written answers from the parents, one particular theme emerged that required more investigation. In phenomenological research this is called revisiting participants to clarify or validate data (Creswell, 2013). Professionals lack of knowledge about the disorder was a major theme in the written responses, along with parents indicating they were responsible for training and information to professionals. More information about how the parents experienced the professionals lack of knowledge about the disorder and how this had impacted the parents were themes that needed validation and clarification in the revisiting of the participants. Therefore, additional oral interviews were completed with four parents. These parents were already part of the study and had provided written answers to the questionnaire. The interviews were audiotaped. These are the questions asked in the oral interviews:

- Professionals' lack of knowledge about the disorder was one of the themes that emerged from data collection. Do you have experience with professionals' lack of knowledge about the disorder, and if so, did the professionals' lack of knowledge about the disorder impact the guidance and follow-up you received?
- Do you believe that lack of knowledge about SMS in the support system and among professionals has led to challenges for you or your child?

4.2.2 Data analysis

The written data were transferred from handwriting to a computer and then were analysed verbatim by the researcher. The interviews were audiotaped and then transcribed and analysed verbatim by the researcher. The data were analysed using the phenomenological methods described in Creswell (Creswell, 2013). The stages in the analysis are similar to thematic analysis (Braun & Clarke, 2006). The following stages were then used in the data analysis: 1) familiarisation of the data by re-reading them repeatedly, 2) development of a list of significant statement, and 3) re-grouping of significant statements into themes (Creswell, 2013). On a more concrete level, all the transcripts were entered into NVIVO qualitative data analysis software (QSR International Pty Ltd., 2015). All the written responses were then re-read several times, before significant statements were collected from the written responses. Then the significant statements were sorted into groups with similar statements. From the groups of similar statements, the statements were grouped into topics, themes and categories. The data from the four in-depth interviews were used to confirm the findings from the open-ended questions and to supplement how a lack of knowledge among the support system and professionals affected the families and persons with SMS.

4.3 Q methodology

In Articles IV and V, the Q methodology was utilised. This methodology was developed and introduced by William Stephenson in 1935 (Stephenson, 1935). The Q methodology is designed to explore subjectivity by identifying unique viewpoints revealed as factor structures (through a by-person factor analysis) (Brown, 1986; Stephenson, 1953). The Q methodology can be regarded as a philosophy of science, a theoretical framework, a research technique for collecting data as well as an analytic method for scientific research into subjectivity (Brown & Good, 2010). The revealed viewpoints represent what is meaningful from the participants' perspectives and is meant to detect their subjective beliefs (Thorsen, 2009; Øverland, Thorsen, & Størksen, 2012). According to Stephenson (1935) *subjectivity* is a behaviour. More specifically, it is how the person sees the situation from his or her own standing or point of view (Stephenson, 1935). Since there is little research regarding how school staff experience students with SMS in schools it is of interest to explore teachers' subjectivity to identify the school staff`s experiences with children who has SMS. Q methodology was therefore considered a relevant choice of method. Q methodology is also considered to be a method well fitted for research in small populations, since it is people, not items that are grouped in the factor analysis (Newman & Ramlo, 2010). Little research regarding how the school staff experience students with SMS in schools, has been detected, and therefore Q methodology seem to be a relevant choice of method. Exploring the subjectivity of how the school staff view their work with children with SMS may lead to new discoveries. The methodology may also open up for themes we would not detect using standardised questionnaire or structured interviews. There are commonly five steps used in Q studies (Van Exel & de Graaf, 2005) (Figure 4-1).

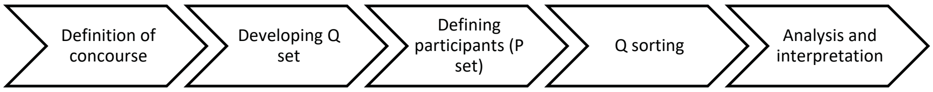


Figure 4-1. The process of a Q study

4.3.1 Definition of concourse

The concourse is defined as the universe of communicability regarding a specific topic - a collection of all possible statements about the subject of concern (Brown, 1993; Van Exel & de Graaf, 2005). It could come from different sources, such as photos, music, interviews, conversations, social media, magazines or scientific articles (Brown, 1980). The concourse in this project was sampled systematically using several sources: published sources (Haas-Givler & Finucane, 2014; Neira-

Fresneda & Potocki, 2015), open-ended questionnaires and standardised questionnaires (Developmental Behaviour Checklist and Vineland Adaptive Behavior Scales). These questionnaires were completed as another part of the project (article I and II).

4.3.2 Developing the Q set

The Q set is the final set of statements. As far as I know, no other studies have been looking at school staffs' experiences with SMS. Finding the concourse for these studies were therefore done by looking into research regarding SMS in general. Approximately 150 statements were collected from the literature and the questionnaires. To identify the concourse the structured questionnaires (VABS II, DBC, SCQ and SRS) used in other studies in this thesis were used, together with published articles concerning challenging behaviours in SMS (Haas-Givler, 2004; Haas-Givler & Finucane, 2014; Poisson et al., 2015). To create a balanced and structured set of statements, Fisher's balanced block design was used to structure and select representative statements from the concourse (Fisher, 1960; Stephenson, 1953). Fisher's balanced block design is a two-dimensional model with effect on one side and levels on the other side (Fisher, 1960). Two different balanced block design were used, one for each Q study in this thesis. In the first study (Table 1) *alone* and *with other students* were used at the effect side of the block design. *Extrovert* and *introvert* were used as levels. In the second study (Table 2) *methods, cooperation* and *guidance/knowledge* were on the effect side of the block, and *behaviours* and *academic* were on the level side. In both block designs extra statements categories was added for statements not covered within the balanced block design. In study 1 I called these "various" and "school staff experiences". The various category included statements not fitting in any of the other categories such as "the student is good at technical things" and "the student can be experienced as very intense". In study 2 the extra category was labelled "school staff's feelings". The same procedure was used for developing the two Q sets. The statements were then narrowed down to 40 (40 in each Q study). The statements were divided into categories and, assessed for duplication and similarities. For duplicated statements, one was kept, and one discarded, and similar statements were combined. The statements were printed on separate cards and numbered arbitrarily, and the generated statements are known as the Q set (Coogan & Herrington, 2011; Van Exel & de Graaf, 2005).

Table 1 Fisher's balanced block design Q study 1

	Extrovert	Introvert	School Staff Experiences
Alone	6 statements	6 statements	12 statements
With other students	6 statements	6 statements	
Various	4 statements		

Table 2 Fisher's balanced block design Q study 2

	Behaviours	Academic	School Staff Feelings
Methods	6 statements	6 statements	5 statements
Cooperation	5 statements	4 statements	
Guidance/Knowledge	7 statements	7 Statements	

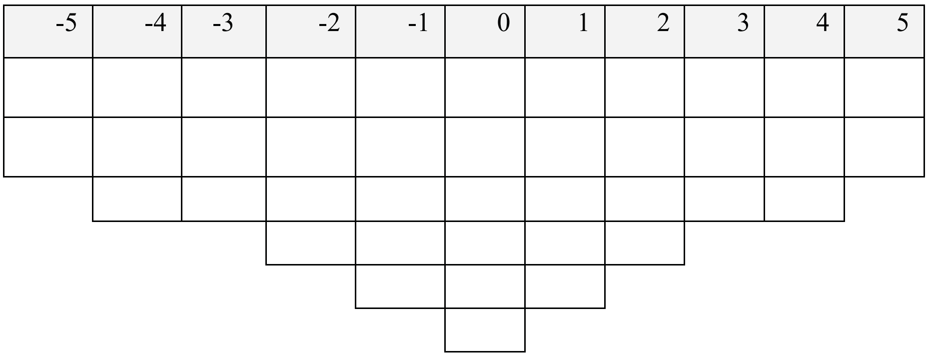
4.3.3 Participants (P set)

The P set refers to the group of participants in the study. The participants sort the statements (Figure 4-2). In these studies, the P set consists of school staff currently working with a student with SMS. The parents of school-age children with SMS were contacted for permission to contact their schools to participate in this study (The Norwegian Ethical Committee required such permission). Then, the principals of the 10 schools (I only know of approximately 10 students in grade school in Norway) were contacted by mail and were asked to distribute the Q sorts to three of their staff members who work with students with SMS. Fourteen (approximately 50%) participants completed the Q sort and returned them. One reminder email was sent to the principals after one month. The school staff represented elementary, middle and high schools.

Figure 4-2. School staff performing a Q sort.

4.3.4 Q sorting

The participants sort the Q set of statements into a grid. In this study, the participants sorted two different Q sorts in a row. The sort represents the participants' viewpoints. The participants receive an instruction with the research theme and are asked to sort the cards from most like to most unlike my viewpoint. In this study, a distribution grid with 11 categories (from +5 to -5) was created to fit 40 statement cards (Figure 4-3). Because the students with SMS are spread geographically all over Norway, and face-to-face Q sorts would be very ineffective, the Q sorts were therefore sent in the mail. The package included information about the study, written instructions, informed consent forms and the two Q sorts. The participants were instructed to sort the cards according to their experiences with the students with SMS. After the sorts, the participants were instructed to write down the numbers of the statements in the correct place in the grid (Figure 4-4). They were also instructed to provide a written rationale for the placement of the two cards placed on the far right (+5) and far left (-5) sides of the grid.



-5	-4	-3	-2	-1	0	1	2	3	4	5

Figure 4-3. The grid used in both Q studies (Articles IV and V)

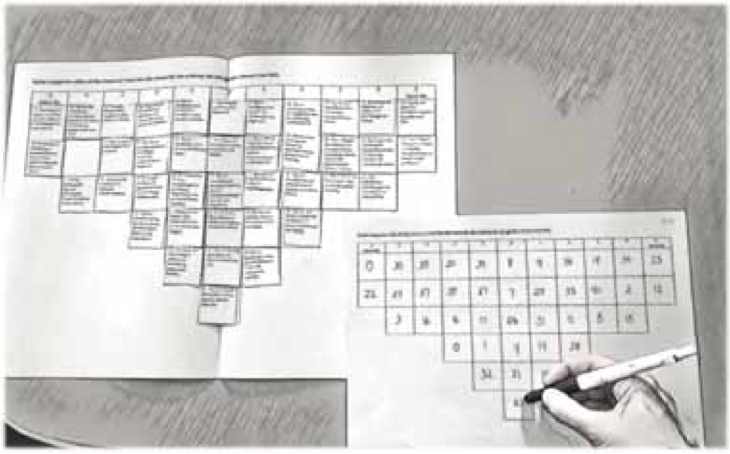


Figure 4-4 The participants writing down the numbers of the statements in the correct place in the grid

4.3.5 Data analysis and interpretation

The completed Q sort is correlated with the other participants' Q sorts (Coogan & Herrington, 2011; McKeown & Thomas, 1988; Schmolck, 2002; Stephenson, 1953). By factor analysing the results, it is possible to identify unique viewpoints/factors (Wheeler & Montgomery, 2009). All of the Q sorts were plotted and analysed using one of the available computer programmes, the PQ Method Programme (Schmolck, 2002), which utilises by-person factor analysis. In the analytic process, the correlation of all Q sorts is calculated. Then, the degree or level of dissimilarity and similarity of points between the individual sorters are calculated. Subsequently, factor analysis is performed to examine how many groupings of similar Q sorts there are. People with similar views (sorts) share the same factor/grouping (Van Exel & de Graaf, 2005). In Q methodology, the statements are utilised to define a factor, unlike traditional factor analysis, in which factor loading is used to interpret factors; moreover, a theoretical factor is constructed by a weighted average of the factor's score for the Q sort associated with this factor (Brown, 1980; Wheeler & Montgomery, 2009). Factor scores are essentially weighted z-scores for each statement in the Q-sample; these scores can be converted into an array of scores (factor array) that correspond to the plus 5 to minus 5 values in the original Q-sort continuum (McKeown & Thomas, 1988). The factors were interpreted based on the characteristic statements of each factor, as well as distinguishing and consensus statements. In addition, written statements regarding why the participants placed the statements on either end of the scale were used to clarify the results.

4.4 Study population (for all three methods)

The participants were recruited through the Frambu Resource Centre for Rare Disorders (Frambu) and the Smith-Magenis Foundations in Norway, Sweden and Denmark (family support groups). All three organisations disseminated information regarding the study via their Facebook sites and email lists. Frambu, which is one of nine publicly funded centres of expertise administered by the Norwegian National Advisory Unit on Rare Disorders, has its own registry, which is based on informed consent. Frambu could therefore send invitations to registered families with a child or adult with a diagnosis of SMS. The Swedish and Danish families were recruited through their respective foundations, both through information via their Facebook sites and through information at their annual gatherings. The only inclusion criterion was a genetically confirmed diagnosis of SMS. The diagnosis was confirmed by review of the genetic testing reports. The parents and patients older than sixteen years old provided written consent to participate in the study. The parents or guardian were the one who decided if the person with SMS had the capacity to consent in this case. If they decided the person with SMS had the capacity to consent, the person had to give their own consent. In the case where the parent or guardian decided the person with SMS did not have the capacity to consent, only the parent or guardian consented. These considerations were in accordance with the ethical approval.

In Norway, Frambu has registered slightly more than 40 persons with SMS. In Sweden and Denmark, the organisations know of approximately 20 persons with SMS in each country. According to the prevalence data (Greenberg et al., 1991), there should be slightly more than 200 persons with SMS in Norway and Denmark and more than 400 in Sweden. In this project, parents of 36 persons with SMS aged between 1 1/2 and 50 years old participated. The distribution across ages is shown in Figure 4-5. Fifteen of the persons with SMS were older than 18 at the time of the study. A total of 18 were from Norway, 13 came from Sweden, and 5 came from Denmark. Twenty fathers and 35 mothers participated. For 19 of the persons with SMS, both parents participated in completed the questionnaires; for sixteen of the persons with SMS, only the mother completed the questionnaires; and for one person with SMS, only the father completed the questionnaire.

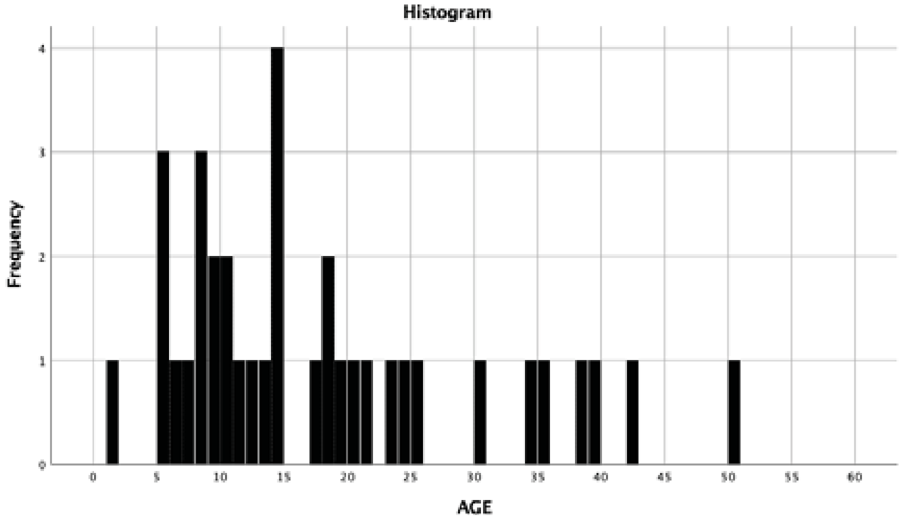


Figure 4-5. Distribution across ages

Seven of the persons with SMS did not have ID, five had mild ID, 15 moderate ID and one severe ID. In seven we did not have access to their degree of ID, either because they were not tested due to their young age (one from Sweden and one from Norway), or because I did not have access to their pedagogical charts (all Danish persons). These measures were collected from either medical or pedagogical charts, not measured explicit for this study. A range of cognitive measures were used to establish the degree of ID, the tests that were mostly used were; Wechsler Intelligence Scale for Children (both R, III and IV), Wechlers Adult Intelligence Scale, Wechlers Preschool and Primary Scale of Intelligence, Bayley Scale of Infant Development and Leiter -R. The tests were done at either the pedagogical centres or the habilitation centres, all by certified personnel.

In Articles I and II, the study population consisted of 28 persons from Norway and Sweden (Figure 4-6). The lack of data from Denmark in this study was due to the time it required to obtained permission from the Ethical Committee in Denmark and denial from the Ethical Committee to use the DBC due to a lack of Danish translation.

In Article III, the study population consisted of information from 37 parents. Sixteen of the questionnaires were answered in writing by both parents, 15 of the 32 questionnaires were answered by only the mother, and only the father answered one questionnaire. All of the parents answered in writing. One parent did not complete the qualitative semi-structured questionnaire (Figure 4-6).

In Articles IV and V, the study population consisted of 14 school staff personnel. Norwegian parents of children with SMS of school age were contacted for permission to contact their schools to participate in this study (the Norwegian Ethical Committee required such permission). Then, principals at 10 schools were contacted by mail asking to distribute the Q sort to three of their staff members working with students with SMS.

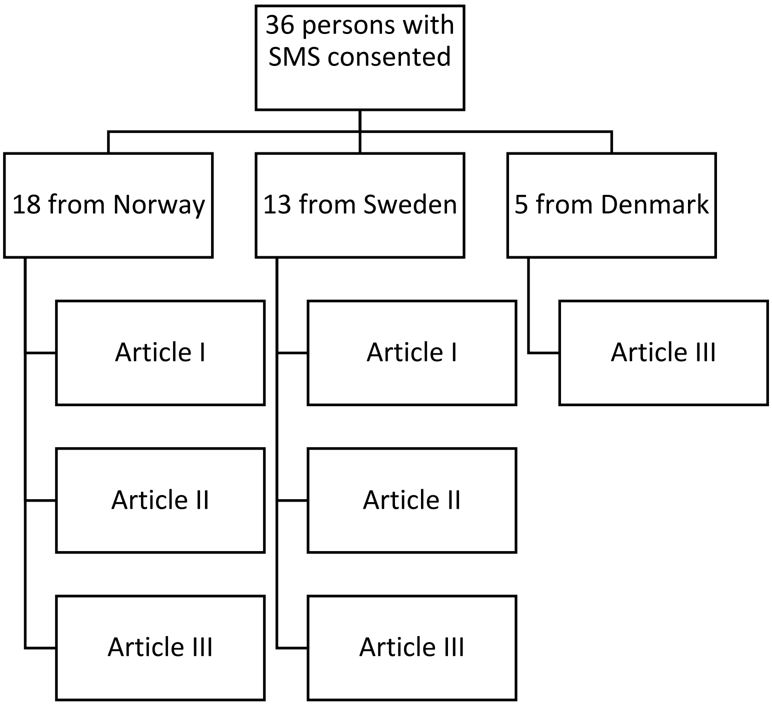


Figure 4-6. Overview of participants in three of the articles

4.5 Trustworthiness

To achieve trustworthy scientific research, issues regarding reliability and validity must be addressed.

In quantitative research, validity refers to whether the instrument measures what it is designed to measure (Field, 2014). Reliability refers to whether the instrument can be interpreted consistently across different situations (Field, 2014). In this study, there are several different quantitative instruments used. Each instrument is presented with information regarding validity and reliability under the description of the instrument earlier in this Methods section.

In qualitative research, the validity is a concern if the results are true or accurate and if they are transferable (Creswell, 2013; Malterud, 2003). In this study, strategies to ensure validity were conducted. Clarification of the researcher's bias was undertaken by bracketing my preconceptions. Member checking using in-depth interviews after analysis of written responses was also performed. Transferability of the results might be possible, and a rich description of the study population has been included to facilitate transferability to similar populations. Reliability in qualitative research often concerns the stability of responses across different coders of the data (Creswell, 2013). In this study, two researchers coded and discussed the themes of both the written responses and the in-depth interviews.

In Q methodology, according to Brown, "There is no external criterion of a person's own point of view" (Brown, 1980 p. 191). The main aim in Q methodology is exploration of subjectivity (Brown, 1980). One validity aspects of Q methodology is how well the Q set is balanced and how the researchers understand the individuals' viewpoints (Størksen & Thorsen, 2011). In this study, measures was undertaken to ensure a balanced Q set utilising Fisher's balanced block design (Fisher, 1960; Stephenson, 1953). A thorough investigation of the participants' written explanations of why they had chosen the specific statements that they placed on either ends of the grid was also conducted to ensure an understanding of the individuals' viewpoints. In Q methodology, you would not expect to obtain reliability coefficients of 1.00 in a test-retest but more likely 0.80-0.90 (Brown, 1980). According to Brown (1980), there are a limited number of viewpoints on any given topic. These viewpoints or perspectives will most likely be revealed in a well-structured Q sample. These studies were, as far as I know, the first studies regarding challenging behaviours in students with SMS in schools. More research is needed to confirm and broaden the findings.

4.6 Ethical aspects

The study was submitted for approval to the Norwegian Regional Committees for Medical and Health Research Ethics (REK # 2015/1026). REK approved of the research project (Appendix 6). The study was also submitted for approval to ethical committees in Sweden and Denmark. Dr. Ann Norgren was responsible for the ethical approval in Sweden and Dr. Mette Møller Handrup was responsible in Denmark.

4.6.1 Researcher's preconceptions

The researcher's preconceptions play a significant role in research. My previous experiences working with rare disorders in general and SMS specifically have most likely influenced this research project. For one thing, it has generated my interest in this topic and influenced the research questions stated in this project. It has also provided me with an in-depth insight into the disorder and a close connection with the families and the professionals working with the families.

Especially in phenomenological studies, it is important to set aside one's own personal experience by writing it down and bracketing it (Creswell, 2013). In the qualitative article (Article III) in this study, the researcher's own experiences and preconceptions were written down and revisited during the development of the questionnaires and interview guide and the analysis process to ensure that the researcher's beliefs and bias did not shape the interpretation.

In the Q studies (Articles IV and V), the selection of statements might be influenced by the researcher's preconceptions. Fisher's balanced block design (Fisher, 1960; Stephenson, 1953) was used to ensure a balanced Q set, and the steps of Q methodological research was further followed (Van Exel & de Graaf, 2005). The participants' own written explanations of why they had placed the statements in the way that they did were analysed to support the viewpoints. Data was analysed in the PQmethod program, where data consisting of arbitrary numbers representing the Q statements were plotted for analyses.

4.6.2 Research involving small populations

Anonymity per se is always an important topic in research, and as researchers, we always want to reassure our research subjects that their data are anonymous. However, this assurance is not always possible when conducting research in rare disorders due to the small population, and many of the patients in the group know (of) each other. Earlier research has discussed the difficulties of maintaining anonymity in research regarding rare disorders due to small affected populations (Griggs et al., 2009; Parker, Ashcroft, Wilkie, & Kent, 2004). This type of privacy is often referred to as "internal confidentiality" (Tolich, 2004) or deductive disclosure (Kaiser, 2009). This breach of confidentiality or privacy can occur when the traits of an individual or groups are made explicit in publications (Kaiser, 2009) or if data are recognisable to other research participants ("insiders") (Tolich, 2004). It is the researcher's obligation to ensure that the individuals' privacy is protected (Alver & Øyen, 2007). The Guidelines for Research Ethics in the Social Sciences, Humanities, Law and Theology (NESH, 2016) points out that researchers must take due caution if individuals can, directly or indirectly, be identified in publications. Almost all research on rare disorders in Norway, where data reported on an individual basis, would have data identifiable to people who know the group, such as family members, persons with the diagnosis and professionals working with one or more persons in the group. There are ways to avoid this identification of persons with the diagnosis to ensure internal confidentiality. One obvious way is to report data on groups of persons with the diagnosis, instead of individuals. However, sometimes this method is not possible due a small sample size. Thus, to render the data less identifiable, they can be reported without referring to geography or gender or using age range instead of accurate ages. All of these tools are typically used to avoid identification in research into rare disorders. However, sometimes it is not possible to use these tools because both gender and accurate age are important to report with the data. It is then of the utmost importance that these issues be clearly disclosed to the participants before they provide their consent.

Internal confidentiality is also often a problem in qualitative research, because the respondents are often described in an elaborated way to give the readers a broad understanding of the participants and the topics that are researched (Kaiser, 2009).

In this study, I am encountering this internal confidentiality problem from two sides: I am conducting research on a small population in which almost everybody knows each other, so the “insider” problem must be addressed; and I am conducting qualitative research, so the internal confidentiality must be addressed in how I am presenting the participants. One of the steps I have undertaken to ensure internal confidentiality is to include patients from Sweden and Denmark, in addition to the Norwegian population of patients with SMS. Doing so provides a larger group to research, rendering the data more difficult to identify, but it also provides three subgroups in which the participants in each group do not know the participants in the other groups very well. I do not report quantitative data on an individual basis but in groups, making identification almost impossible. The qualitative data are more difficult to deidentify, but that I have three subgroups that do not know each other across the groups makes it more difficult to identify individuals in the data. In the qualitative data, neither gender or age is of interest on an individual basis, and omitting this information makes it more difficult to identify individuals and therefore ensures better internal confidentiality.

Research regarding rare disorders will almost always pose challenges in publishing since the population is small, and recognition of persons with the diagnosis is possible. This fact is important for the participants to be aware of, and steps to render the data as unrecognisable as possible are important. It remains important to conduct research on rare disorders since information is scare.

4.6.3 Research involving vulnerable groups

This study involved persons with ID, and they are considered an especially vulnerable group in research (Liamputtong, 2005). In addition, some persons with ID have limited ability to consent to participation. Almost everybody debating vulnerable persons in research refers to Silva's (1995) definition regarding nursing research. According to Silva (1995), vulnerable persons often need some safeguards to ensure that their rights and welfare are protected. In U.S. federal regulations governing human research (Protection of Human Subjects, 2009), mentally disabled persons are defined as vulnerable populations. Research involving vulnerable populations is often under-represented in the scientific literature, often due to the more rigorous ethical considerations to obtain approval of the research projects (Moore & Miller, 1999). Ellingsen (2015) discussed that denying a population possibilities to participate in research in a systematic manner weakens the research, but denying a person his or her right to participate in research, based on a belonging to a population, is also an ethical question in itself (Ellingsen, 2015). In regard to rare disorders, it is important to include everyone in the population, both because of the size of the population and because of the general lack of information regarding all ages. Including adults, even if they might not have competency to provide their consent, warrants us to be extra sensitive when developing studies. This study includes children with ID, for whom the parents provide consent, but also adults (older than 16) with ID for whom both the parent/guardian and the person him- or herself provide consent. Only the adults viewed as having the capacity to give consent do so; I left it to the parents/guardian to decide whether the adult with SMS has the capacity to provide free and informed consent (Psykisk helsevernloven, 2002). The capacity to consent is not a static concept but is dependent on the situation, the project, and the information provided (De nasjonale forskningsetiske komiteene, 2009). In this study, I developed an easy-to-read information letter for the participants so that they can either read it themselves or have somebody read and explain it to them (Appendix 5). The parents or guardian were the one who decided if the person with SMS had the capacity to consent in this case. If they decided the person with SMS had the capacity to consent, the person had to give their own consent. In the case where the parent or guardian decided the person with SMS did not have the capacity to consent, only the parent or guardian consented. These considerations were in accordance with the ethical approval.

Including vulnerable persons in this study forces me to thoroughly examine the methods that I use to ensure that I do no harm to the participants. In this study, I mostly rely on information from the parents or guardians.

It is important to also include the voices of persons with disabilities in research (Ellingsen, 2015). In this thesis, their voices have not been heard due to the time and limitations on a PhD dissertation, but a new project has already been started focusing on how persons with SMS view their own diagnoses.

5 Summary of results

In the following section, the results of the articles included in this PhD thesis are presented.

5.1 *Article I*

This article explored autism spectrum disorders (ASD) in Smith-Magenis syndrome. Parents of 28 persons with SMS aged between 5 and 50 years old participated in this study; 11 of the persons with SMS were older than 18 at the time of the study. A total of 12 came from Sweden, and 16 were from Norway. The mean age was 17.5 years old with a range from 5.2-50.5. More than 50% scored above the cutoff on the Social Communication Questionnaire (SCQ), and more than 96% scored in the mild-moderate to severe range on the Social Responsiveness Scale (SRS). A significant gender difference was found in ASD symptomatology using the SCQ and SRS. Approximately 3 females per male above the SCQ cutoff. The same differences were not found in the intellectual level, adaptive behaviours, or behavioural and emotional problems. Gender had an independent contribution in a regression model predicting the total SCQ score, and neither the Vineland Adaptive Behavior Scale II nor Developmental Behaviour Checklist had an independent contribution to the SCQ scores. In conclusion, a clear, reversed gender difference in ASD symptomatology in persons with SMS was found. This outcome might be relevant in the search for female protective factors assumed to explain the male bias in ASD.

5.2 *Article II*

This article explores the Developmental Behaviour Checklist (DBC) profiles of persons with SMS and how they relate to measures of ASD symptoms, adaptive level and age**.** Parents of 28 persons with SMS aged between 5 and 50 years old participated in this study; 11 of the persons with SMS were older than 18 at the time of the study. A total of 12 came from Sweden, and 16 were from Norway. The mean age was 17.5 years old with a range from 5.2-50.5. The DBC results from 28 persons with SMS were analysed. DBC-Total scores decreased with age, but they still showed a mean clearly greater than the cutoff of 46. The differences between the age groups <9 years old and 9-17 years old ($p=0.024$) and between the age groups <9 years old and >18 years old ($p=0.007$) are significant. There is no significant difference between the age groups 9-17 years old and >18 years old. There are significant differences between the youngest and oldest age groups in all but the Communication Disturbance and Social Relating subscales. Between the youngest and middle groups, there are no significant differences when we examine p-values. There is a large effect between the youngest and middle groups on the Anxiety subscale when we use Cohen's D. Between the middle group and the oldest group, the only significant difference is in the Disruptive/Antisocial subscale. A significant decrease in behavioural and emotional problems with age was found in SMS. Adaptive skills were related to the behavioural problems, although this outcome must be further explored.

5.3 Article III

This article explores the experience of having a child with a rare disorder. Parents (31 mothers and 17 fathers) of 32 persons with Smith-Magenis syndrome (SMS) participated in this study. A phenomenological approach was used to divide the data into topics and themes. Four themes emerged: behavioural challenges displayed, parents' strategies for meeting the challenging behaviours, parents' experiences of their own competencies and parents' experiences of professionals' competencies and understandings regarding children with SMS and their behavioural challenges. We found that parents of children with SMS experience that they are exposed to severe challenging behaviours from their children. The parents believe that they experience more misunderstandings with professionals and that the challenging behaviours increase because there are some specific characteristics of SMS of which professionals are not aware or do not consider in their support services.

5.4 Article IV

This article explores utilising Q methodology regarding how school staff experience the behaviours of children with SMS in school and how they cope when working with these children. An important aim in this article was to investigate whether and what type of challenging behaviours students with SMS display in schools. Varimax rotation revealed two distinct viewpoints regarding the school staff's experiences with challenging behaviours in students with SMS, namely: 1) managing challenging aggressive and self-injury behaviours in schools; and 2) struggling with intense non-physical challenging behaviours in school. In viewpoint 1, school staff experience a range of challenging behaviours in school, both aggressive behaviours and acting out behaviours. The staff manages these behaviours and has a positive attitude towards both the work and the students. Staff experienced that the students become angry at school; they hit, scream, kick and have self-injurious behaviours, but the school staff still experienced the work as positive and challenging in a good way to work with these students. In viewpoint 2, school staff experienced that the students are challenging to work with, especially because of the intensity of their behaviours, but the staff is positive towards the work and the students. However, the staff experiences demanding situations especially if the staff members are alone with the students. In this viewpoint the staff experienced that these students have more non-physical behavioural problems, such as being very intense, craving attention and pushing buttons. The staff holding this view do not experience the acting out behaviours, such as kicking, screaming and self-injurious behaviours, as being as problematic as the more non-physical behaviours. In this view, the staff members were positive towards working with these students, but they also found it demanding and struggled with being alone with the students with SMS. The staff experienced the students as emotionally immature, having a lack of impulse control, lacking the ability to focus on schoolwork over time and having trouble sitting quietly by themselves. Most of the consensus statements was about how the school staff experienced the students' behaviours and not about the school staff's coping and emotions regarding the behaviours. According to the results of this study, there must be a greater emphasis on education and advising and supporting the school staff's work with the non-physical challenging behavioural aspects of teaching children with SMS, as well as the focus on challenging aggressive behaviours.

5.5 Article V

This article explores the school staff´s experiences of how to manage the challenging behaviours of students with SMS. In this study, the staff`s work methods and resources working with students with SMS in school were investigated utilising Q methodology. Four factors regarding the teachers' views concerning methods and resources in their work were identified: 1) in control, 2) struggling, 3) strugglers relying on parents and 4) support dependent. In the first viewpoint, the school staff received guidance and information regarding SMS and manage the work well. The staff members enjoy their work and feel safe, although the students display challenging behaviours. In the second viewpoint, the school staff experiences are that it is difficult to work with students with SMS because of the challenging behaviours, and the staff members struggle because they must do things a little differently than they are doing with other students. They also believe that it is difficult to inform parents of challenging behaviours because of the parents' feelings, but they work well with the parents. In this group, they also lack support from the leadership at the schools and from other colleagues. In the third viewpoint, the school staff has received information and guidance from the parents and not as much from the school leadership or guidance from anyone other than the parents (pedagogical centres, for example). In the fourth and last viewpoint, the school staff has received guidance and training regarding SMS and support from the school leadership and colleagues. The staff have not received much information regarding the disorder from the parents and are not working much with the parents. Fourteen statements do not distinguish between any pair of the viewpoints. The staff agree, across all viewpoints, that preparations both ahead of activities and to be ahead of difficult situations are important for students with SMS. The staff members also agree, across viewpoints, that they do have a structure that prevents challenging behaviours. Several of the statements regarding academic work with the students are consensus statements, and they have scores in the middle (+2 to -2). It seems as it is consensus regarding academic work not being a priority for these students. To be in control and feel safe when working with students with SMS, school staff is dependent on support from the school's leadership and colleagues, in addition to cooperation with parents. School staff feeling safe and in control seems to be important for shifting the focus from handling challenging behaviours to academic work regarding students with SMS.

6 Discussion

In this PhD thesis, the focus has been on exploring challenging behaviours in persons with SMS and how their parents and school staff are coping. One of the objectives was to explore the relationship among specific characteristics of the syndrome, communication, challenging behaviours and ASD (Articles I and II). Another of the main objectives was to examine how the school staff (Articles IV and V) and parents (Article III) experience handling the challenging behaviours. Children of school age spend almost 1/3 of their day in school, and adults with SMS do spend time in school if they have access to adult special education. It is therefore important to determine how to manage and reduce challenging behaviours in schools and other learning situations regarding persons with SMS.

The results of the five enclosed articles are discussed separately. In this section, some of the results of the five articles are discussed across two main topics; Challenging behaviours in SMS in schools and Challenging behaviours in SMS. These two topics are used to discuss the results from the five different articles across each other to widen the findings in each article. Some practical implications are also suggested.

Individual development plays a central role both in Bronfenbrenner's bioecological model and in the PPCT model (Bronfenbrenner, 1977; Bronfenbrenner & Morris, 1998). The characteristics of the person with SMS influence the different microsystems and the overall environment with which he or she interacts (Bronfenbrenner, 1995; Bronfenbrenner & Morris, 1998). Sameroff developed a model of how these characteristics (genotype and phenotype) influence the microsystems and environment in general (environtype) (Sameroff, 2009). I therefore attempt to discuss the findings of this thesis in the context of how development and change can occur, according to Bronfenbrenner and

Sameroff. I start with how school staff member experience working with students with SMS and how challenging behaviours in schools appear from the school staff's view.

6.1 Challenging behaviours in SMS in schools

One important finding of these studies is that students with SMS display challenging behaviours in schools, and school staff copes with these challenges in a diverse way. In the first Q study (Article IV), two viewpoints were revealed, and they differ both in the type of challenging behaviours viewed as problematic and in how well the staff manages the challenging behaviours. In the second Q study (Article V), the four identified viewpoints especially differed in how they perceived support from school leadership and colleagues, as well as cooperation with parents. The results showed that both support from the school leadership and cooperation with parents were important to how well the school staff managed the challenging behaviours.

According to Bronfenbrenner, communication both within and between systemic levels is important (Bronfenbrenner, 1979). In the microsystem, Bronfenbrenner emphasised activities, roles and relations and their influences on each other in a child's development (Bronfenbrenner, 1979). As mentioned earlier, Bronfenbrenner adjusted the description of the microsystem at a later stage, including the influence of other persons' (significant persons) distinctive characteristics on the microsystem (Bronfenbrenner, 2005). Using Bronfenbrenner's bioecological model, I assume that how well the school staff handles the challenging behaviours and the support that they have both within the microsystem (from colleagues and school leadership) and between microsystems (cooperation with parents) can influence the development of the student. In Sameroff's model of development, how the school staff manages the challenging behaviours will be part of the environtype, but the environtype will also consist of how the school leadership and colleagues support the staff, how the participation with parents works, and so on. The environtype will then influence the student's behaviours, i.e., the phenotype. The school environment is in this way a part of what is shaping the student's development. Some of the school staff members indicated that they were tired of their job, experiencing it as demanding being alone with the student, and they scored low on thinking that it is challenging in a good way to work with students with SMS (Article IV). Several studies have indicated that challenging behaviours can influence the student-teacher relationship in a negative way (Hamre & Pianta, 2005; Patterson & Fisher, 2002). Pianta's research also indicated the importance of a positive relationship between teacher and student to the effect of both academic and social gain (Pianta & Hamre, 2009; Pianta & Stuhlman, 2004). Several reports regarding special education in Norway (Barneombudet, 2017; OECD, 2005) have indicated a lack of resources or a lack of use of resources regarding special education. Among other tasks, the principals at the schools are responsible for both the daily management and the development of the organisation (the school) (The Education Act, 1998).

The statement regarding difficulties in explaining the students with SMS to other staff at the school received a high agreement score from several school staff across the viewpoints (Article IV), which could indicate a lack of knowledge in the school system regarding SMS in general and how the challenging behaviours manifest. How well the school staff members are coping could depend on how supported they feel by colleagues and the school leadership (Lazarus & Folkman, 1984). Knowledge and support can lead to more problem-focused coping than emotion-focused coping (Lazarus & Folkman, 1984). Lack of knowledge in any of the micro- or macrosystems around the person with SMS can influence development. Lack of knowledge and information is also in accordance with what Harazni and Alkaissi (2016) found in their research regarding teaching children with ADHD.

In Q study 2 (Article V), there was agreement across all four viewpoints that structure, preparation, clear boundaries and being ahead of the student were needed for these students. This required structure is in accordance with the high levels of ASD in these students (Article I). Focusing on structure and preparing the students before activities can lower the anxiety and influence the experience of stress for the students.

6.1.1 Schools staffs' experiencing the challenging behaviours of students with SMS

In the first Q study (Article IV), the findings indicate that the school staff is divided regarding how they experience the challenging behaviours of students with SMS in the schools. The two different viewpoints included 1) managing aggressive and self-injury behaviours in school and 2) struggling with intense non-physical challenging behaviours in school. The non-physical challenging behaviours are behaviours such as lack of impulse control, lacking the ability to focus, pushing school staff's "buttons", being demanding, picking on things and talking all the time. It does seem as though the school staff experiences the more intense non-physical challenging behaviours as more demanding than the aggressive and self-injury behaviours. There was consensus among the staff in the two viewpoints regarding how the staff experienced the students' behaviour (such as "the student has good impulse control" and "the student never destroys things at school"). The school staff also agrees that it is positive to work with these students, and that they know what to do when challenging behaviours occur. They also experience it as somewhat difficult to explain to other teachers about the student and how to handle her/his behaviours. The distinguishing statements were especially those regarding how the staff handled the behaviours. The staff who loaded on viewpoint 2 were more scared when the student lost control, they experienced it as more demanding working alone with these students and they also experienced that they did get tired of their job sometimes. The staff loading on viewpoint 1 experienced it as challenging in a good way working with these students.

Lazarus described coping as having the skills to manage external or internal demands that are taxing or that exceed one's resources (Lazarus & Folkman, 1984). Since some of the school staff members struggled with the challenging behaviours of students with SMS, it seems that the demands exceed their skills to manage the behaviours that they are experiencing, and they are not coping. They lack knowledge about how to deal with these types of behaviours. Coping is important in how we manage stressful situations (Drageset, 2014; Lazarus & Folkman, 1984). The school staff requires information and knowledge of how to handle the different types of challenging behaviours in students with SMS. By adding these, their perceptions about these students may change, as well as their self-efficacy when addressing the challenging behaviours and the needs of their students, thus giving teachers more tools for a more effective coping process'. Both problem-focused coping and emotion-focused coping are important in these situations. They must also learn how to manage their own emotions in the situations. Due to the high prevalence of challenging behaviours in students with SMS, school staff will encounter these challenges. School staff requires tools and knowledge to both handle the physically aggressive behavioural challenges and the intense non-physical challenging behaviours in schools, as well as tools to manage their own emotions in the situations.

6.1.2 How school staff handle the challenging behaviours in students with SMS

In the second Q study (Article V), how the school staff handles the challenging behaviours in students with SMS and the support needed were investigated. Four distinct viewpoints were discovered: 1) in control, 2) struggling, 3) strugglers relying on parents and 4) support dependent. Those loading on viewpoint 1 (in control) distinguish from the other viewpoints on the statements that indicate that they do not believe the challenging behaviour stop them from having an academic focus and they report not to be afraid when students get angry. In this viewpoint they are also looking forward to their workdays because of the students' charm, humour and love. Those loading on viewpoint 2 (struggling) distinguish from the other viewpoints on statements such as believing it is difficult to work with these students because of their challenging behaviour and thinking it is hard when parents get upset because of the feedback from the school staff. They lack support from both colleagues and the leadership at the school and do not receive sufficient follow up after difficult episodes. Several statements distinguish viewpoint 3 (strugglers relying on parents) from the other viewpoints. School staff holding this view experience to receive good support from the parents and do not lack training regarding SMS but report they have not received any training in how to handle the challenging behaviours. Still, they report not to get afraid when the students get angry. Finally, those loading on viewpoint 4 (support dependent) distinguish from the other viewpoints on statements such as getting support after difficult episodes and having no problem following the recommendations regarding the challenging behaviours. The staff holding this view has not been trained by the parents and value the cooperation with the parents less than the staff in the other three viewpoints. In all the viewpoints, the staff agrees that both preparation for the student ahead of the activities and to be ahead of difficult situations are important for students with SMS. Across all the viewpoints they also agree that they do have a structure that prevent challenging behaviours. Several of the consensus statements regard academic work with the students with SMS and they have scores in the middle. It looks like it is consensus around the fact that academic work is not a priority for these students. This will be discussed further later in the Discussion section.

An important finding of this study was the relation between the support from the school leadership and how well the school staff handles the challenging behaviours (Table 3). It seems as though support from school leadership and colleagues is important for school staff members to experience that they are in control and to feel safe working with students with SMS. Those loading on viewpoint 2 seem to be in a particularly difficult position experiencing lack of support from school leadership and colleagues and experiencing it as challenging to work with parents.

Table 3 Viewpoints divided on parents support and leadership support

		Support from school leadership and colleagues	
		Positive	Negative
Parent support and cooperation	Positive	Viewpoint 1, in control	Viewpoint 3, strugglers relying on the parents
	Negative	Viewpoint 4, support dependent	Viewpoint 2, struggling

Support and cooperation with parents are also important pieces, but if the staff only has these pieces and not support from the leadership and colleagues, it seems as if it is difficult to manage the challenging behaviours in the schools. Lack of leadership support was also found by Harazni and Alkaissi (2016) in research regarding teaching children with ADHD. Cooperation between the different microsystems, such as school and parents, is a crucial part of the mesosystem in Bronfenbrenner's bioecological model (Bronfenbrenner, 1995). Bronfenbrenner's PPTC model incorporates how the different parts of the model influence the person's development (Bronfenbrenner & Morris, 1998). Both the context, consisting of the micro- and mesosystems (among others), and the process, consisting of the interaction between the individual and the environment, are important. These factors are also crucial parts of Sameroff's transactional development model (Sameroff, 2009). How the school leadership and colleagues support school staff working with students with SMS is an important part of the environment and can influence the development of the student.

Another finding in this study is that the school staff of male and female students seems to manage challenging behaviours differently. Most of the staff members working with male students seem to load on viewpoint 1: in control. In this viewpoint, it seems as though they both work well with parents and receive support from the school leadership and colleagues. This finding could indicate that it is more challenging to work with females with SMS than males. It is especially in the social domain that females have more problems than males (Article I). It might be that problems in the social area impose more challenges on the school staff than other behavioural challenges. Article I also indicate that more females than males scored in the moderate and severe/profound level of ID and had lower scores on adaptive behaviour measured by the Vineland Adaptive Behavior Scale, but neither difference was significant. In general, it seems as though some of the important skills needed in schools, such as social competence, adaptive behaviours and cognitive level, are lower in females than males. It seems that school staff members working with males manage challenging behaviours better than school staff working with female students with SMS. The gender differences are discussed further below.

6.1.3 Comparison between the two school staff studies (Articles IV and V)

In the first Q study of school staff's experiences with challenging behaviours in students with SMS in schools, two viewpoints emerged: 1) managing challenging aggressive and self-injury behaviours in school; and 2) struggling with intense, non-physical challenging behaviours in school. In the second Q study, on how the school staff manages challenging behaviours in students with SMS in schools, four viewpoints emerged: 1) in control, 2) struggling, 3) strugglers relying on parents and 4) support dependent. Both Q studies are independent and have a different set of statements; therefore, it is difficult to compare them. However, since it seems as if, in both studies, there are some staff members who manage better than others, I wanted to compare the results to investigate whether there are similarities in the two studies (Table 1).

Table 4. Comparison of factors on the two Q studies.

		Q study 2 – How to handle challenging behaviours			
		Factor 1 – In control	Factor 2- Struggling	Factor 3 – Strugglers relying on parents -	Factor 4 - Support dependent
Q study 1 – Experience and coping of challenging behaviours	Factor 1 - Managing	TA TA T U O		T T	T T
	Factor 2 - Struggling		U TA O	T	

T: Teacher, TA: Teaching assistant, O: Other school education, U: Unknown profession

More than half of the participants who loaded on viewpoint 1, managing challenging aggressive and self-injury behaviours in school, in the first study also loaded on viewpoint 1, in control, in the second study. None of the participants who loaded on viewpoint 1 in the first study, loaded on viewpoint 2, struggling, in the second study. Conversely, almost all of the participants loading on viewpoint 2, struggling with intense non-physical challenging behaviours in school, in the first study loaded on viewpoint 2, struggling, in the second study. It looks as though those who are struggling with non-physical challenging behaviours in school also struggle with a perceived lack of leadership support and somewhat with cooperation with the parents. Lack of support from the leadership at schools has been identified as a major stressor for school staff (Shernoff, Mehta, Atkins, Torf, & Spencer, 2001). Shernoff et al.'s (2001) research also found that teachers believed that a lack of parental support from other sources (such as parenting workshops, mental health resources) prevented the parents from supporting the school staff. I found in Article III that parents of persons with SMS experienced a lack of support in general.

Another factor regarding the difference in how school staff manages different challenging behaviours might lie in the focus on regulations of the use of restrictive practises in schools. Restrictive practises mean "any practise or intervention that has the effect of restricting the rights or freedom of movement of a person with disability" (NDIS Quality and Safegurads Comminsion). These restrictions may include seclusion and restraint. There has been a focus on the use of restrictive practises in schools in Norway in recent years (Højmark, 2016). There are no laws or regulations regulating the use of restrictive practises in schools, which means that restrictive practises are not allowed to be used in schools. Use of restrictive practises are only allowed to be used in cases of necessity or self-defence, either to protect others or you self from risk (Straffeloven, 2005). School leadership has focused on other strategies than restrictive practises to handle challenging behaviours. This increased focus on challenging behaviours and alternative interventions might have led to more support for school staff working with students with aggressive and acting out behaviours (Hansen & Østvold, 2015). The results of Article IV reveal that this outcome might not be the case with non-physical challenging behaviours for students with SMS and that the school staff working with those students still lacks support.

6.1.4 Focus on academic work for students with SMS

One of the findings in this study from the second Q study (Article V) is the lack of focus on academic work. Neither of the statements regarding academic work truly ends up on either end of the grid. They are almost all concentrated from -2 to +2 in all four viewpoints, with a few exceptions. It seems as though the school staff has very strong opinions or attitudes towards the challenging behaviours, cooperation with parents or support from school leadership and colleagues. In regard to the academic work, these strong opinions or attitudes seem to fade away. Udwin et al. (2001) found a lack of progress in educational achievement from childhood to adulthood in persons with SMS. They also found low abilities in other areas, such as independence in daily living skills and occupational achievement. This discrepancy between different abilities and the cognitive level has been attributed to behavioural challenges (Udwin et al., 2001). Udwin et al. (2001) asked whether this lack of ability could be a function of limited educational input or a ceiling in abilities regarding persons with SMS. In Norway, all students have a right to receive an education and to have a school environment adapted to their abilities (The Education Act, 1998). The Education Act states that all students are entitled to a physical and psychosocial environment conducive to learning (The Education Act, 1998 § 9a). This study indicates that the school staff might have a limited academic or educational focus. It seems as if the challenging behaviours do capture much of the focus regarding this disorder in school. In Article II, an indication was detected of a relation between daily living skills and challenging behaviours. Therefore, an effort to shift the focus from only handling the challenging behaviours towards a focus on learning and educational outcomes might provide a decrease in challenging behaviours. In general education, one of the suggestions for preventing challenging behaviours in school is to provide explicit and engaging academic instructions (Alter et al., 2013).

6.2 Challenging behaviours in SMS

Knowledge regarding challenging behaviours is important if we want to improve the behaviours. Knowing why, when, where and how challenging behaviours occur is necessary (Stoesz et al., 2016). In this section, how the challenging behaviours appear across different aspects of the life of a person with SMS is discussed, which could provide a broader picture of the challenging behaviours necessary to understand the behaviours. Knowledge about both the environment (environtype) and the phenotype is important to changing development and challenging behaviours (Sameroff, 2009).

6.2.1 ASD in SMS

ASD symptomatology are more prevalent in genetic syndromes than in the general population (Richards et al., 2015). This is also evident in SMS (Laje et al., 2010). Research over the years have also indicated an association between ASD symptomatology and greater severity of ID (Richards et al., 2015). In this thesis, I didn’t investigate the prevalence of ASD per se, since this has been established previously, but more the relations between ASD symptomatology and other characteristics. One interesting research topic arising the last years, concerns how specific behavioural outcomes are connected to the underlying genetic difference between and within syndromes (Oliver, 2019). Not only whether ASD symptomatology in genetic syndromes is associated with the degree of ID, but also to what degree social communication and repetitive behaviours in specific genetic disorders differ from social communication and repetitive impairment in idiopathic autism (Richards et al., 2015). This was not the topic of this thesis, but in the first article I found a gender difference in ASD symptomatology, favouring the females. It was especially in the social domain females with SMS differed from females with other aetiological pathways to ASD (Article I). Differences in social preference and social motivation in persons with SMS have previously been researched (Wilde, Mitchell, & Oliver, 2016; Wilde et al., 2013). In article III in this thesis parents indicated controlling behaviours from their children as a major issue in this syndrome. Both differences in social communication and repetitive behaviours may be mistaken as ASD symptomatology, especially if measured with less sensitive measures such as SRS and SCQ but may only be phenotypic symptoms of SMS not necessarily fulfilling criteria for an ASD diagnosis.

6.2.2 Challenging behaviours and relations to other symptoms

In the two first articles, the nature of the challenging behaviours in persons with SMS and the relations between the symptoms to suggest some intervention regarding the challenging behaviours were explored. Individual characteristics, such as gender, age, communication, behaviours and intelligence, were investigated to find relations with the challenging behaviours. In the first article, a reversed gender ratio in ASD was found. Analysis of any of the other symptoms also found a gender difference but revealed a non-significant difference in the adaptive behaviour composite score (VABS). Neither the developmental level nor behavioural problems could explain the gender difference in ASD (Article I). In Article II, a more direct look was taken into the behavioural problems and into whether anything had an influence on the challenging behaviours. A significant decrease in challenging behaviours with age was found. This outcome is the opposite of what was reported earlier regarding challenging behaviours and age in SMS (Finucane, Dirrigl, & Simon, 2001; Neira-Fresneda & Potocki, 2015; Poisson et al., 2015). No significant relations between challenging behaviours and other symptoms regarding SMS were detected. Neither adaptive behaviours nor communication had a significant relation with behavioural and emotional problems in SMS. The only strong relation that I found was between challenging behaviours and activities of daily living (ADL) skills, which might be of great importance and should be investigated further. ADL skills are important to functioning in kindergarten, schools and society in general. The direction of the relation is not indicated; therefore, I do not know whether poor ADL skills lead to more challenging behaviours or whether challenging behaviours lead to poorer ADL skills. The results of poor ADL skills are in concordance with Udwin et al. (2001), who found little independence in daily living skills, especially in the adult population with SMS. However, this relationship is possibly suitable for interventions since it is a possible skill to implement in the schools.

The mean percentile on the Disruptive/Antisocial subscale in DBC, which mainly measures aggression, was 87.57. The range was in the range of the 50th to 98th percentiles, with one person on the 50th percentile but all of the others between the 78th and 98th percentiles (Article II). The subscale of disruptive/antisocial behaviours in DBC does not separate between proactive and reactive aggression, only confirming that there are aggression issues. There are persons with SMS who have no ID or mild ID. It would therefore be interesting to also investigate the different types of aggression that they display as a possible route to differentiating interventions. Examining both common characteristics and individual characteristics described in developmental psychology in relation to challenging behaviours could provide us with input on where to focus our interventions and being able to predict challenging behaviours.

6.2.3 Challenging behaviours in SMS across settings and situations

In these studies, information regarding the challenging behaviours of persons with SMS in different ways and across different situations has been collected. The parents completed standardised questionnaires and described their children's challenging behaviours qualitatively. The school staff described the challenging behaviours by sorting different type of behaviours in a Q methodology Q sort. In general, challenging behaviours as described in earlier research were identified (Sloneem, Oliver, Udwin, & Woodcock, 2011). Both aggressive behaviours and self-injury were identified in Articles I and II, and such behaviours were also described by the parents in Article III and by school staff who loaded high on the viewpoints in Article IV. In particular, confirming that these types of behaviour occur in schools are important. No scientific studies investigating challenging behaviours in schools regarding students with SMS have been performed before, as far as I know.

In the qualitative study of parents' experiences, two new aspects surfaced regarding challenging behaviours that have not been thoroughly researched before. The first subject regarded inappropriate behaviours; such as undressing, self-stimulation, talking to strangers and repeatedly asking questions. These behaviours have been observed in SMS (Wilde et al., 2016), but in this study, the inappropriate behaviours came across as more troublesome than previously documented (Poisson et al., 2015; Wilde et al., 2016). Farrugia (2009) investigated how inappropriate behaviours in children with ASD affected the stigmatisation of the parents and found high levels of stigmatisation and adaptation of their everyday lives due to the inappropriate behaviours. This type of behaviour also seemed to be frequently observed in the schools by school staff (Article IV). Inappropriate behaviours in school could lead to poorer peer relations and influence interactions with other students

(DuPaul & Weyandt 2006). The other aspect was regarding controlling behaviours. This type of challenging behaviour is well known in ASD (Constantino & Charman, 2016). In SMS, this behaviour is displayed by controlling how the parents sit, what they wear, what they say and which words they use (Article III). I have heard parents discuss this theme but have never seen it mentioned in the published research. It may also be that this type of controlling behaviour also is a part of the intensive, non-physical challenging behaviour the school staff in article IV struggled with. The individual characteristics of a person with SMS will impact the individual, the context and the process in Bronfenbrenner's PPTC model (Bronfenbrenner & Morris, 1998). In Sameroff's model of development, these behaviours are somewhat influenced by the genotype, and they influence the environtype (Sameroff, 2009).

6.2.4 Challenging behaviours in SMS across age

In two of the articles, a difference in challenging behaviours across ages (Articles II and III) was found. In Article II, significant age group differences in the DBC Total, with a decrease with age, were presented. The studies in this thesis included more adults with SMS than did earlier studies, but a decrease in challenging behaviours from age group 1 (<9 years) to age group 2 (9-17 years) is still interesting, although the decrease was not significant in all of the subscales. It is also important to underscore that the levels of challenging behaviours in adults with SMS are still high and far greater than the cut off on the DBC Total. In Article III, a difference was found in the number of challenging behaviours reported with more challenging behaviours reported by parents of children younger than 18. Both of these findings are the opposite of what was reported earlier in research, in which challenging behaviours were found to increase with age (Finucane et al., 2001; Neira-Fresneda & Potocki, 2015; Poisson et al., 2015). The studies in this thesis included more adults than previous research, which might be part of the reason for the different results, but this finding requires further research. In the two

Q studies, the number of students with SMS was too small to examine age differences. A decrease in challenging behaviours with age is positive information for both parents and school staff.

6.2.5 Gender differences in SMS

Gender differences were found in three of the articles (I, III and V), and it seems as though females with SMS have more challenges than males. In the first article, approximately three females per male greater than the SCQ cutoff was found, which is exactly the opposite of what is expected in a sample with idiopathic ASD. It is particularly in the social domain of ASD that females with SMS differ substantially from females with other aetiological pathways to ASD. This reversed gender difference could prove useful in more general ASD research when searching for the mechanisms underlying female protective effects (Lai, Lombardo, Auyeung, et al., 2014).

The reversed gender ratio of ASD symptoms identified in this study cannot be explained by differences in either developmental level or the number of challenging behaviours. The clinical diagnoses of intellectual disability differ between genders, and a tendency for poorer development in females (VABS II total 53) than males (VABS II total 62) was found. This difference was not significant. A similar gender difference was found in Article III. Parents of females with SMS reported more challenging behaviours, more adaptations and a greater lack of competence and support.

According to the last article, the school staff working with male and female students seems to handle challenging behaviours differently. Most of the staff members working with male students seem to load on viewpoint 1: in control. In this viewpoint, it seems as though they both work well with the parents and attain support from the school leadership and colleagues. This finding could indicate that it is more challenging to work with females with SMS than males. No gender differences were found in the first Q study (Article IV). Three other articles present gender differences in SMS (Edelman et al., 2007; Huang et al., 2016; Laje et al., 2010). In the study by Edelman et al. (2007), the authors found some gender differences, with the females showing more problems. Most of them were somatic (myopia, cold hands and feet, eating/appetite problems and possible hypersensitivity (problems finding shoes to fit)), but they also found that females had significantly higher frustration with communication levels.

Gender is a distinctive characteristic around which we build our beliefs and expectations (McClintock et al., 2003). In Bronfenbrenner's bioecological model, the microsystem includes significant others' distinctive characteristics, such as their systems of belief (Bronfenbrenner, 2005). One of the beliefs that school staff might bring to its work is that males will display more challenging behaviours than females since most of the research has drawn this conclusion (McClintock et al., 2003). In contrast, people often do not expect this level of challenging and aggressive behaviours from female students; therefore, challenging behaviours are experienced as more of a struggle than the same behaviours displayed by male students.

6.3 Limitations and strengths

The use of multimethod design may be a limitation in this thesis. A PhD study has some restrictions regarding time and resources. The use of three different methods investigating the topic from different views (parents and school staff) is challenging. One of the weaknesses is that I have not been able to examine each view in depth. Neither did I manage to explore the children`s views in this PhD theses. However, I believe the use of multimethod design have given some important results that may help children with SMS, their parents and their teachers. The study also identifies themes that ought to be further researched.

The main limitation of this study is the sample size. Even though about 50% of the population is included the sample is too small to be confident in some of the quantitative results. This power problem is particularly evident when investigating subsamples and when conducting linear regression.

The small number of participants, especially in the quantitative studies and also to an extent in the Q study, is a limitation. This issue is a known challenge in research regarding rare disorders (Griggs et al., 2009). This limitation must be borne in mind when drawing conclusions, especially regarding the quantitative results. I have attempted to rectify this issue by including persons of all ages and from three countries to have as large a sample as possible.

The use of indirect methods is another limitation. The challenging behaviours of persons with SMS in this PhD study are researched utilising questionnaires completed by parents or caregivers, written responses and interviews with parents and Q statements sorted by school staff. I have not conducted any direct observations or interviews with the persons with SMS. This has some implications regarding ASD in SMS. The use of checklists such as SRS and SCQ instead of the more in-depth measurements such as ADI-R and ADOS have implication on the level of certainty of the ASD symptomatology described in this thesis.

Yet another limitation is the use of cross-sectional data, which prevents me from concluding regarding development over time. In particular, Article II, in which challenging behaviours between different age groups are discussed, would have improved with longitudinal data. The differences between the age groups could be due to cohort differences or rapport bias. Other characteristics, such as ASD and adaptive behaviours, were also investigated to minimise these biases.

There is also a limitation concerning the level of ID of the persons with SMS in this thesis. First of all, I do not have information about the level of ID for all the persons in these studies. Level of ID is only used in the first two articles, and for all persons included in those articles information about level of ID is given. Furthermore, the most important limitation probably is that I did not measure the level of ID myself. Since administering formal psychometric assessments might be difficult with people med SMS, due to the maladaptive behaviours, sleep disturbance, and difficulties in language skills (M. R. Smith et al., 2009) and the focus of this thesis was challenging behaviour, and use of indirect methods, I chose to collect formerly administered tests of ID. Due to these limitations, level of ID have only played a supportive role in the analysis. The last limitation concerning level of ID, is that it seems like that the population in this study have a different cognitive level than populations in other SMS studies (Dykens, Finucane, & Gaylay, 1997; Madduri et al., 2006; Martin et al., 2006; Osório et al., 2012; Udwin et al., 2001). In most studies regarding cognitive profile of SMS only mean and range are reported (Dykens et al., 1997; Madduri et al., 2006; Osório et al., 2015) and almost all research indicates ID from mild to moderate, but several studies have a range that exceed IQ points of 70. A few studies report the number of participants on each level of ID. In Martin et al. (2006) 28% fell within the borderline ID, and 6% in the low average range of cognitive ability. Most research and descriptions on SMS, includes ID as one of the symptoms (Dykens et al., 1997; Elsea & Girirajan, 2008; Gropman et al., 2006; A. C. Smith, Dykens, & Greenberg, 1998a). The diversity of SMS has changed from the first description by Smith et al. (1986) and with a more widespread use of genetic testing, more persons with SMS without ID may be detected (Prescott, 2013). Even though this sample of persons with SMS may have more participants without ID than other samples with SMS, all other measures in this thesis are consistent with other characteristics of SMS. All participants in this thesis scores above the cutoff for severe behavioural and emotional problems in the DBC and the scores of ASD symptomatology are similar to those reported in other SMS research (Laje et al., 2010). In spite of the broader diversity in ID in this sample, I believe the results are comparable and may be generalised to other population of persons with SMS.

The interpretation of both themes in the qualitative study and the factors in the Q studies may deviate from how the parents or school staff actually experience it. In the qualitative analysis, a member check was performed in the oral interviews with a few of the participants to confirm and check if the analysis of the written responses were interpreted accurately. No such member check was performed regarding the Q studies, but the participants own written explanation of why they placed the specific statements on +/- 5 were used to interpret the factors.

My previous experience with and knowledge of SMS might also constitute a limitation. Especially in regard to analysing the data. I might have had preconceptions that influenced the analytic process, especially in the qualitative methods and selection of statements in the Q methodology.

As with all methods, Q methodology has its' limitations. Some of the limitations and strengths are presented in chapter 4.5 (about validity and reliability in Q studies) and 4.6. (about preconceptions, small samples). First of all, results from Q studies cannot be generalised (John & Montgomery, 2016). Even though I cannot generalise to all school staff working with students with SMS, the results from these Q studies may guide future focus and research. Also, the fact that the participant can only respond to pre-determined statements may be perceived as a limitation (Cross, 2005). The statements in the Q studies have been selected systematically utilising Fisher's balanced block design and abductive reasoning (Brown, 1986; Fisher, 1960; Haig, 2008). The Q set could have been broader by including more statements regarding challenging behaviours in school and how school staff handle these challenges, based on the SMS literature. I chose to only include behavioural characteristics regarding SMS since some of these characteristics are specific to SMS and that the specific characteristics were a theme in this thesis.

There might be a limitation in how the Q sorts were executed. I mailed the participants and did not perform the sort face-to-face. In a face-to-face setting, misunderstandings can be solved, and body language can also be considered. It is recommended that the Q sorting is followed by an interview (Van Exel & de Graaf, 2005). In this interview the participants are asked why they sorted the way they did. In this study, the participants answered this in writing. A face-to-face interview of participants who loaded high on each factor would have be beneficial and would be important to include in later Q studies in this population. Studies have shown that Q sorts sent in the mail or performed using a computer have no differences in reliability or validity as compared to interview-based (face-to-face) Q sorts (Reber, Kaufman, & Cropp, 2000; Van Tubergen & Olins, 1979).

However, it is of great interest to perform a Q methodology study with children with SMS to obtain "a snapshot" of their subjectivity regarding their lives with SMS (Brown, 2006; Stephenson, 1953; Øverland et al., 2012). In this PhD study, the time limit and complexity of including the persons with SMS prevented me from doing so, but a study of how persons with SMS experience their own syndrome is already planned.

This study is, as far as I know, the first Q study performed regarding SMS. It appears to be a useful method for exploring school staff's viewpoints in handling challenging behaviours in schools. Some of the results, such as lack of leadership support and being tired of their jobs, might have been difficult to reveal in an interview setting or standardised questionnaires.

The strengths of this PhD study are the use of multimethod design, to explore challenging behaviours in persons with SMS from various perspectives. The use of qualitative, quantitative and Q methodologies in a multimethod design studying the same concept (challenging behaviours in persons with SMS) strengthens the validity (Abowitz & Toole, 2010; Esteves & Pastor, 2004). These methods have led to some new findings presented in the summary section and that are further summarised in the concluding comments section.

Another strength is that almost 50% of the known population with SMS in Norway and Sweden participated in the study. As described in this project, these families struggle greatly in their daily lives, which might be why half of the population chose not to participate in this study.

6.4 Concluding comments

The first aim of this PhD thesis was to explore the characteristics of SMS in the Scandinavian population and the relations between the different specific characteristics (Articles I and II). I found important information valuable to the staff responsible for the adaptation of persons with SMS in schools. In Article I, I found that more than 50% scored above the cutoff on SCQ, and 96% scored in the mild-moderate to severe range on the SRS. The high level of autism spectrum symptoms is important to be aware of when planning learning for persons with SMS. The two most interesting findings in these two articles are the reversed gender ratio of autism spectrum disorder in SMS (Article I) and the decreases in behavioural and emotional problems with age (Article II). The decreases in behavioural and emotional problems with age are especially interesting since earlier research on SMS has reported the opposite (Finucane et al., 2001; Neira-Fresneda & Potocki, 2015; Poisson et al., 2015). This decrease might also indicate that the work that schools are doing, related to challenging behaviours, is working towards decreasing the behaviours. There is a need for research into what the schools are actually doing to decrease challenging behaviours. Another finding in Article II is the negative relation between VABS ADL subscale and challenging behaviours. This finding indicates that those with the most challenging behaviours have the poorest ADL skills. I cannot define the direction of this relation and can therefore not tell whether poor ADL skills lead to more challenging behaviours or whether more challenging behaviours lead to poor ADL skills. However, teaching ADL skills is something that kindergarten and school staff is able to do and focusing on these skills might be important to prevent challenging behaviours.

The second aim was to investigate the experiences and how school staff manages and handles challenging behaviours in persons with SMS (Article IV). The importance of this aim was to investigate whether and what type of challenging behaviours students with SMS display in schools. Two distinct viewpoints were found: 1) managing challenging aggressive and self-injury behaviours in school and 2) struggling with intense non-physical challenging behaviours in school. The school staff in viewpoint 1 experiences a range of challenging behaviours, both aggressive behaviours and acting out behaviours. The staff members manage these behaviours and have a positive attitude towards both their work and the students. Staff experienced that the students become angry at school, and they hit, scream, kick and have self-injurious behaviours, but the school staff still experienced it as positive and challenging in a good way to work with these students. School staff members in viewpoint 2 experience that the students are challenging to work with, especially because of the intensity of their behaviours, but the staff is positive towards its work and the students. However, the staff experiences demanding situations, especially if alone with the students. The staff experienced that these students have more non-physical behavioural problems, such as being very intense, craving attention and pushing buttons. The staff members holding this view do not experience the acting out behaviours, such as kicking, screaming and self-injurious behaviours, as being as problematic as the more non-physical behaviours. In this view, they were positive towards working with these students but also found it demanding and struggled with being alone with students with SMS. In this view, the staff experienced the students as emotionally immature, having a lack of impulse control, lacking the ability to focus on schoolwork over time and having difficulty sitting quietly by themselves. The implications of these different views are discussed in Article IV. The importance of this aim was to investigate the school staff's beliefs regarding challenging behaviours and the type of challenging behaviours that students with SMS display in schools.

In this aim, I also focused on how the school environment can adapt to meet the students' needs (Article V). Four viewpoints were revealed regarding what the school staff needed in support to handle the challenging behaviours: 1) in control, 2) struggling, 3) strugglers relying on parents and 4) support dependent. In the first view, the school staff members received guidance and information regarding SMS and are handling the work well. They enjoy their work and feel safe even though the students display challenging behaviours. In the second view, the school staff members experience is that it is difficult to work with students with SMS because of the challenging behaviours, and they struggle because they must do things a little differently than they are doing things with other students. They also believe that it is difficult to inform parents about challenging behaviours because of the parents' feelings, but they work well with the parents. In this group, they also lack support from the leadership at the schools and from other colleagues. In the third view, the school staff members receive information and guidance from the parents and not as much from the school leadership, as well as guidance from sources other than the parents (pedagogical centres, for example). In the fourth and last view the school staff members receive guidance and training regarding SMS and have support from the school leadership and colleagues. They have not received much information regarding the disorder from the parents and are not working much with them. In all of the viewpoints, the staff agrees that both preparation for the student ahead of the activities and being ahead of difficult situations are important for students with SMS. Across all of the viewpoints, the staff members also agree that they do have a structure that prevents challenging behaviours. Several of the consensus statements regard academic work with the students with SMS, and they have scores in the middle (2 - -2). It appears that there is consensus around academic work not being a priority for these students. The implication of these different views is discussed in the article (Article V) and in the discussion section of this thesis.

The last aim of this thesis concerns the parents' experiences of having a child with SMS, with a focus on the challenging behaviours (Article III). Four themes emerged in this study: behavioural challenges displayed, parents' strategies for meeting the challenging behaviours, parents' experiences of their own competence and parents' experiences of professionals' competence and understanding regarding children with SMS and their behaviour challenges. I found that parents of children with SMS experience that they are exposed to severe challenging behaviours from their children. The parents believe that they experience more misunderstandings with professionals and that the challenging behaviours increase because there are some specific characteristics of SMS that professionals are not aware of or do not consider in their support services. How the professionals experience the lack of knowledge and experience with SMS is important to investigate at a later point.

The use of a multimethod design and data from different sources afforded me the opportunity to explore challenging behaviours in SMS from different perspectives. It has been useful, and new knowledge has been found due to exploring the same topic with several methods and across different informants (school staff and parents) and settings (home and school).

6.5 Practical implications and future challenges

Based on the results of this thesis, I have some suggestions regarding work with persons with SMS and their families. In this thesis, I have established that students with SMS have a variety of challenging behaviours in school (Article IV). It specifically looks as though students with intense non-physical challenging behaviours are more of a challenge to work with, likely due to a lack of knowledge about how to manage this type of challenging behaviour. It also appears as though the school staff members experiencing the intense, non-physical challenging behaviours lack the support from the school leadership that they need to handle their jobs (Articles IV and V). The lack of knowledge and lack of support could be risk factors for stress (Lazarus & Folkman, 1984).

I also found gender differences, specifically in autism spectrum disorders (Article I). This reversed gender difference inspired me to examine gender differences in how both school staff and parents described and handled challenging behaviours. I found both a gender difference in how parents described the challenging behaviours displayed by their children (more challenging behaviours described by parents of females with SMS) (Article III) and gender differences in how school staff is handling the challenging behaviours in school (most school staff members working with males with SMS seem to experience being in control) (Article V).

The importance of cooperation with parents was also an important finding in Article V, but Article III draws a picture of parents pushed to their limits. The parents themselves experience a lack of knowledge in how to handle challenging behaviours but still must be the one to educate the professionals, including school staff, regarding the disorder. Based on these findings, I suggest further research into how to provide better support for parents, as well as relations with school issues.

The parents experience that there are some specific characteristics of SMS that professionals are not aware of or do not consider in their support services. Parents express that this lack of understanding exposed them to both more behavioural challenges from their children and more misunderstandings with the professionals. It is important to also investigate how the professionals view the experience of giving guidance regarding a disorder they probably not have any experience with from before and knowledge are scarce.

It is important to educate school staff to handle both challenging, aggressive and self-injury behaviours and intense non-physical behaviours when working with students with SMS. It is also important to educate or inform school staff about the challenges of cooperating with the parents of students with severe challenging behaviours. The school leadership should take note of the importance of their support, along with support from colleagues.

The future challenges in this field of educating students with SMS include determining how to impact or change challenging behaviours. I have found that there could be a relationship between ADL skills and challenging behaviours, and this connection must be further researched. Another challenge is how to change the focus from only focusing on the challenging behaviours to focusing more on academic work. It is important that these students also have a learning focus in schools. Finally, it is important to explore how the persons with SMS themselves experience the disorder. Ecological perspectives are important to ensure that children grow up in a good, secure environment.

7 References

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PART II

8 Article I-V

Paper I

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Molecular Autism

RESEARCH Open Access

Reversed gender ratio of autism spectrum disorder in Smith-Magenis syndrome

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Abstract

Background: A substantial amount of research shows a higher rate of autistic type of problems in males compared to females. The 4:1 male to female ratio is one of the most consistent findings in autism spectrum disorder (ASD). Lately, the interest in studying ASD in genetic disorders has increased, and research has shown a higher prevalence of ASD in some genetic disorders than in the general population.

Smith-Magenis syndrome (SMS) is a rare and complex genetic syndrome caused by an interstitial deletion of chromosome 17p11.2 or a mutation on the retinoic acid induced 1 gene. The disorder is characterised by intellectual disability, multiple congenital anomalies, obesity, neurobehavioural abnormalities and a disrupted circadian sleep-wake pattern.

Methods: Parents of 28 persons with SMS between 5 and 50 years old participated in this study. A total of 12 of the persons with SMS were above the age of 18 at the time of the study. A total of 11 came from Sweden and 17 were from Norway.

We collected information regarding the number of autism spectrum symptoms using the Social Communication Questionnaire (SCQ) and the Social Responsiveness Scale (SRS). Adaptive behaviour was also measured using the Vineland Adaptive Behavior Scale II. The level of intellectual disability was derived from a review of the medical chart.

Results: We found significant gender differences in ASD symptomatology using the SCQ and SRS questionnaires. We found approximately three females per male above the SCQ cutoff. The same differences were not found in the intellectual level and adaptive behaviour or for behavioural and emotional problems.

Gender had an independent contribution in a regression model predicting the total SCQ score, and neither the Vineland Adaptive Behavior Scale II nor the Developmental Behaviour Checklist had an independent contribution to the SCQ scores.

Conclusion: We found a clear reversed gender difference in ASD symptomatology in persons with SMS. This may be relevant in the search for female protective factors assumed to explain the male bias in ASD.

Keywords: Gender, Autism symptomatology, Smith-Magenis syndrome

Background

A substantial amount of research shows a higher rate of autistic type problems in males compared to females. The 4:1 male to female ratio is one of the most consistent findings in autism spectrum disorder (ASD) research [1–3], and a gender difference has been a part of the description of ASD since the first characterisation of the disorders.

ASD occurs in conditions with X-linked recessive inheritance, but because of the rarity of these disorders, this inheritance cannot explain the male bias in prevalence of ASD [4]. The fact that most ASD risk loci are found in autosomal regions makes the male bias in ASD largely unexplained [5]. Most current data suggest that the male bias is more likely to be due to female protective factors rather than male-specific risk factors, but comprehensive molecular explanations are lacking for both [6].

Gender ratios in ASD differ substantially from study to study. Among individuals with ASD and normal cognitive functioning, gender differences as high as 9:1 have been reported [7]. A newer systematic review and meta-

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analysis from Loomes et al. [8] found a male to female ratio closer to 3:1 than 4:1. According to Loomes et al. [8], the main reasons for this change were both how ASD was diagnosed and what population were used to investigate the male to female ratio in ASD in different studies. Loomes [8] found that studies screening the general populations for ASD had a lower male to female ratio than studies investigating population with pre-existing diagnosis. In cohorts with ASD in combination with intellectual disability, the ratio varies between 2:1–7:1 [2, 4]. Loomes et al. [8] also found a lower male to female ratio in their meta-analysis in the subgroup of the studies including participants with lower IQ. Epidemiological studies describe the degree of intellectual disability and the ascertainment approach as major explanations behind the varying ratios that were reported [9].

The particular biological aetiologies of autistic problems are probably also relevant, even when the degree of intellectual disability (ID) is controlled for, but such a line of investigation has not yet been explored. How different biological pathways to ASD differ in the ASD-gender ratio may shed light on basic ASD biology.

ASD is in the Diagnostic and Statistical Manual V (DSM V) referred to as a dyad of impairments; difficulties in social interactions and social communications; and restricted and repetitive behaviour, interests, and activities [10]. Gender differences in profiles of autistic symptoms have a limited research base [11]. Several studies [11–13] have found that males have more restricted and repetitive behaviours than females. Some studies have found that females have more impairment in social reciprocity and communication than males, but these findings are not consistent [13]; others have found that females with ASD have better sociability skills than males with ASD [14].

Lately, the interest in studying ASD in genetic disorders has increased, and research shows a higher prevalence of ASD in some genetic disorders than in the general population [15]. The focus so far has been on the prevalence and phenomenology in different syndromes, and further studies are required to tell us more about the differences in ASD phenomenology between ASD in genetic syndromes and idiopathic autism. Using the Autism Screening Questionnaire (ASQ), Oliver et al. [16] found a high level of autism (> 45%) in individuals with Cornelia de Lange syndrome (CdLS) and fragile X syndrome (FXS) (only males with FXS participated in the study) but lower levels in individuals with cri du chat syndrome (CDCS), Angelman syndrome and Prader Willis syndrome (PWS) (< 20%). Individuals with Lowe syndrome and Smith-Magenis syndrome (SMS) were more in the middle with approximately 35% scoring above the cutoff for autism. No significant gender differences in any of the syndromes were found. Another study concerning tuberous sclerosis (TSC) found no significant differences between females and males regarding ASD [17]. Recently, Nærland et al. [18] published an article regarding gender differences in Down syndrome. The gender ratios in their sample were approximately 2M:1F, which is slightly less than expected in idiopathic ASD with the same degree of ID.

SMS is one of the rare disorders where ASD has been described as a prominent part of the disorder [19] but also a disorder where gender differences in ASD symptoms, favouring females, have been found. Laje et al. [19] found that females had higher *T* scores on the Social Responsiveness Scale (SRS) total and on the 'Social cognition' and 'Autistic mannerisms' subscales. In this study, they did not control for the gender differences already accounted for in the gender-specific norms. They did not find any significant gender differences in Social Communication Questionnaire (SCQ) scores [20].

SMS is a rare and complex genetic syndrome caused by an interstitial deletion of chromosome 17p11.2 [21] or a mutation on the retinoic acid induced 1 (RAI1) gene [22]. Most SMS patients have a deletion containing 76 genes [23], but the patients with mutations in the RAI1 gene also display most of the core features of SMS, which indicates that the RAI1 gene is a dosage-sensitive gene responsible for most of the symptoms in SMS patients [24]. The disorder is characterised by intellectual disability, multiple congenital anomalies, obesity, neurobehavioural abnormalities and a disrupted circadian sleep-wake pattern [25]. The incidence of SMS is estimated to range from 1:15,000–1:25,000 births [26]. Delayed diagnosis is common, although the use of array-CGH and SNP-array analyses in routine clinical practice, together with greater recognition of the syndrome in the last decade, has led to earlier diagnosis [27].

Children and adults with SMS appear to have unique neurobehavioural problems that are challenging for both parents and professionals. These problems include sleep disturbances, self-injurious and maladaptive behaviours, stereotypies, and sensory integration disorders [28]. A thorough investigation of aggressive behaviours of a cohort with SMS showed that self-injurious behaviour, physical aggression and destructive behaviour were all significantly more prevalent in persons with SMS compared with a cohort of persons with IDs of mixed aetiologies [29]. In this study, 96.9% of participants displayed self-injurious behaviour, 87.5% exhibited physical aggression, 81.3% showed destructive behaviour and 43.8% were verbally aggressive [29]. SRS scores consistent with ASD have also been identified in almost 90% of the investigated populations with SMS [19]. A progression of autistic-like behaviour has also been described in young children with SMS [20]. A study comparing several genetic disorders (PWS, FXS, CdLS, CDCS, etc.) found that persons with SMS scored higher (were more impaired) than PWS and CDCS in the social domain, but in the two other domains (communication and repetitive behaviour), they did not differ from the other groups [16].

In addition to the study by Laje [19] mentioned earlier, two other studies have looked at gender differences in SMS [19, 30]. In an animal model study, Huang et al. [24] found a sexually dimorphic phenotype regarding obesity in mice (females were significantly more obese than males) with loss of RAI1 functions, but due to high mortality, they did not investigate this any further and the cause of the sexually dimorphic phenotype is not clear. Edelman et al. [30] found some somatic differences between males and females such as myopia, cold hands and feet, eating/appetite problems and possible hypersensitivity (problems finding shoes to fit) in females; Edelman et al. also found that females had more frustration with communication than males.

Measuring ASD in genetic syndromes is fraught with some difficulties. Individuals with known genetic syndromes are usually excluded from the standardisation of ASD assessment tools, and it is known that degree of intellectual disability influences these tools [31]. Additionally, it is recently documented that the commonly used ASD assessment tools are highly influenced by parent-reported behavioural and emotional problems [32]. In the SMS population with its varying cognitive abilities and high rates of behavioural problems, it is therefore important to control for these factors when making claims about ASD symptomatology.

The main aim of this study was to investigate gender differences in rates and profile of ASD symptoms in SMS when controlling for rates of emotional and behaviour problems and adaptive behaviour as a proxy for developmental level. Based on previous research and our own clinical experiences, we hypothesised that the usual increased rate of ASD symptoms in males (the male bias) would be absent in a sample of individuals with SMS.

Methods

Recruitment and participants

This study was part of a larger assessment study of SMS in Norway and Sweden. The participants were recruited through Frambu Resource Centre for Rare Disorders (Frambu) and the Smith-Magenis Foundations in Norway and Sweden (both family support groups). Both organisations spread information regarding the study via their Facebook sites and email lists. Frambu, which is one of nine publicly funded centres of expertise administered by the Norwegian National Advisory Unit on Rare Disorders, has its own register, which is based on informed consent. Frambu could therefore send invitations to registered families with a child or an adult with a diagnosis of SMS. The Swedish families were recruited through the Swedish Smith-Magenis Foundation both through information via their Facebook site and through information at their annual gathering. The only inclusion criterion was a genetically confirmed diagnosis of SMS. The diagnosis was confirmed by review of the genetic testing reports. The parents and the patients above the age of 16 provided written consent to participate in the study.

Parents of 28 persons with SMS aged between 5 and 50 years participated in this study; 12 of the persons with SMS were above the age of 18 at the time of the study. A total of 11 came from Sweden and 17 were from Norway (all the Norwegian patients were recruited through Frambu). In Norway, we know of 36 patients diagnosed with SMS and in Sweden 20; we have thus included approximately 47% of the Norwegian and approximately 55% of the Swedish SMS population. In Norway, 58% ($n = 21$) are females and in Sweden, 50% ($n = 10$) are females.

The level of ID was derived from a review of the medical charts. Consents were given to collect medical charts from the paediatric/habilitation and pedagogical centres. The levels of ID were collected from these charts. There was a wide variety as to who administered the test, with what instrument and at what age the level of ID was established.

Demographics

The demographics are displayed in Table 1. The mean age was 18.5 with a range from 5.1–50.5. The intellectual disability (ID) level was available from medical charts; seven of the patients did not have ID. It seems that more females had lower levels of ID, but this gender difference was not significant (asympt. $p = 0.07$).

Table 1 Demographics

	Total	Females	Males
N	28	15	13
Mean age	18.5	16.2	22.2
Range	5.1–50.5	5.1–33.9	5.1–50.5
Genetics			
Deletion	25	12	13
Mutation	3	3	0
ID grade			
No ID	7	3	4
Mild	5	1	4
Moderate	15	10	5
Severe/profound	1	1	0

Instruments

The Social Communication Questionnaire (SCQ) is a standardised screening tool for ASD [33]. The SCQ was used to assess the number of autism symptoms [33]. The questionnaire is used from the age of four. It contains 40 items, which are answered with 'Yes' (= 1) or 'No' (= 0) and comes in two versions. SCQ-Current covers the individual's behaviour during the most recent 3 months, whereas SCQ-Lifetime is based on the individual's entire developmental history. Both versions give a single total score, where a score of 15 or above is regarded as an indicator of possible ASD. The SCQ are also scored in three different domains: the reciprocal social interaction domain, communication domain and repetitive domain. In this study, the SCQ-Lifetime questionnaire was used [34]. In the initial standardisation of the assessment tool, a good reliability was reported with a Cronbach's alpha of 0.84–0.93 across the age groups and a Cronbach's alpha of 0.81–0.92 across the diagnostic groups [33]. Rutter et al. [33] also measured the validity and found a correlation of 0.71 between SCQ and the Autism Diagnostic Interview-Revised (ADI-R). In two groups of children with Down syndrome (DS) with ASD and DS without ASD, Magyar et al. [35] investigated the validity of SCQ and found that it did discriminate between the two groups. Children with DS and ASD obtained a significantly higher total score on the SCQ than children with DS only. SCQ is used in research on different genetic disorders [35, 36] including SMS [19].

The Social Responsiveness Scale (SRS) is a 65-item, quantitative parent-reported or adult self-reported measure that assesses social impairment associated with ASD [37]. The SRS enquires about specific and observable elements of reciprocal social behaviour (39 items), social use of language (6 items) and behaviour characteristics of children with autism and other PDDs (20 items), and it generates a standardised score. In addition to a total score, SRS consists of five subscores: Social Awareness, Social Cognition, Social Communication, Social Motivation and Autistic Mannerisms. In the initial standardisation of the questionnaire, the reliability was tested across gender and parents' and teachers' reports and in clinical settings. A good reliability was reported across these groups with a Cronbach's alpha of 0.93–0.97 [37]. The validity of the SRS has also been evaluated and a strong association between the SRS and the ADI-R was found [37]. Recently, in a large sample of idiopathic ASD, the SRS scores were shown to be influenced by rates of behavioural problems [32]. We therefore use both the SCQ and the SRS in this study and we assess the effect of behavioural problems. Since the SRS *T* score norms are different for males and females, we chose to use raw scores in addition to *T* scores when comparing the genders.

The Developmental Behaviour Checklist (DBC) [38, 39] is a questionnaire completed by parents or other primary caretakers or teachers that report problems over a 6-month period. Each behavioural description is scored on a 0, 1 and 2 rating where 0 = 'not true as far as you know', 1 = 'somewhat or sometimes true', and 2 = 'very true or often true'. Five versions of the Checklist are available: the Parent/Carer version (DBC-P), the Teacher version (DBC-T), the Adult version (DBC-A), the Short-form (DBC-P24) and the Monitoring chart (DBC-M). In this study, the DBC-P was used.

The Vineland Adaptive Behavior Scale II (VABS II) [40, 41] is a semi-structured interview or rating form of the parents or caregivers that assesses the everyday behavioural functioning of children and adults from birth throughout life. In this study, both the interview form (Norwegian cohort) and the parent/caregiver rating form (Swedish cohort) were used. The scales yield standard scores (mean = 100: one standard deviation (SD) = 15) in the domains of communication, daily living skills, socialisation and motor function, as well as a total sum score on adaptive behaviour composite. Each domain contains several subdomains. Motor function can only be assessed in children less than 6 years of age. In this study, the Norwegian and Swedish versions of the scales based on Scandinavian normative data were used. VABS II is a standardised and validated tool. Many studies have confirmed its reliability and validity making this measure one of the most widely used assessment tools of adaptive behaviour [42]. This tool has also been used with SMS several times [43, 44].

Table 2 Social Communication Questionnaire scores

	Total ($N = 27$[a])	Females ($N = 15$)	Males ($N = 12$)	p value (Cohen's d)
SCQ total (SD)	16.04 (6.10)	19.07 (4.77)	12.25 (5.55)	0.003 (1.32)
Reciprocal Social Interaction (SD)	5.19 (3.05)	6.87 (2.83)	3.08 (1.78)	0.000290 (1.60)
Communication (SD)	5.07 (2.73)	5.93 (1.98)	4.00 (3.22)	0.086 (0.72)
Repetitive behaviour (SD)	4.81 (2.19)	5.27 (1.91)	4.25 (2.45)	0.252 (0.46)
	N(%)[ratio]	*N*(%)	*N*(%)	
Number above cutoff (≥ 15)	14(52)[2.93]	11(73)	3(25)	0.021

[a]One parent did not return the SCQ questionnaire

The SRS, SCQ and DBC were all mailed to the parents after they consented to participate in the study. The parents filled in the information at home and mailed the questionnaire back to the researchers in a prepaid envelope. The VABS II were conducted in two different ways; the Norwegian cohort was interviewed on the telephone, and the Swedish cohort was mailed the parent/caregiver rating form together with the other questionnaires. The difference in procedure was due to language issues of performing the telephone interview with the Swedish cohort.

These instruments were chosen, instead of the gold-standard instruments ADI-R and Autism Diagnostic Observation Schedule (ADOS), because of their ease of use, because they have been used earlier with SMS, and to assess persons scattered around Norway and Sweden with the least possible burden for the patients.

Statistical analysis

Data were compiled for statistical analysis using the Statistical Package for the Social Sciences (SPSS) version 23 (IBM). Analysis of group differences in the degree of ID was conducted with the Mann-Whitney independent sample test. Descriptive statistics were derived, and the total scores and subscores obtained from the SRS and the SCQ were analysed as continuous dependent variables using t tests. The ratio was calculated as number of females above the cutoff on the SCQ total score divided by the number of males above the cutoff. Effect sizes (Cohen's d) were calculated using Social Science Statistics' online resources. The two-sided Fisher's exact test was used to test the proportion of males and females above the SCQ cutoff and in the different SRS classifications. Multiple regression analyses were conducted to assess the impact of 'gender', 'DBC' and 'VABS II standard scores' on the 'total SCQ score'. The normality of the residuals was checked using the visual inspection of P-P plots. Due to the combination of dichotomous and continuous predictor variables, we report the standardised coefficients (β), in addition to unstandardized B.

Results

Social Communication Questionnaire

The SCQ scores from 27 patients were analysed. A total of 52% scored above the cutoff (≥ 15). The females had higher scores on both the SCQ total score and all domains, but only the total SCQ score and the reciprocal social interaction domain showed a significant gender difference. A total of 25% of the males and 73% of the females scored above the ≥ 15 cutoff ($p = 0.021$). This provides a gender ratio of 3:1 and favours the females. All the SCQ scores are summarised in Table 2. The means for the males and females on each SCQ subdomain score are plotted in Fig. 1. How the ID grades are distributed between the males and females with SCQ scores above versus below the ASD cutoff are displayed in Table 3 (females) and Table 4 (males).

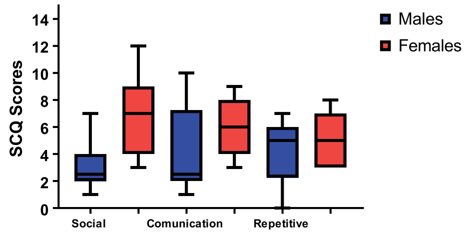

Fig. 1 SCQ subdomain scores divided by males and females

Social Responsiveness Scale

The SRS scores from 28 patients were analysed. A total of 71% of the scores were in the severe range, and 25% were in the mild to moderate range. Only 4% were in the normal range. Total scores and all subscales were higher in females on both standardised and raw scores. The gender difference was significant only in the subscales of Social Awareness and Social Cognition. The total T score and the raw score of Social Awareness and Social Cognition also had between large and very large effect sizes on the differences between males and females. A total of 87% of the females' and 54% of the males' scores were within the severe range, 13% of the females' and 38% of the males' scores fell in the mild to moderate range and 8% of the males' scores was in the normal range. All the SRS scores are summarised in Table 5.

Vineland Adaptive Behavior Scale II

The VABS II scores from 24 patients were analysed. All the VABS II scores are summarised in Table 6. There was a difference in the adaptive behaviour composite score, between males and females, but the differences were not significant.

Table 3 ID grade and SCQ-cutoff crosstabulation males

			SCQ-cutoff		Total
			≤ 15	≥ 15	
ID grade	No ID	Count	3	0	3
		% within ID grade	100.0	0.0	100.0
	Mild	Count	2	2	4
		% within ID grade	50.0	50.0	100.0
	Moderate	Count	4	1	5
		% within ID grade	80.0	20.0	100.0
Total		Count	9	3	12
		% within ID grade	75.0	25.0	100.0

Table 4 ID grade and SCQ-cutoff crosstabulation females

			SCQ-cutoff		Total
			≤ 15	≥ 15	
ID grade	No ID	Count	1	2	3
		% within ID grade	33.3	66.7	100.0
	Mild	Count	0	1	1
		% within ID grade	0.0	100.0	100.0
	Moderate	Count	3	7	10
		% within ID grade	30.0	70.0	100.0
	Severe	Count	0	1	1
		% within ID grade	0.0	100.0	100.0
Total		Count	4	11	15
		% within ID grade	26.7	73.3	100.0

Developmental Behaviour Checklist

The DBC scores from 27 patients were analysed and all scores were above the clinical cutoff (≥ 46). All the DBC scores are summarised in Table 7. The DBC did not have the same gender differences that we observed in the SRS and SCQ, except from the subscale Social Relating, where we found a strong tendency for more problems among the females (Cohen's d 0.85).

Effect of gender when controlling for developmental level and behavioural problems

To determine the impact of gender on the SCQ score when controlling for developmental level (VABS II standard score) and amount of emotional and behavioural problems (DBC total score), a linear regression was conducted with the total SCQ score as the dependent variable. Measuring IQ in individuals with SMS is known to be problematic due to their behavioural characteristics. Therefore, we use data from the VABS II as a proxy for developmental level.

When gender, VABS II and DBC were entered as covariates, we obtained a highly significant model of the SCQ score ($R^2 = 0.60$, $F = 8.8$, $p = 0.0008$). Only gender had an independent contribution on the model ($\beta = 0.70$, $p = 0.0003$); VABS II ($\beta = -0.13$, $p = 0.44$) and DBC ($\beta = -0.16$, $p = 0.31$) had no independent contribution.

A similar linear regression was conducted with the SRS total raw score. When gender, VABS II and DBC were entered as covariates, we still obtained a significant model of the SRS total raw score ($R^2 = 0.46$, $F = 5.1$, $p = 0.010$). Both gender ($\beta = 0.46$, $p = 0.022$) and DBC ($\beta = 0.48$, $p = 0.013$) contributed to the model. VABS II ($\beta = 0.04$, $p = 0.836$) had no independent contribution. More details from the models are displayed in Table 8.

Discussion

This study explored a number of ASD symptoms across gender in a Scandinavian SMS sample. The approximately three females per male above the SCQ cutoff is exactly the opposite of what we would expect to find in a sample of idiopathic ASD. It is particularly in the social domain of ASD that females with SMS differ substantially from females with other aetiological pathways to ASD.

The reversed gender ratio of ASD symptoms identified in this study cannot be explained by differences in neither developmental level nor in the amount of emotional and behavioural problems. The clinical diagnoses of intellectual disability differ between the genders, and we found a tendency for poorer development in females (VABS II total 53) than males (VABS II total 62), but this difference was not significant. In the regression model, the VABS II score did not have an independent contribution to the SCQ score. Emotional and behavioural problems, as measured with the DBC, did not differ between the sexes. In the regression model of the SRS, we

Table 5 Social Responsiveness Scale scores

	Total ($n = 28$)	Females ($N = 15$)	Males ($N = 13$)	Significant p value (Cohen's d)
Total T score (SD)	82.29 (12.63)	89.73 (9.88)	73.69 (9.77)	0.000 (1.63)
Social Awareness raw score (SD)	12.43 (2.73)	13.60 (2.53)	11.08 (2.36)	0.011 (1.03)
Social Cognition raw score (SD)	11.68 (5.36)	19.47 (5.00)	13.46 (3.82)	0.001 (1.35)
Social Communication raw score (SD)	27.93 (7.70)	29.60 (6.72)	26.00 (8.56)	0.233 (0.47)
Social Motivation raw score (SD)	13.07 (5.26)	14.47 (4.91)	11.46 (5.36)	0.137 (0.58)
Autistic Mannerisms raw score (SD)	21.14 (5.97)	21.73 (5.99)	20.46 (6.12)	0.585 (0.21)
Total raw score	91.32 (20.60)	98.87(17.65)	82.62(20.93)	0.038 (0.84)
SRS classification	N(%)[ratio]	N(%)	N(%)	
Normal (>60)	1(4)[0]	0	1(8)	[a]
Mild–moderate (60–75)	7(25)[0.35]	2(13)	5(38)	[a]
Severe (< 75)	20(71)[1.61]	13(87)	7(54)	0.096

[a] Not applicable due to small sample

Table 6 Vineland Adaptive Behavior Scale II scores

	Total (*N* = 24[a])	Females (*N* = 13)	Males (*N* = 11)	Significant *p* value (Cohen's *d*)
VABS II standard score (SD)	56.88 (12.86)	52.85 (12.69)	61.64 (11.89)	0.094 (0.71)
Communication (SD)	57.92 (14.12)	54.38 (13.25)	52.09 (14.59)	0.193 (0.55)
Daily activities (SD)	61.79 (12.79)	62.69 (12.44)	60.73 (13.71)	0.719 (0.15)
Socialisation (SD)	62.54 (10.36)	59.46 (9.03)	66.18 (11.05)	0.123 (0.67)

[a]Four parents were not available for telephone interview

found that DBC contributed in addition to gender. This probably indicates that the SRS is more sensitive to behavioural problems than the SCQ is [32, 45]. The SRS places a heavier emphasis on the reciprocal social interaction trait in ASD, whereas the SCQ places a similar emphasis on all three ASD domains [45].

Neither Oliver [16] nor Vignoli [17] found any significant gender differences in ASD symptomatology in other rare genetic syndromes such as cri du chat syndrome, Cornelia de Lange syndrome, Prader Willis syndrome or tuberous sclerosis complex.

We wanted to investigate whether a difference in ASD symptomatology could be the result of females having more severe phenotypes than males and if it could be linked to levels of ID or whether the emotional and behaviour problems in SMS affected gender differences. We found a strong tendency for lower degrees of ID in females than in males, but the difference was not significant. But as the difference is approaching significance (0.07), it would be interesting to investigate further if there could be a real gender difference in ID levels in SMS. As mentioned before, the accuracy of our ID levels is questionable and therefore not used to draw any conclusions. In general, administering formal psychometric assessments is often reported to be very difficult with people with SMS, due to the maladaptive behaviours, sleep disturbance and difficulties in expressive language skills [46].

The observed gender differences in ASD do not seem to be related to the main genetic mechanisms for SMS. The RAI1 mutations, associated with less severe SMS phenotype, were more frequent in females (3/20%) with more ASD symptoms than in males (0/0%) who had less ASD symptoms. The group of individuals with RAI1 mutations was too small to be tested as a separate subgroup in any of the analyses.

Current research suggest that female protective factors are more important than particular male-linked risk in explaining the male bias in ASD, but the mechanisms behind such female protection are not established [4, 6]. Whatever the female protective factor turns out to be, the current data suggest that it is not present in females with SMS.

We found three other papers presenting gender differences in SMS [19, 24, 30]. In the study from Edelman et al. [30], the authors found some gender differences, with the females showing more problems. Most of them were somatic (myopia, cold hands and feet, eating/appetite problems and possible hypersensitivity (problems finding shoes to fit)), but they also found that females had a significantly higher frustration with communication level. Neither of the questionnaires used in our study found a significant gender difference regarding communication, but a more thorough investigation of communication profiles in this syndrome would be beneficial both to investigate the gender difference more and to propose possible interventions. The study by Laje et al. [19] indicated an absence of the usual gender difference regarding ASD measured with SRS but not SCQ. In our study, we find gender differences both in the SRS and in the SCQ measure, both showing more problems among the females. It is particularly the social domain of ASD that has an unusual male/female ratio. Females with SMS have significantly more social problems than males. We did not find any difference in repetitive behaviour. Laje et al. [19] found a gender difference, favouring the females, in two subscales on the SRS but not in the total raw score or on the SCQ. In our study, we found a

Table 7 Developmental Behaviour Checklist scores

	Total (*N* = 27[a])	Females (*N* = 15)	Males (*N* = 12)	Significant *p* value (Cohen's *d*)
DBC total percentiles (SD)	84.44 (13.19)	83.73 (13.87)	85.33 (12.83)	0.759 (0.12)
Disruptive/antisocial percentiles (SD)	85.63 (16.24)	83.47 (18.45)	88.33 (13.26)	0.433 (0.30)
Self-absorbed percentiles (SD)	80.67 (12.47)	80.53 (11.89)	80.83 (13.68)	0.953 (0.02)
Communication disturbance percentiles (SD)	75.93 (20.75)	73.73 (20.76)	78.67 (21.33)	0.551 (0.23)
Anxiety percentiles (SD)	61.11 (28.28)	62.53 (29.15)	59.33 (28.31)	0.776 (0.11)
Social Relating percentiles (SD)	42.96 (24.82)	51.60 (25.28)	32.17 (20.33)	0.036 (0.85)

[a]One parent did not correctly fill out the questionnaire

Table 8 Regression model summary

	SCQ total			SRS raw score		
Factors	*B*	*p*	95%	*B*	*p*	95%
Constant	12.44	0.204	− 7.38/32.25	− 21.43	0.615	− 109.37/66.50
Gender	8.30	0.0003	4.34/12.24	20.95	0.022	3.42/38.48
VABS II	− 0.08	0.44	− 0.21/0.10	0.96	0.836	− 0.74/0.61
DBC	− 0.06	0.31	− 0.25/0.08	− 0.07	0.013	0.23/1.68
Model's R^2		0.60			0.46	
Model's *p* value		0.0008			0.010	

B unstandardized B, *Sig* significant level, *95%* confidence interval for B for each factor

gender difference in ASD symptomatology, but neither in our study nor in the study by Laje et al. [19] could this difference be explained by differences in other traits in the syndrome. IQ level, adaptive behaviour and general emotional and behaviour problems have been investigated. A more thorough investigation of gender differences in adaptive behaviour profiles and the emotional and behaviour problems would be beneficial, alongside further molecular research regarding possible sexually dimorphic processes in SMS.

Limitations

Assigning a formal diagnosis of ASD to individuals with a known genetic syndrome is a matter of some debate [15]. In the current study, we only used the SCQ and the SRS as a measure of the number of ASD symptoms; we did not observe or use diagnostic instruments such as ADI-R or ADOS. Hence, we do not have data on how many actually fulfil the criteria for an ASD diagnosis.

Measuring IQ in individuals with SMS, as mentioned earlier, is known to be problematic due to their behavioural characteristics. Therefore, data from the VABS II were used as a proxy for developmental level. Even though VABS II cannot substitute a formal psychometric assessment such as IQ tests, consistency has been demonstrated between formal IQ tests and the VABS II [41]. In this study, we used developmental level instead of intellectual level/disability in most of our analysis, due to the fact that we ourselves did not collect the ID levels and could not guarantee for their validity.

Conclusion

We found a clear reversed gender difference in the number of ASD symptoms in persons with SMS. This female bias in ASD symptoms is not explained by differences in the developmental level or the amount of emotional and behavioural problems. The deletion that is known to cause SMS is located on chromosome 17 (17p11.2), and there is no known reason to expect gender differences in any traits in this autosomal condition. The finding of a clear gender difference is therefore notable, and the mechanisms behind this require further study. A previous study found a sexually dimorphic phenotype in eating behaviour in mice with loss of RAI1 functions [24]. Whether this is related to our finding should be explored. Knowledge about the biological underpinnings of the reversed ASD gender ratio may be of relevance to understand gender differences in other biological pathways to ASD. The female protective factors assumed to explain the male bias in ASD seems to be lacking in SMS.

Abbreviations

ADI-R: Autistics Diagnostic Interview-Revised; ADOS: Autism Diagnostic Observation Schedule; ASD: Autism spectrum disorder; DBC: Developmental Behaviour Checklist; ID: Intellectual disability; RAI1: Retinoic acid induced 1; SCQ: Social Communication Questionnaire (SCQ); SMS: Smith-Magenis syndrome; SRS: Social Responsiveness Scale; VABSII: Vineland Adaptive Behavior Scale II

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Availability of data and materials

The datasets used and analysed during the current study are available from the corresponding author on reasonable request.

Authors' contributions

HEN, AN and TN planned and designed the study. HEN, BMA and AN collected the clinical information. HEN and TN analysed the data and interpreted the results. HEN wrote the first draft of the manuscript. All the authors contributed to the manuscript and read and approved the final manuscript.

Ethics approval and consent to participate

The study was approved by the regional ethics committee in Norway (REK 2015/1026). Written informed consent was given by parents/guardians to allow the researchers access to medical records from all hospitals in Norway.

Consent for publication

All the participants consented to publication.

Competing interests

The authors declare that they have no competing interests.

Publisher's Note

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Paper II

Age related changes in Behavioural and Emotional Problems in Smith-Magenis Syndrome measured with the Developmental Behavior Checklist (DBC)

ABSTRACT

Smith–Magenis syndrome (SMS) is a genetic syndrome most often caused by a deletion on chromosome 17 or more rarely by a mutation in the retinoic acid-induced 1 gene. The aim of this study was to investigate the Developmental Behavior Checklist profile (DBC) of persons with SMS and the associations between behavioural and emotional problems, age, gender, adaptive behaviour and autism symptomatology.

Twenty-eight persons with SMS were represented by their parents in this study.

DBC-Total scores are reduced with age, but they still show a mean that is clearly above the cut-off of 46. The differences between the age groups <9 years and 9-17 years ($p=0.024$) and between the age groups <9 years and >18 years ($p=0.007$) are significant.

We found a significant decrease in behavioural and emotional problems with age in SMS. We did not find a relationship between adapted behaviour and communication and behavioural and emotional problems.

KEYWORDS

Smith-Magenis syndrome, rare disorders, behaviour and emotional problems, adapted behaviour

INTRODUCTION

Smith–Magenis syndrome (SMS) is a rare, neurobehavioural disorder most often caused by a deletion of chromosome 17p11.2 (A. C Smith et al., 1986) or more rarely, haploinsufficiency

of the retinoic acid-induced 1 (RAI1) gene (Edelman et al., 2007; Slager, Newton, Vlangos, Finucane, & Elsea, 2003). The disorder is a multiple congenital anomalies and intellectual disability syndrome (Falco, Amabile, & Acquaviva, 2017).
More advanced genetic analysis and a more knowledge regarding syndrome, have the last years lead to more patients being diagnosed with SMS earlier than before (Gropman, Duncan, & Smith, 2006).

The unique neurobehavioural problems displayed by children and adults with SMS are challenging for both parents and professionals. The syndrome's severe behavioural problems include self-injury behaviours such as self-biting, slapping and skin-picking as well as prolonged outbursts, tantrums and aggressive behaviour (Dykens & Smith, 1998; Finucane, Dirrigl, & Simon, 2001). The challenging behaviours are believed to increase with age and intellectual level (Neira-Fresneda & Potocki, 2015). Sleep disturbances, present in 88% of the SMS patients, have been associated with an unusual inverted circadian melatonin rhythm (De Leersnyder et al., 2001; Gropman et al., 2006; Gropman, Elsea, Duncan, & Smith, 2007; A.C. Smith, Dykens, & Greenberg, 1998). The sleep disturbances include difficulty getting to sleep, frequent nocturnal awakenings, early sleep offset, and daytime sleepiness with a need for daytime naps (Neira-Fresneda & Potocki, 2015). Early expressive speech delays with and without hearing loss are observed, as are mild to severe cognitive deficits (Madduri et al., 2006; Martin, Wolters, & Smith, 2006; Udwin, Webber, & Horn, 2001). Autism spectrum disorders have also been identified in almost 90% of the investigated populations with SMS (Laje et al., 2010; Martin et al., 2006). A recent study found a reversed gender difference in autism symptomatology (Nag, Nordgren, Anderlid, & Naerland, 2018). This study only found a gender difference in the Social Relation sub-scale on the Developmental Behaviour Checklist (DBC) (Nag et al., 2018).

Behaviour and emotional problems (BEP), communication problems, autism spectrum disorder and sleep problems are aspects of SMS that have significant impacts on the lives of families. Hodapp et al. (1998) found that parents of children with SMS reported greater pessimism and more parent- and family problems compared to families coping with Down syndrome. Other studies of caregivers for children with SMS show increased distress in the form of depression, anxiety, and sleep problems (Foster, Kozachek, Stern, & Elsea, 2010). Additional findings indicate that levels of stress among caregivers are related to the level of behaviour challenges displayed by the child with SMS (Fidler, Hodapp, & Dykens, 2000; Hodapp et al., 1998).

The prognosis of the SMS patient is closely linked to their BEP (Poisson et al., 2015), and there is a need for more research that explores relations between BEP and other characteristics.

The aim of this study was to investigate the DBC profile of persons with SMS and the associations between BEP, age and other characteristics such as, gender, communication, ASD and adapted behaviour.

METHOD

Participants

This study was part of a larger assessment study of SMS in Norway, Sweden and Denmark. The study was approved by the Ethics Committees in all three countries. Written consent was provided, either by the parents, for persons under sixteen years old, or by both the parents and the person with SMS for persons above sixteen years old.

The participants consisted of parents of persons with SMS. Twenty-eight persons with SMS met the inclusion criterion of a genetically confirmed diagnose. Their age was between 5 and 50 years. Eleven of the persons with SMS were adults (above 18 years old). The level intellectual disability (ID) was retrieved from medical or pedagogical charts. In two of the participants, the level of ID was missing. The demographics are displayed in Table 1.

Table 1.

Demographics

	Total	<9 years	9-17 years	>18 years
N	28	8	9	11
Mean Age	17.5	6.2	11.9	29.5
Range	5.2-50.5	5.2-8.3	9.5-14.9	18.6-50.5
Gender				
Male	12	3	3	6
Female	16	5	6	5
Genetics				
Deletion	25	7	7	11
Mutation	3	1	2	0
ID grade	26	7	8	11
No ID	6	1	4	1
Mild	5	1	2	2
Moderate	15	5	2	8

Measures

The Developmental Behaviour Checklist (DBC) (Einfeld & Tonge, 1992, 2002) is a questionnaire which is completed by parents or other primary carers or teachers; this questionnaire is used to report problems over a six-month period. It is an instrument for assessment of a broad range of behavioural and emotional problems of persons with developmental and intellectual disabilities (Dekker, Nunn, & Koot, 2002; Einfeld & Tonge, 1995). The DBC have a confirmed reliability and validity (Dekker et al., 2002; Einfeld &

Tonge, 1995). The DBC has been proved useful to use in research of prevalence of psychopathology in the population of persons with ID (Einfeld & Tonge, 1995).

The Vineland Adapted Behavior Scale II (VABS II) (Sparrow, Balla, & Cicchetti, 1984; Sparrow, Balla, & Cicchetti, 2005) is a semi-structured interview. It can be completed by parents or caregivers that is used to assess the everyday skills (adaptive behaviour) of children and adults from birth throughout life. The VABS II is a standardised and validated tool. Many studies have confirmed its reliability and validity, and it is a widely used tools, and one of few tools for assessing adaptive behaviours translated to Norwegians with Scandinavian norms (de Bildt, Kraijer, Sytema, & Minderaa, 2005; Sparrow et al., 2005). Other studies have found a strong evidence for applicability of VABS in the population with ID (de Bildt et al., 2005).

The Social Communication Questionnaire (SCQ) is a standardized screening tool for the evaluation of communication forms and social function in children or adults in order to exclude autism or an autism spectrum disorder (ASD) (Rutter, Bailey, & Lord, 2003). A review of different assessment of rating scales for ASD found the SCQ performing well on psychometric properties, and is useful as a screening instrument (Norris & Lecavalier, 2010).

The Social Responsiveness Scale (SRS) is a 65-item, quantitative parent-reported measure that assesses social impairment associated with autism spectrum disorders (Constantino J. N & Gruber, 2005). Studies have found this instrument valid and reliable for measuring autistic traits (Bölte, Poustka, & Constantino, 2008; Constantino et al., 2003). It has also deemed feasible for research studies of autism spectrum conditions (Constantino et al., 2003).

Data analysis

Statistical Package for the Social Sciences (SPSS) version 23 (IBM) was used for statistical analysis.

Pearson's r was calculated using bivariate correlation analysis. Significance levels of both 0.05 and 0.01 was used. To make sure basic assumptions for parametric tests were met, the data were checked for normality by visually inspect histogram, Q-Q plot and Shapiro-Wilk's W test. The data proved normally distributed and therefore parametric tests were used. The participants were divided into three age groups (< 9 years, 9-17 years and > 18 years) to investigate whether the BEP measured with the DBC changes with age. Social Science Statistics' online resources was used for calculating effect sizes (Cohen's D). The T-test was performed to investigate group differences.

Linear regression analyses were conducted with DBC Total as the dependent variable and with ID grade, gender, age, VABS (including the communication sub-scale), SRS and SCQ as covariates. Due to low sample size, we only included a maximum of 4 covariates for each model. P-P plots was used to check the normality of residuals. We report the standardised coefficients (β) because we have both dichotomous and continuous predictor variables. Due to modest sample size, we do not correct p-levels for multiple tests, but we report effect sizes for improved interpretation of the comparisons conducted.

RESULTS

Table 2 displays the participants' scores on VABS II, SCQ and SRS. This information will be used as background to investigate the DBC profiles as well as associations between the characteristics and BEP.

Table 2.

Characteristics of the participants

	N	Mean	Range	SD
VABS Standard Scores	24	55.17	21-78	14.56
VABS Communication	24	56.46	24-95	15.47
VABS ADL	24	60.50	33-85	13.85
VABS Socialization	24	60.46	38-81	10.45

SCQ Total Score	27	15.70	3-25	5.76
SCQ Reciprocal Social Interaction	27	5.15	1-12	2.69
SCQ Communication	27	5.07	1-10	2.66
SCQ Repetitive Behavior	27	4.52	0-8	2.26
SRS Total Score	28	83.21	55-102	13.12
SRS Social Awareness	28	73.71	52-98	11.18
SRS Social Cognition	28	76.43	50-113	13.80
SRS Social Communication	28	75.04	55-93	11.17
SRS Social Motivation	28	69.18	40-89	13.26
SRS Autistic Mannerisms	28	95.54	65-135	18.46

DBC

The DBC data from 28 participants were analysed. The results of the DBC-Total and sub-scales and are presented in Table 3. A total of 93% of scores were above the cut-off on the DBC-Total.

Table 3.

Developmental Behaviour Checklist Scores (Percentiles)

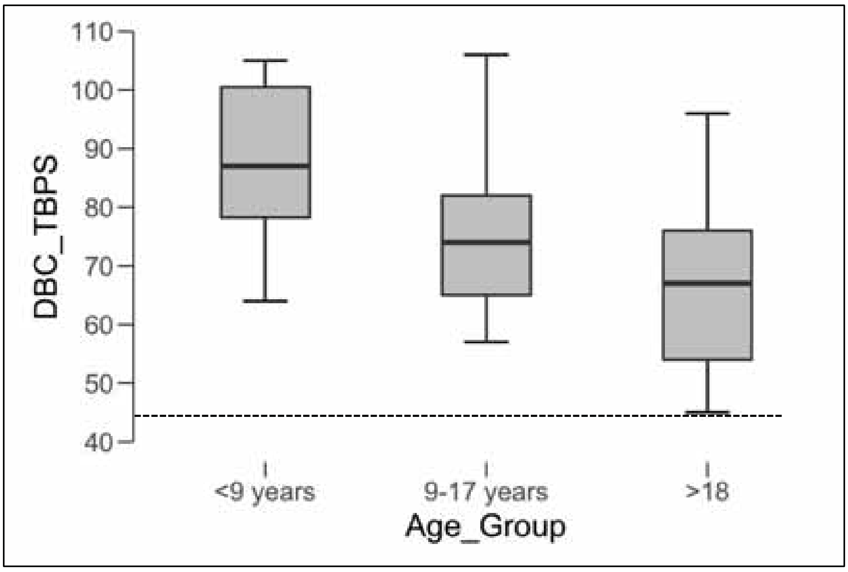
	Mean (SD)
DBC-Total	86.50(11.08)
Disruptive/Antisocial	87.57(12.81)
Self-Absorbed	81.50(12.82)
Communication Disturbance	77.64(17.31)
Anxiety	63.57(26.08)
Social Relating	45.14(25.85)

DBC: Relation to age

DBC-Total scores are reduced with age, but they still have a mean that is clearly above the cut-off of 46. The DBC Total percentiles had a significant negative correlation with age in the whole group (r=-0.430, p=0.022). The results of the age groups are presented in Figure 1. The difference between the age group <9 years and that of 9-17 years is significant (p=0.024), as is that between the age groups of <9 years and >18 years (p=0.007). There is no significant difference between the age groups of 9-17 years and >18 years.

Figure 1.

Developmental Behavior Checklist Total Score distributed across the age groups



Legend: The box indicates the 25th to 75th percentiles, the line in the box indicates the mean and the whiskers the minimum and maximum. The dotted line indicates the cut-off on the DBC.

The mean percentile profile in the different age groups are presented in Table 4. In general, both DBC Total and all subscales, except social relating are decreasing with age. In both DBC Total and sub-scale 1-4 the mean percentile in all age groups are relatively high (above 70th percentile. The only exception is in the Anxiety sub-scale where both age group 2 and 3 have a mean percentile in the 50th percentile. Social relating seems to be a relative strength in SMS with mean percentiles below the 50th percentile.

Table 4

Mean DBC Percentiles across age groups

	Age groups		
	1 <9yrs (N=8)	2 9-17yrs (N=9)	3 >18yrs (N=11)
DBC Total Behaviour Problem Score	93.75	87.33	80.55*/**
DBC Disruptive/Antisocial	94.25	92.00	79.09*
DBC Self-Absorbed	88.50	82.67	75.45*
DBC Communication Disturbance	84.25	74.44	75.45*
DBC Anxiety	82.75	59.56	52.91
DBC Social Relating	36.25	47.78	49.45

*Significant different from age group 1 at 0.05 level (2-tailed)
**Significant different from age group 2 at 0.05 level (2-tailed)

The mean, SD and significant group differences in each sub-scale are presented in Table 5. There are significant differences between the youngest and oldest age group in all but the Communication Disturbance and Social Relating sub-scales. Between the youngest and middle group there are no significant differences when looking at the p values, but there is a large effect in the Anxiety sub-scale using Cohen's D. Between the middle group and the oldest group, the only significant difference is in the Disruptive/Antisocial sub-scale. None of the other measures, such as VABS or SCQ had a similar decrease in scores with age SCQ had an increase in scores with age, but the difference was not significant (p=0.071, D=0.29).
In all sub-scales except, social relating, there is a decrease with age. In social relating there is an increase from age group 2 to 3. This decrease is not significant.

Table 5.

DBC across age groups

	Age groups			Group differences		
	1 <9yrs (N=8)	2 9-17yrs (N=9)	3 >18yrs (N=11)	1 vs 2 Cohens's D (p)	2vs3 Cohen's D (p)	1vs3 Cohen's D (p)
DBC Total Behaviour Problem Score (SD)	87.50 (14.92)	75.22 (15.89)	66.91 (16.87)	0.80 (0.123)	0.51 (0.275)	1.29 (0.014)
DBC Disruptive/Antisocial (SD)	31.13 (3.31)	30.44 (5.39)	23.36 (8.49)	0.15 (0.762)	1.00 (0.044)	1.21 (0.026)
DBC Self-Absorbed (SD)	30.63 (9.68)	24.78 (7.24)	20.91 (6.64)	0.68 (0.176)	0.56 (0.229)	1.17 (0.019)
DBC Communication Disturbance (SD)	9.88 (3.09)	8.22 (3.49)	8.55 (4.41)	0.50 (0.321)	0.08 (0.860)	0.35 (0.476)
DBC Anxiety (SD)	7.25 (2.66)	4.44 (3.13)	3.45 (1.81)	0.97 (0.067)	0.39 (0.387)	1.67 (0.002)
DBC Social Relating (SD)	2.25 (2.44)	3.22 (2.11)	3.91 (3.65)	0.43 (0.391)	0.23 (0.623)	0.53 (0.280)

Relation to adaptive level

The results are presented in Table 6.

Table 6.

Correlations with DBC

Correlations

		DBC Total Score percentile	DBC Subscale 1 percentile	DBC Subscale 2 percentile	DBC Subscale 3 percentile	DBC Subscale 4 percentile	DBC Subscale 5 percentile
VABS COMMUNICATION	Pearson Correlation	-,162	-,010	-,280	-,190	-,183	-,140
	Sig. (2-tailed)	,449	,962	,184	,374	,392	,516
VABS ADL	Pearson Correlation	-,405*	-,079	-,482*	-,428*	-,355	,012
	Sig. (2-tailed)	,049	,715	,017	,037	,089	,956

VABS SOCIALIZATION	Pearson Correlation	-,064	,111	-,138	-,118	-,277	-,139
	Sig. (2-tailed)	,766	,607	,519	,584	,191	,516
VABS Standard Scores	Pearson Correlation	-,134	,078	-,217	-,122	-,239	-,250
	Sig. (2-tailed)	,531	,717	,308	,571	,260	,239
SRS Total Score	Pearson Correlation	,375*	-,124	,398*	,325	,349	,577**
	Sig. (2-tailed)	,049	,529	,036	,092	,068	,001
SCQ Total Score	Pearson Correlation	-,180	-,483*	,020	,208	-,070	,473*
	Sig. (2-tailed)	,368	,011	,921	,298	,729	,013

*. Correlation is significant at the 0.05 level (2-tailed).
**. Correlation is significant at the 0.01 level (2-tailed).

Both the DBC total and the DBC sub-scales for disruptive/antisocial and communication disturbance were related to the VABS ADL sub-scale with a negative correlation.

SCQ Total had a negative correlation with the disruptive/antisocial sub-scale and a positive correlation with the social-relation sub-scale. SRS Total had a positive correlation with DBC Total score percentile and the sub-scales self-absorbed and social relating.

Several linear regressions were conducted with DBC Total as the dependent variable and with ID grade, gender, age, VABS (including the communication sub-scale), SRS and SCQ as covariates. When both gender and age were entered as covariates with VABS and SRS, we obtained a significant model (R^2= 0.64, F= 8.6, p= 0.00039). SRS (β=0.829, p= 0.00027), age (β= -0.479, p= 0.003) and gender (β= -0.671, p= 0.003) each had an independent contribution to the model. VABS (β=-0.244, p= 0.143) had no independent contribution. Removing gender from the model still yielded a significant model (R^2= 0.42, F= 4.88, p= 0.010). Both SRS (β=-0.42, p= 0.029) and age (β=-.43, p= 0.021) made independent contributions, but they dropped to the 0.05 level. Including only VABS and SRS did not give a significant model (R^2= 0.24, F= 3.35, p= 0.055). All three models are presented in Table 7.

Table 7.

Regression model summary

	DBC Total		DBC Total		DBC Total	
Factors	β	Sig	β	Sig.	β	Sig.
Constant		0.015		0.070		0.304
VABS	-0.225	0.143	-0.076	0.667	-0.052	0.793
SRS	0.829	0.00027	0.415	0.029	0.477	0.023
Age	-0.479	0.003	-0.43	0.021		
Gender	-0.671	0.003				
Model´s R^2	0.64		0.42		0.24	
Model´s p value	0.00039		0.010		0.055	

β =Standardised B, Sig.=significant level

DISCUSSION

The aim of this study was to investigate the relations between behavioural and emotional problems and other characteristics in SMS such as communication, adaptive behaviour and autism symptomatology.

We found significant age group differences in the DBC Total, with a decrease with age. This finding is the opposite of earlier research that reports an increase in behavioural problems in SMS with age (Finucane et al., 2001; Neira-Fresneda & Potocki, 2015; Poisson et al., 2015). This study included more adults with SMS than did earlier studies, but we still found a decrease in behavioural and emotional problems from age group 1 (<9 years) to age group 2 (9-17 years), even though the decrease was not significant in all sub-scales. Another study with 267 persons with either Williams syndrome (WS), Down syndrome (DS), Fragile X syndrome (FXS) or Prader-Willis syndrome (PWS) measured behavioural and emotional problems using the DBC two times, four years apart (Einfeld, Tonge, Turner, Parmenter, & Smith, 1999). Neither of the syndrome groups in this study had a significant change from Time 1 to Time 2. Einfeld et al (1999) found that persons with WS had the highest mean around 55 at Time 1, this decreased to less than 50 at Time 2. Persons with PWS had an increase from Time 1 to Time 2 from a mean around 52 to a mean around 57. The mean DBC Total in our sample was higher.

The decrease of behavioural and emotional problems with age is positive information for families that struggle with the behavioural and emotional problems of SMS. It is also important to underline that the levels of behavioural and emotional problems in adults with SMS are still high and far above the cut off on the DBC Total.

In this study as many as 93 % scored above the cut off on the DBC Total. The high percentage of persons scored above the cut off on the DBC Total was not surprising. Earlier research have shown that SMS often scores higher on self-injuries behaviour and aggressive behaviour than other comparable disorders (Arron, Oliver, Moss, Berg, & Burbidge, 2011). In the study by Arron et al. (2011), 92.9 % of the population with SMS displayed self-injuries behaviours and 73.8 displayed physical aggression. In earlier research concerning Cornelia de Lange syndrome and Down syndrome 38 and 31.3 % scored above the cut off (Basile, Villa, Selicorni, & Molteni, 2007; Bourke et al., 2008). in all age groups than any of the syndromes investigated by Einfeld et al(Einfeld et al., 1999). SMS is often compared to PWS, both due to both syndromes struggle with obesity, but also regarding challenging behaviours (Alaimo et al., 2015; Bouras, Dykens, & Smith, 1998). In a study utilising DBC to investigate the behavioural phenotype of PWS, Holland et al (2003) reported both DBC Total scores and scores on all sub scales lower than what we found in any of the three age groups in our study.

Investigating relations between the behavioural and emotional problems and other characteristics in SMS is an important step towards finding treatments that regulate the challenging behaviours that characterizes SMS. In this study, we investigated a range of characteristics as they related to SMS, but few of them had a significant relation to the behavioural and emotional problems measured by the DBC.

Both communication and level of intellectual disability have been associated with behavioural problems (McClintock, Hall, & Oliver, 2003). A large meta-analysis have found a significant association between low language abilities and problem behaviour (Chow & Wehby, 2018). In this study, we did not measure intellectual level -only reported previously measured ID grade, but we did measure adapted behaviour. Neither adaptive behaviour nor communication had a relation with the DBC in this study. A more specific communication and language assessment is needed to investigate if there is an association between communication issues and challenging behaviour in SMS.

The strongest correlation we found in this study was between SRS and sub-scale 5 (social relation), which is not surprising. The most interesting correlation, and the one most worthy of investigating further, might be the correlation between the DBC total and several sub-scales and the VABS ADL sub-scale.

All participants in our study have known challenging disorders, even though not all of them have a clinically diagnose of emotional and behavioural disorders in addition to SMS. They have all been in contact with resource centres for rare disorders in Sweden and Norway and their challenging behaviour are followed by either their regional or local professionals. Knowledge regarding how the handle challenging behaviours in SMS are scarce (Poisson et al., 2015) and more research are needed, in addition a continuous effort to spread information to the local communities and professionals close to the families.

In this study, we did not perform a thorough investigation of either intellectual level or communication, and that information is needed to further investigate if there is a relation between behavioural problems and ID or communication.

CONCLUSIONS

We found a significant decrease in behavioural and emotional problems with age in SMS. We did not find a relationship between, on the one hand, adapted behaviour and communication and, on the other hand, behavioural and emotional problems.

LIST OF ABBREVIATIONS

SMS	Smith-Magenis syndrome
RAI1	Retinoic acid induced 1
BEP	Behavioural and emotional problems
ADL	All Day Activities
ID	Intellectual disability
SCQ	Social Communication Questionnaire (SCQ)
SRS	Social Responsiveness Scale
DBC	Developmental Behavior Checklist
VABS	Vineland Adaptive Behavior Scale

DECLARATIONS

Ethics approval and consent to participate:

This study was approved by the regional ethics committees in Norway, Sweden and Denmark.

Consent for publication

All the participants consented to publication.

Availability of data and materials

The datasets used and analysed during the current study are available from the corresponding author on reasonable request.

Competing interests

The authors declare that they have no competing interests.

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Paper III

Article

Parental experiences with behavioural problems in Smith–Magenis syndrome: The need for syndrome-specific competence

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Abstract

The experience of having a rare disorder was summarised in a large study as ‘falling outside the vast field of knowledge of the professionals’. Parents (31 mothers and 17 fathers) of 32 persons with Smith–Magenis syndrome (SMS) participated in this study. A phenomenological approach was used to analyse the data into topics and themes. Four themes emerged: behavioural challenges displayed, parents’ strategies for meeting the challenging behaviours, parents’ experiences of their own competence and parents’ experiences of professionals’ competence and understanding regarding children with SMS and their behaviour challenges. We found that parents of children with SMS experience that they are exposed to severe challenging behaviours from their child. The parents believe that they experience more misunderstandings with professionals and that the challenging behaviours increase because there are some specific characteristics of SMS that professionals are not aware of or do not consider in their support services.

Keywords

Smith–Magenis syndrome, challenging behaviours, parents’ experiences, rare disorders, support

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Background

The experience of having a rare disorder was summarized in a large study by Grut et al. (2008b) as 'falling outside the vast field of knowledge of the professionals'. Because professionals rarely possess knowledge of rare disorders, they do not know how to treat and follow-up the person with the diagnosis and their family (Grut et al., 2008a). Trulsson and Klingberg (2003) found in their qualitative study that professionals' knowledge about the specific rare disorders is very important for these families. A study in Sweden concluded that parents of children with rare disorders experienced more stress, especially related to their own lack of competence, social isolation and emotional demands, and that they were less satisfied with the services provided by habilitation centres compared to parents of children with more common disabilities (Dellve et al., 2006). Dellve et al. (2006) also found that parents of children with behaviour challenges reported a higher level of stress than parents of children without behaviour challenges.

Smith–Magenis syndrome (SMS) is a rare, complex genetic syndrome caused by an interstitial deletion of chromosome 17p11.2, (Smith et al., 1986) or a mutation on the retinoic acid induced 1 (RAI1) gene (Slager et al., 2003). The disorder is characterized by intellectual disability, multiple congenital anomalies, obesity, neurobehavioural abnormalities and a disrupted circadian sleep–wake pattern (Chen et al., 2015; Poisson et al., 2015). The incidence of SMS is estimated to be 1:15,000–1:25,000 births (Dubourg et al., 2014; Greenberg et al., 1991; Huang et al., 2016). Delayed diagnosis is common, although more widespread recognition of the syndrome and introduction of new genetic technology in the last decade have led to earlier diagnosis (Gropman et al., 2006).

The majority of children and adults with SMS have behavioural problems, including self-injury, tantrums and stereotypies (Gropman et al., 2007; Poisson et al., 2015). Sleep disturbances, such as nocturnal awakenings and daytime sleepiness, are present in 88% of SMS patients and are partly attributed to an inversion of the circadian rhythm of melatonin distribution (De Leersnyder et al., 2001; Poisson et al., 2015). Cognitive impairment ranges from mild to severe, and expressive language delay with or without hearing loss is observed (Madduri et al., 2006; Martin et al., 2006; Osório et al., 2012; Udwin et al., 2001). Autism spectrum disorders (ASDs) have also been identified in almost 90% of the investigated populations with SMS (Laje et al., 2010; Nag et al., 2018; Osório et al., 2015). These unique neurobehavioural problems are challenging for both parents and professionals.

Variable levels of cognitive impairment have been documented through a variety of psychometric tests that assess adaptive behaviour, intelligence quotient and speech and language development (Madduri et al., 2006; Osório et al., 2012). Both behaviour problems and the need for assistance in many situations last into adulthood (Udwin et al., 2001).

Challenging behaviours, communication problems, ASDs and sleep problems are aspects of this disorder that have a great impact on these families' lives. Hodapp et al. (1998) found that parents of children with SMS reported greater pessimism and parent and family problems compared to families coping with Down syndrome. Other studies about caregivers for children with SMS showed an increased amount of distress in terms of depression, anxiety and sleep problems (Foster et al., 2010). There are also findings that support that the level of stress in caregivers is related to the level of behaviour challenges the child with SMS displays (Fidler et al., 2000; Hodapp et al., 1998). These three studies are all primarily quantitative and use standardized questionnaires. To our knowledge, no other qualitative research regarding parents of children with SMS' own experiences of handling the behavioural problems has been published. The topic is complex. The possibility to ask open-ended questions may provide answers and insight not found using standardized questionnaires.

The main aim of this study was divided into three areas: (a) explore the parents' experience of the behavioural problems of SMS, (b) how the parents viewed their own competency and their experiences in handling the behaviour problems their children with SMS display and (c) the type of support the parents obtained for the behavioural problems. We also looked for differences in reports from parents of females with SMS and males with SMS and differences by age.

Methods

Design

This is a qualitative study utilizing a phenomenological approach to investigate the parents' lived experiences (Creswell, 2013). The researchers decided to use written responses to open-ended questions and in-depth interviews to supplement the written responses. In phenomenological research, significant statements are highlighted, and these statements are divided into themes organized by the description of the parents' experience of the phenomenon (Creswell, 2013). After descriptions and themes have been obtained, the researcher may revisit the participants again to clarify or validate the data (Creswell, 2013). An important step in phenomenological studies is to set aside your own personal experience by writing it down and bracketing it (Creswell, 2013). Bracketing means that the researcher identifies personal experiences (beliefs, feelings and perceptions) with the phenomenon and sets it aside to be able to focus on the experiences of the participants (Creswell, 2013). Edmund Husserl, the founder of phenomenology, called this *epoché* and developed it as a method to be better able to describe the phenomenon exactly as experienced (Husserl, 1954 [1939]). In this study, the researchers' own experiences and preconceptions were written down at the beginning of the project. This was then revisited during the analysis process to ensure that the preconceptions had not influenced the results in a substantial way.

Recruitment and participants

This study was part of a larger study of SMS in Norway, Sweden and Denmark. The participants were recruited through Frambu Resource Centre for Rare Disorders (Frambu) and the Smith–Magenis Foundations in Norway, Sweden and Denmark (family support groups). The organizations disseminated information about the study on their Facebook pages and email lists. Frambu, which is one of nine state-financed centres of expertise administered by the Norwegian National Advisory Unit on Rare Disorders, has its own register, which is based on informed consent. Frambu could therefore send invitations to registered families with a child or an adult with a diagnosis of SMS. Swedish and Danish families were recruited through family support groups, both through information on their Facebook pages and through information given at their annual gatherings. The only inclusion criterion was that they had a son or a daughter with a genetically confirmed diagnosis of SMS, with no age limit. The parents consented to participate in the study. The parents who participated in the oral interviews were recruited through the Norwegian foundation's Facebook page, where they responded to a request to participate in an interview.

The study was approved by the Norwegian Regional Committees for Medical and Health Research Ethics (REC # 2015/1026).

A purposive sample of parents of 32 persons with SMS between 1½ years and 50 years participated in this study. Sixteen of the questionnaires were answered in writing by both parents, 15 of the 32 questionnaires only the mother answered, and 1 only the father answered. All the parents answered in writing. All persons with SMS had a genetically confirmed diagnosis of SMS. Four of the persons had RAI1 mutation, the other 28 individuals had a deletion. Of the 32 persons with SMS, 14 were males and 18 females. Thirteen of them were above the age of 18 at the time of the study. Seventeen came from Norway, 11 came from Sweden, and 4 came from Denmark. A convenience sample of four mothers participated in oral interviews were all from Norway, and their children ranged in age from 10 years to 22 years.

Methodological approach

Open-ended questions were provided to the participants to answer in writing through a questionnaire. The questions were stated in their native language (Norwegian, Swedish or Danish), and the participants provided written answers in their native language. All of the authors understand and can read all the three Scandinavian languages in addition to English. The questions were formulated on the basis of a literature review and Frambu's experience with the population. These are the questionnaire questions (translated into English by the authors):

- Does your child have behaviour challenges? If yes, please describe:
- What do you think is the cause of the challenging behaviours?
- How do you or did you feel about your own competence handling your child's challenging behaviours?
- What type of support and/or guidance did you receive for handling your child's challenging behaviours?
- What type of support and/or guidance would you have preferred for handling your child's challenging behaviours?
- What types of interventions have been tried for your child's challenging behaviours, and how did they work out?

After an analysis of the written answers from the parents, one particular theme emerged that required more investigation. Therefore, additional oral interviews were completed with four parents. These parents were already part of the study and had provided written answers to the questionnaire. The interviews were audiotaped. These are the questions asked in the oral interviews (translated to English by the authors):

- Professionals' lack of knowledge about the disorder was one of the themes that emerged from data collection. Do you have experience with professionals' lack of knowledge of the disorder, and if so, did the professionals' lack of knowledge about the disorder impact the guidance and follow-up you received?
- Do you believe lack of knowledge of SMS in the support system and among professionals have led to challenges for you or your child?

Data analysis

The written data were transferred from handwriting to a computer by one of the researchers and then analysed word for word. The audiotaped interviews were transcribed by the same researcher and analysed word-by-word. All the data were analysed using the phenomenological method described by Creswell (2013). The following stages were used in the data analysis: becoming familiar with the data by reading it repeatedly, developing a list of significant statements and regrouping the significant statements into themes (Creswell, 2013). After the regrouping of

Table 1. Topics, themes and categories.

Behaviours	Behavioural challenges displayed by the persons with SMS	Self-injurious behaviours Aggressive behaviours Inappropriate behaviours Controlling behaviours
	Parents' strategies for meeting the challenging behaviours of their children with SMS	Adaptation Adaptation to an extreme Just handling it
Competencies	Parents' experiences of their own competence with their child with SMS and their behaviour challenges	Lack of understanding and knowledge Not getting any assistance Learning by doing
	Parents' experiences of professionals' competence and understanding regarding children with SMS and their behaviour challenges	Professionals' lack of experience and knowledge The parents as educators Professionals' lack of understanding of the syndrome and the impact on the families

SMS: Smith–Magenis syndrome.

statements into themes, four parents were revisited in an interview to clarify, validate and broaden the findings. At the end, both the written data and the interviews were analysed as a whole. The statements were also visually analysed based on demographic data to look for differences by gender or age. The citations were translated to English by the authors.

Results

Two major topics with four themes emerged from analysing the data. Topics with themes and categories are presented in Table 1. Both the written responses and the oral interviews were analysed as a whole. When theme 4 emerged (parents' experiences of professionals' competence and understanding regarding children with SMS and their behaviour challenges), a need for clarification and broadening became necessary. Therefore, four oral interviews were included in the study to supplement the written responses.

Theme 1: Behavioural challenges displayed by the persons with SMS

Parents described self-injurious behaviours, such as tearing off nails, pinching themselves, hitting themselves and biting their hands. Behaviours such as banging their head into the walls or the bed were also reported. With regard to aggressive behaviours, we found descriptions of screaming, throwing items, destroying items, rage, hitting people around them, kicking and other types of physical attacks on siblings and other family members. As for inappropriate behaviours, we found behaviours such as bad or offensive language, refusing to do as told, undressing, self-stimulation and rapid mood swings.

> ... bothering siblings and doing things she knows is not ok. We also categorize it as challenging behaviour when she talks to strangers and repeatedly asks questions.

The parents also described behaviours such as constantly doing the opposite of what was requested, protesting, arguing and seeking conflicts. Impulsive and repetitive behaviours were also reported. With respect to controlling behaviours, we found behaviours such as controlling what parents should say, which words to use.

> He needs to be the one who decides. To be in control.

The parents also described situations when the person with SMS behaved differently with their parents and with the professionals.

> . . . it has been hard because they do not understand why he behaves like this towards us because he doesn't do that towards them?

It seems to be a clear gender bias in the behaviour reported by the parents. Parents of females reported more challenging behaviours than parents of males. There are no apparent differences in the type of behaviour reported between parents of males or females.

There are also more challenging behaviours reported by parents of children under the age of 18 than by parents of adults.

Theme 2: Parents' strategies for meeting the challenging behaviours of their children with SMS

Parents described common adaptations such as being proactive, providing predictability and aiming at prevention.

> We adapt everything around him to avoid anxiety, uncertainty, irritations, disappointment, etc. That makes things better.

Always planning for predictability became important for avoiding situations that might trigger challenging behaviours. Parents described more far-reaching adaptations, such as fixing hooks on all the doors in the house, parents sleeping outside the bedroom door of their child for years and installing physical tools to protect family members, such as restraints in the car. Some families rarely did activities together but divided to avoid triggers. One couple reported that they sometimes play-acted that they were dead to try to stop their child's behaviour.

> I have learned not to react to everything; today, we live a weird life where I move and act in a certain way not to set of my child

Some parents talked about just handling the challenging behaviours of their child with SMS.

> When we try to 'ignore' the aggressive outburst and just shield others or objects around, the outburst subsides faster.

The parents reported that handling and enduring the different situations became harder over time. They reported that this increased difficulty was partly due to the behaviours evolving into being more challenging as their child grew bigger and partly due to their own tiredness and lack of sleep over many years. They also expressed that it was easier to handle the behaviours at home than outside their home, especially as their children became stronger and more force was needed to handle them. Some parents explained that they had to adhere to strict rules, never breaking them to avoid challenging behaviours. They told of years of yelling, feeling guilty, tears and despair and that handling the behaviours demanded that you always were 100% present. Some of them reported that lack of energy reduced their ability to handle the challenging behaviours. When they did have enough energy, they handled the behaviours, and they were able to receive guidance and help. Some parents characterized their situation as lifelong parenthood.

> . . . we have tried everything, begged her to stop, cried in despair, hold her, not hold her . . .

There was also a gender difference in this theme, with parents of females with SMS reporting the need for more adaptations and more examples of just handling the challenging behaviours. No age differences were found in this theme.

Theme 3: Parents' experiences of their own competence with their child with SMS and their behaviour challenges

The parents described their own lack of understanding and knowledge about the syndrome and their handling of the challenging behaviours. They described that anger, frustration and sorrow were caused by the lack of knowledge and that they did not have the strategies or the tools to deal with these behaviours. Some parents said that many of the symptoms were easier to understand when their child was diagnosed with SMS.

> It would have been good to have the correct diagnosis earlier to adapt in a better way . . . . if we had known that the sleep issues were because of SMS, we would have acted differently. Now we thought it was because of us . . . that we didn't act firmly enough (when he was little). We didn't understand the reason for the challenging behaviours.

Regarding not getting any assistance, the parents described situations when they did not get any support in handling the challenging behaviours. They asked for assistance from the municipality but did not receive any. They emphasized that they needed guidance with strategies and solutions.

> We would have taken all the courses and classes if they had been offered.
>
> Nothing has been offered from the municipality, only by our own connections and by paying for it ourselves.

One family reported good guidance regarding what triggered the aggressive behaviours. The parents informed about how they developed strategies and knowledge themselves, learning by doing.

> We have learned different ways to handle the outbursts but have to renew ourselves all the time.

Some told about learning from other SMS families and the Internet.

A gender difference was found in parents reporting their own lack of competence and lack of support, with more reports from parents of females with SMS. No age differences were found in this theme.

Theme 4: Parents' experiences of professionals' competence and understanding regarding children with SMS and their behaviour challenges

Some parents reported that they had been offered support, but from professionals that didn't have the necessary specific knowledge of the syndrome, and therefore, the advice and recommendations didn't fit or work out. They told about how professionals reacted and acted considering the children's unusual sleep problems, variations in their developmental profiles and the fact that the person with SMS behaved distinctively differently at home towards their parents than outside the home towards professionals. The parents experienced inadequate advice telling them not to let their child sleep during daytime to make her or him sleep better at night; typically, they received this advice when their child was a toddler. They experienced a change when the kindergartens, schools or healthcare centres received information of the syndrome. One parent wrote that when the professionals were informed of the syndrome, the staff made changes leading to a decrease in this child's outbursts.

> I think it was because it was not specific to this diagnosis. I think it was that. Because I always experienced that it didn't fit (the advice).

On the other hand, some parents reported that they received qualified support from habilitation centres, pedagogical centres and kindergartens and schools.

Some parents expressed that professionals didn't consider the rare disorder but acted as if the child just had an intellectual disability or ASD.

> It is not enough to just look at the level of ID . . . you have to specify things about the syndrome also . . . I think.

Parents mentioned that the demands often were set too high. The professionals assumed that the children were more competent than they were, and they underestimated the amount of support that the person with SMS needed.

> . . . he cannot handle that independence, he doesn't have a chance. But they all think he does, and the school thinks he does . . . .

This last experience was particularly related to kindergartens and schools, and for some families, it was a successful approach, but it was not for others. Some parents said that schools are familiar with intellectual disability and ASDs and want the child with SMS to fit into one of these categories and that did not work very well. The parents also explained that they were responsible for providing information and training for professionals working with their child. Both professionals from the kindergartens and schools and from healthcare institutions relied on information from the parents, and the parents felt that it was their responsibility to see that their child received correct follow-up and treatment.

> It is we that have been educating the preschool, school, and health care centres.

Furthermore, the parents described situations when they felt that the professionals were of the opinion that the parents should have handled the situations differently. They told about professionals' lack of understanding and how the professionals' actions influenced how the person with SMS behaved at home with the family. They also mentioned what it meant to them always having to describe their child in negative words. Some parents expressed how the professionals' lack of understanding of the syndrome made them feel like an annoying whiner. Some parents, though, stated that it felt like someone was 'taking care of their soul', when the professionals showed that they understood.

> I don't think they understand how bad it is a home. I don't think they have a chance to understand it . . . . I had, to be honest, problems to understand it myself, when she was younger.

Neither any gender nor age differences were found regarding parents reports in this theme.

Discussion

The most important finding from this study is that the parents experienced that there are some specific characteristics of SMS that professionals are not aware of or do not consider in their support services. Parents' expressed that this lack of understanding exposed them to both more behavioural challenges from their child and more misunderstandings with the professionals. According to the parents, one of these characteristics is the uneven developmental profile. Persons with SMS often have developmental asynchrony with a discrepancy between intellectual development and emotional development in particular. This asynchrony means that they master skills at one level, but their emotional development and reactions are like a small child (Haas-Givler and Finucane, 2014). As one parent said:

> Because she looks very well-functioning, but she is not well-functioning at all, she needs a lot more support than you see at first glance.

This profile may be found in other disorders as well, such as Williams syndrome (Fu et al., 2015), where they often have better development of expressive language than other abilities. This developmental asynchrony is one issue that makes living with a person with SMS challenging (Haas-Givler and Finucane, 2014). As the cognitive skills and demands progress, the environment is not adapted to their young emotional style and reactions (Haas-Givler and Finucane, 2014). In addition, children and adults with SMS behave very differently with and without their parents. They often have challenging behaviours they only display at home (Poisson et al., 2015). Aggressive behaviours in SMS are usually directed towards close relatives such as parents (Poisson et al., 2015). These two characteristics, the uneven profile and the display of the more severe behaviours directed towards close relatives, amplify the parents' problems. These two characteristics are not necessarily noticeable when professionals meet the persons with SMS for the first time. Professionals need knowledge of the syndrome and the ability to listen to the parents to become aware of these characteristics and the impact they have on the families' lives.

Another important finding in this study is the extreme adaptations some of these families implement. Behaviours such as pretending to be dead to stop your child's outburst or sleeping outside your child's bedroom door for years are extreme and indicate to which length these parents are willing to go to take care of their child and themselves. Professional support and guidance in how to handle their child's behaviours at home will be valuable for these families. Extreme adaptations need to be seen in relation to many parents' reports that they didn't receive any help with the behavioural challenges. Some parents were also of the opinion that if they had obtained help, the help would have been given from unqualified or unknowledgeable professionals.

The challenging behaviours in SMS have been described earlier (Sloneem et al., 2011). In the open-ended questionnaire we used in this study, the parents were asked to name the challenging behaviours their child displayed instead of checking them off a list, which is a more common approach in research on behavioural challenges. Self-injury, aggressive behaviours and inappropriate behaviours are well known within the syndrome, even though inappropriate behaviours were more emphasized and more often mentioned by the parents than anticipated and described earlier. Controlling behaviours are not mentioned in the literature in any extensive way. Parents attending courses at Frambu have been talking about these behaviours, but in this study, it came across as a substantial challenge for the families. This type of behaviour is an important topic to explore further.

There have previously been reported a gender difference regarding SMS and ASDs (Nag et al., 2018). A visual inspection of the demographics behind the statements was done in this study, and there might be a gender difference also in other areas than ASD, such as the number of challenging behaviours reported. Parents of females with SMS reported more challenging behaviour, more adaptations and more of a lack of competence and support. Since this is a qualitative study, the results need to be interpreted carefully and further investigated.

A similar visual inspection of the demographics behind the statements was done regarding differences between children below the age of 18 and adults. A difference was found in the number of challenging behaviours reported with more challenging behaviours reported by parents of children under the age of 18. This is the opposite of what is reported earlier in research where the challenging behaviour is found to increase with age (Finucane et al., 2001; Neira-Fresneda and Potocki, 2015; Poisson et al., 2015). Our study included more adults than previous research and that may be one of the reasons for the different results, but this needs further research. This is a qualitative study, and the results need to be interpreted carefully.

Some of the results in this study are similar to other research on parenting a child with a rare disorder (Griffith et al., 2011; Strehle and Middlemiss, 2007). The professionals' lack of knowledge about the rare disorder was confirmed in Griffith et al.'s study from 2011 (Griffith et al., 2011). Research emphasizes that parents often need information about the main aspects of a specific syndrome and that specific characteristics of genetic syndromes influence the type of guidance the parents require (Pearson et al., 2018). Knowing about specific difficulties associated with a genetic syndrome may help with adapting to the environment in a different and more adequate way (Oliver et al., 2010). Specifically, having information about the link between the genetic disorder and the challenging behaviours seems to lead to a change in attitude among professionals from blaming the parents and the person with the diagnosis to increased optimism with regard to the possibility of change and increased willingness to help (Oliver et al., 2010).

Griffith et al. (2011) studied mothers' experiences of social care support and medical services for their adult offspring with rare syndromes and found that both the professionals and the mothers first categorized their offspring as a person with an intellectual disability and second with a rare genetic disorder. The parents in our study expressed that they found it difficult that professionals only defined their children as having an intellectual disability or ASD. They experienced that this definition led to an insufficient adaptation of measures and more behaviour challenges.

Research over recent years has focused on how the professionals' lack of knowledge put a strain on the person with the rare diagnosis and their parents (Haas-Givler and Finucane, 2014). However, with regard to SMS, knowledge is even more important. Some of the specific characteristics of this diagnosis have a substantial negative impact on the person with SMS and the whole family (Poisson et al., 2015). This impact is especially true if the support systems do not acknowledge these specific characteristics and seek advice and training to improve their counselling (Haas-Givler and Finucane, 2014).

In a study by Hodapp et al. (1998) comparing parents of children with SMS and Prader–Willi syndrome, they found a correlation between families' stress level and the size of their support group, both private and professional. In the SMS group, professionals were more often listed in the families' support group. Therefore, qualified professionals may be of greater importance to these parents than parents of children with other disorders. Other studies confirm that parents of children with SMS experience higher levels of family problems than parents of children with Down syndrome and Williams syndrome (Fidler et al., 2000) and parents of children with different aetiologies of intellectual disability (Hodapp et al., 1998). Sarimski (1997) found a correlation between parental stress and perceived social support. Our study also supports the findings that families having a family member with SMS are in great distress. Professionals, municipalities and other support systems need to be aware of these families and their overall needs for persistent and competent support.

Some of the results in this study agree with research on persons with disabilities, including research concerning children with behaviour challenges (Awijma et al., 1997; Griffith and Hastings, 2014; Richman et al., 2009). A high level of challenging behaviours have been correlated with elevated parental stress (Richman et al., 2009). A review from 2014 (Griffith and Hastings, 2014) confirms some of the findings from our study: the lack of support for challenging behaviours, the fact that the level of need exceeds the level of services and the lack of skilled support. The review also reported families who were satisfied with their received support, which we also found in our study. Future research should identify the families who are positive about the support they receive and identify the characteristics of both the families and the support so that different types of support systems may learn from it.

Limitations and strengths

One limitation in this study is that we chose to use open-ended questions in the questionnaire. Utilizing qualitative analysis of written responses has been questioned, both because of difficulties interpreting the written answers and the danger of asking leading questions. When asking respondents to write their answers down, you lose the ability to code and analyse body language or clarify orally (Ryen, 2002). It may also be an obstacle, both for the respondents and in the analysis, if the respondents are not comfortable with or have trouble expressing themselves in writing (Creswell, 2013; Ryen, 2002). We still chose this method because, to our knowledge, this study is the first qualitative study of parents of children with SMS focused on behaviours. It was important to be able to include parents from Norway, Sweden and Denmark, and we would have faced complications with both logistics and language if we chose face-to-face interviews. We assess using open-ended questions also as a strength because this opened up for statements and themes that have never before been considered in research on SMS. We added face-to-face interviews with four parents to strengthen and broaden our findings.

A limitation in phenomenological research is that the researchers' own preconception and experiences may influence the analysis process and the selection of citations.

Limitations in generalizability or transferability are often raised regarding qualitative research (Creswell, 2013; Malterud, 2001). The possibility to transfer the results to similar populations is often a goal in quantitative research, but this is not the main purpose in qualitative research. Some of our results may be unique to the Scandinavian population due to the similarity in the social systems they benefit from. These systems may be different in other countries. It is a strength to the transferability possibilities that we have participants from three different countries and that we have recruited approximately 50% of the known population in both Sweden and Norway.

Conclusions

In this study, we found that parents of children with SMS experience that they are exposed to both more behaviour challenges from their child and more misunderstandings with professionals and they believe that is because there are some specific characteristics of SMS that professionals are not aware of or do not consider in their support services. In particular, the uneven profile and the display of the more severe behaviours directed towards close relatives seems to amplify the parents' problems. We also found that these parents implement some extreme adaptations to meet the challenging behaviour that their children with SMS display, and there is an unmet need for qualified and knowledgeable support for these families. It is especially important that professionals working with persons with SMS or families where a person is diagnosed with SMS increase their knowledge about the disorder. According to the parents, the professionals need to make an effort to listen to the families regarding what type of support and help they need and try to base their actions on their views.

Authors' note

The study was approved by the Norwegian Regional Committees for Medical and Health Research Ethics (REC # 2015/1026).

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Paper IV

School Staff's Experiences and Coping related to the Challenging Behaviours of Children with Smith-Magenis Syndrome in Schools: A Q Methodological Study

Abstract

The aim of this study is to use Q methodology to explore how school staff experience the behaviours of children with Smith-Magenis Syndrome (SMS) in school and how they manage working with these children. Q methodology utilise by-person factor analysis to investigate subjectivity. Fourteen school staff of students with SMS in Norway participated and sorted 40 statements according to their own experience working with a student with SMS. Two distinct viewpoints were revealed, namely, 1) Managing challenging aggressive and self-injury behaviours in school where school staff experienced a range of challenging behaviours, especially aggressive behaviours. 2) Struggling with intense non-physical challenging behaviours in school where school staff experienced behaviours such as the students being very intense, craving attention and pushing buttons. In conclusion, there must be a greater emphasis on education and advising and supporting school staff's work with the non-physical challenging behaviours aspects of teaching children with SMS, as well as a continued focus on challenging aggressive behaviours.

Keywords: Smith-Magenis syndrome, challenging behaviours, school, school staff, Q-methodology, coping

Challenging behaviours are a complex concept and are used to describe several types of behaviours in kindergartens, schools, at home and in society in general. Challenging behaviours often have a negative impact on persons' learning performance and become a challenge for the learning environment (Roland, Øverland, & Byrkjedal-Sørby, 2016). Challenging behaviours includes behaviours that are self-injurious behaviours, aggression, stereotyped behaviours and destruction of property (McClintock, Hall, & Oliver, 2003). Persons with intellectual disability (ID) have a heightened risk of developing challenging behaviours (McIntyre, 2008). One of the disorders associated with ID and challenging behaviours is Smith-Magenis Syndrome (SMS) (A. C. Smith, Dykens, & Greenberg, 1998). Children with SMS has a complex behavioural profile that often prove challenging for school staff, and one of the most crucial elements of successful school environment is the student – teacher match (Haas-Givler & Finucane, 2014).

Children with SMS in School

SMS is a rare, complex genetic syndrome caused by an interstitial deletion of chromosome 17p11.2 (A. C Smith et al., 1986) or a mutation of the retinoic acid induced 1 (RAI1) gene (Slager, Newton, Vlangos, Finucane, & Elsea, 2003). The disorder is characterized by ID, multiple congenital anomalies, obesity, neurobehavioural abnormalities and a disrupted circadian sleep-wake pattern (Williams, Zies, Mullegama, Grotewiel, & Elsea, 2012). The incidence of SMS is estimated to be 1:15,000-1:25,000 births (Greenberg et al., 1991). In Norway, Frambu Resource Centre for Rare disorders have registered approximately 40 persons with SMS.

Children and adults with SMS appear to have unique neurobehavioural problems that are especially challenging for school staff. Many of the physical and medical symptoms of

SMS have a direct impact on educational functioning (Haas-Givler & Finucane, 2014). These problems include sleep disturbances, self-injurious and aggressive behaviours, stereotypes and sensory integration disorders (Martin, Wolters, & Smith, 2006; A. C Smith et al., 1986). Autism spectrum disorders (ASD) has also been identified in almost 90% of the investigated populations with SMS (Laje et al., 2010; Martin et al., 2006). A high level of social motivation, attention seeking behaviours and high levels of attachment to favourite people, along with an insatiable' need for individualised attention from adults are also reported (Wilde, Mitchell, & Oliver, 2016).

The aforementioned characteristics may present incidence on students' outcomes and wellbeing at school. Such characteristics make that both educational and behavioural interventions for students with SMS are extremely challenging to implement for both parents and professionals' (Neira-Fresneda & Potocki, 2015) and, moreover, they affect to teacher-child relationship in a negative way (Haas-Givler, 2004). Teacher-child relationships are critical for educating students (no matter their disability status) since they do have an influence on children's adaptation to school and their social and emotional development and academic success (Pianta & Stuhlman, 2004). The teacher-child relationship in children's early years has been found to be predictive of the behaviour issues (Hamre & Pianta, 2001). Teacher's beliefs are important to identify because they relate to their priorities and decision making in the classroom, which directly influences the children's well-being and education (Thorsen, 2009).

Bearing in mind the challenging behaviours present in students with SMS and that this influences their relationships with their teachers and teachers' expectations, there is a pressing need to investigate the current behaviours of students with SMS in schools and how the school staff perceive them and manage their work with these children. This is important for students' wellbeing and success. Notwithstanding this, if one dives into the published research, there is a lack of studies addressing this topic, with some studies using Q methodology regarding the perceptions of kindergarten teachers and schoolteachers (Subba, Bru, & Thorsen, 2017; Øverland, Thorsen, & Størksen, 2012). To fill the gap concerning SMS students and teachers' perceptions and coping strategies when teaching these students, the goal of this work was to explore the challenging behaviours that students with SMS display in school and explore how school staff experience and manage these behaviours.

Methods

Q methodology was developed and introduced by William Stephenson in 1935 (Stephenson, 1935). Q methodology is both a philosophy of science, a theoretical framework, a research technique for collecting data and an analysis method for scientific research of subjectivity (Brown & Good, 2010). There are commonly five steps used in Q studies (Van Exel & de Graaf, 2005): a) definition of concourse, b) developing the Q set, c) defining the participants, d) the Q sorting and analysis and e) interpretation.

Definition of Concourse

The concourse is defined as a collection of all possible statements of the subject in concern (Van Exel & de Graaf, 2005). The concourse could come from different sources, such as photos, music, interviews, conversations, social media, magazines or scientific papers (Brown, 1980). In this Q study open-ended questionnaire and standardized questionnaires (Developmental Behaviour Checklist and Vineland Adaptive Behavior Scales), completed by the parents of persons with SMS as part of a larger study, and published sources (Haas-Givler & Finucane, 2014; Neira-Fresneda & Potocki, 2015) were used to identify the concourse.

Developing the Q set

The Q set consist of the statements that are being sorted. Approximately 150 statements were collected. To create a balanced and structured set of statements, Fisher's balanced block design was used to structure and select representative statements from the concourse (Fisher, 1960; Stephenson, 1953). Fisher's balanced block design is a two-dimensional model with effect on side and levels on the other side (Fisher, 1960). A 2 x 2 block design with two main dimensions (extrovert versus introvert behaviours and behaviours occurring alone or together with other students) were used to ensure coverage of a wide range of statements. An additional statements category of 'school staff experiences' was added. Also, a 'various' category was added, including statements not fitting in any of the other categories such as 'the student is good at technical things' and 'the student can be experienced as very intense' (see Table 1).

The statements were reduced to 40 statements by grouping them in similar groups. From the statements that addressed the same issue, one statement was selected, or statements were combined. The statements were printed on separate cards and numbered arbitrarily, and the generated statements are known as the Q set (Coogan & Herrington, 2011; Van Exel & de Graaf, 2005).

[Table 1 near here]

Participants (P set)

The P set refers to the group of participants in the study. In this study the P set consist of school staff currently working with a student with SMS in grade school. Principals of 10 schools (we only know of approximately 10 students in grade school in Norway) were contacted by mail and were asked to distribute the Q sort to three of their staff members who work with students with SMS. Fourteen (47 %) participants completed the Q sort and returned them. The participants' information is indicated in Table 2. Three of the participants worked in special education schools, the rest (eleven) worked in regular education settings. Six participants were special education teachers, the rest did not have any special education training. Six of the participants had worked with the student with SMS for 1-2 years, five had worked with the student with SMS 4-5 years, and one had worked with the student for 7 years. Two participants did not indicate how long they had worked with the student with SMS. No one had worked with other students with SMS before.

[Table 2 near here]

Q Sorting

The participants were instructed to sort the Q set of statements into a grid from most like to most unlike my experiences regarding the students with SMS. In this study a distribution grid with 11 categories (from +5 to -5) was created to fit 40 statements cards (Fig. 1). Because the students with SMS are spread geographically all over Norway and face-to-face Q sorts would be very ineffective, the Q sorts were therefore sent in the mail. Studies have shown that Q sort sent in the mail or performed using a computer has no difference in reliability or validity than interview based (face-to-face) Q sorts (Reber, Kaufman, & Cropp, 2000; Van Tubergen & Olins, 1979). After the sorts, the participants were instructed to write down the numbers of the statements in the correct place in the grid. They were also instructed to provide a written rationale for the placement of the two statements placed on the far right (+5) and far left (-5) sides of the grid.

[Figure 1 near here]

Data Analysis and Interpretation

The completed Q sort was correlated with the other participants' Q sorts (Coogan & Herrington, 2011; McKeown & Thomas, 1988; Schmolck, 2002). All the Q sorts were plotted and analysed using one of the available computer programmes, the PQ Method Programme (Schmolck, 2002) that utilized a by-person factor analysis. In the analysis process the correlation of all Q sorts was calculated. Then, the degree, or level of dissimilarity and similarity of points between the individual sorters were calculated. After that, a by-person-factor analysis was performed to examine how many groupings of similar Q sorts there were. People with similar views (sorts) shared the same factor (Van Exel & de Graaf, 2005). In Q methodology, the statements are utilized to define a factor, a theoretical factor is constructed by a weighted average of the factor's score for the Q sort associated with this factor (Brown, 1980; Wheeler & Montgomery, 2009). Factor scores are essentially weighted z-scores for each statement in the Q set; these scores can be converted into an array of scores (factor array) that correspond to the plus 5 to minus 5 values in the original Q sort continuum (McKeown & Thomas, 1988). The factors were interpreted based on the characteristic statements of each factor, as well as distinguishing and consensus statements. In addition, written statements regarding why the participants placed the statements on either end of the scale were used to enlighten the results. The statements and factor scores are presented in Table 3.

[Table 3 near here]

Ethical Considerations

This study was part of a larger study regarding SMS. This study was approved by the Norwegian Ethical Committee (2015/1026). The participants signed an informed consent form.

Results

A Varimax rotation revealed two distinct viewpoints regarding school staff's experiences with challenging behaviours in students with SMS. Two factors or viewpoints were chosen based on both a visual inspection of the scree plot and the eigenvalues. The two viewpoints were labelled 1) Managing challenging aggressive and self-injury behaviours in school and 2) Struggling with intense non-physical challenging behaviours in school. These two factors extracted a 42 and 18 percent variance, respectively. Ten participants loaded on factor 1, and four participants loaded on factor 2 (Table 4). Five of the six special education teachers loaded on factor 1. The four participants loading on factor 2 had all different levels of education and work positions in the schools, but they all worked in regular education settings. There were no differences in which factor they loaded on depending on the gender of the student with SMS.

Table 5 provides an overview of the statements that have been ranked high or low on the two factors.

[Table 4 and 5 near here]

Consensus Statements

Consensus statements do not distinguish between any of the identified factors (Van Exel & de Graaf, 2005). In this study, there was consensus between the two factors on more than half of the statements (23 statements). The consensus statements are presented in Table 3. Most of the consensus statements were regarding how the school staff experienced the students' behaviours, not the school staff's coping and emotions regarding the behaviours. Regarding the staff's emotions and coping, they agreed that it is positive to work with these students (Table 3, statement 36, +5 and +4), and they know what to do when challenging behaviours are displayed (Table 3, statement 38, +4 and +3). In both factors, the staff think it is somewhat difficult to explain to other teachers about the student and how to handle their behaviours (Table 3, statement 40, -1 and -1). One participant explained why they placed statement 40 (Table 3), I think it is easy to explain to the other teachers about the student and how to handle situations, on the least like (-5) in a good way:

> *To get insight into the challenges regarding this student you must have known him for a while; therefore, it takes time before you experience the need of the adaptations. And, therefore, it is difficult to explain well some of the importance of following the rules and routines that we have with this student.*

Interpretation of factors

Factor 1: Managing challenging aggressive and self-injury behaviours in school. Ten school staff sorts defined the first factor. The main aspects of this factor are that the school staff experience a range of challenging behaviours, especially aggressive behaviour, as well as acting out behaviours, and that the staff are handling these behaviours and have a positive attitude towards both their work and the students. One of the participants described why they put statement 27 (Table 3), the student never gets angry with screaming, kicking and hitting, on least like (-5) this way:

> *When he gets angry, he can boycott, destroy for others, scream, yell, lay down on the ground, bite, spit, scratch, kick and hit. He is doing this towards himself, but mostly towards us adults and very rarely towards other students*.

The view represented by this factor was that the staff experience that the students get angry at school, they hit, scream, kick and have self-injurious behaviour, but the school staff still experienced it as positive and challenging in a good way to work with these students. One of the participants put the following statement, working with this student is challenging in a good way (Table 3, statement 30) on most like (+5) and explained the choice of the most positive statement as follows:

> *A lot of joy, laughter and care that he and his parents are sharing. Instructive and demanding in a good way.*

The staff who hold this view experience numerous acting out and challenging behaviours but are managing it and know what to do. The staff experience a balance of challenging behaviours but also see the positive traits in the student, such as humour, politeness, and social and caring behaviour. Another participant put statement 40 (Table 3), I think it is easy to explain to the other teachers about the student and how to handle situations, on the least like (-5) and explained it as follows:

> *The student is complex. It takes a long time to get to know him. It takes a long time before he shows his true self.*

The school staff who hold this view are not tired of their work (Table 3, statement 9, -4) although they are working with students who are very preoccupied with adults (Table 3, statement 31, -3 (negative stated)) and have a lack of impulse control (Table 3, statement 15, -3 (negative stated)).

Factor 2: Struggling with intense non-physical challenging behaviour. The second factor identified was defined by four the school staff (n=4). The main view of this factor was that the school staff experienced that the students are challenging to work with especially because of the intensity of their behaviour, and the staff are positive towards their work and the students. However, the staff experience demanding situations especially if they are alone with the students. The view of this factor was that the staff experienced that these students have more non-physical behaviour problems such as being very intense, craving attention and pushing buttons. One of the participants who holds this view explained their placement of statements 20 (Table 3), the student can be experienced as very intense, and 19 (Table 3), I have not experienced the student getting angry at school, in the most like (+5) this way:

> *I experience this student as very intense, talks all the time and picks on everything if he is allowed to.*

> *I have not experienced that the student gets angry. I have seen him upset and frustrated, but not angry.*

The staff holding this view do not experience the acting out behaviour, such as kicking, screaming and self-injurious behaviour, as problematic as the more non-physical behaviour. In this view, they were positive towards working with these students but also found it demanding and struggle with being alone with the student with SMS. In this view, the staff experienced the students as emotionally immature, having a lack of impulse control, lacking the ability to focus on school work over time and having trouble sitting quietly by themselves. Another participant described why choosing statement 14 (Table 3), I think it is demanding to be alone with the student, in the most like (+5) this way:

> *The student demands a lot of one person. You always need to be prepared for a mood swing. It is very important to have more than one person working with this student. You will need a break in between the battles.*

In this view, the participants also described that the students with SMS were more drawn towards adults than other students. As one participant said regarding why they chose statement 31 (Table 3), the student is not preoccupied with adults, for the least like (-5):

> *The student is very preoccupied with adults and contact with the adults. It often gets complicated with other children. They are more unpredictable.*

The staff who hold this view also agreed with the statements that they are sometimes tired of their job (Table 3, statement 9, +2) and that these students sometimes provoke them (Table 3, statement 23, +2).

Distinguishing Statements

A statement is distinguishing when a statement's score goes beyond the difference score. The difference score is the degree of difference between a statement's score on any two factor's that is required for it to be statistically significant (Van Exel & de Graaf, 2005). The distinguishing statements are presented in Table 3. How the staff handled the behaviours differs: The participants who held the view of factor 2 were more scared when the student lost control (Table 3, statement 39, -4 and 0), and the participants who held the view of factor 1 believed it was more challenging in a good way working with these students (Table 3, statement 30, 5 and 1). There were also differences in how demanding they experienced working alone with these students (Table 3, statement 14, -1 and 4) and if they did get tired of their job sometimes (Table 3, statement 9, -4 and 2). The two factors also had a different view of the acting out behaviours (Table 3, statement 27, -5 and -3) and the self-injury behaviours (Table 3, statement 6, 4 and 1).

Discussion

Classroom disruption and student behaviours have become one of the biggest issues in classrooms today (Alter, Walker, & Landers, 2013; Bru, 2009). One of the main symptoms of SMS is challenging behaviours (Poisson et al., 2015). As far as we know, no one have specifically researched the challenging behaviours of students with SMS in schools. The aim in this study was to explore challenging behaviours in students with SMS, and to explore how school staff experienced and managed these challenging behaviours.

The main finding in this study were the two distinct viewpoints that were discovered in this analysis: 'Managing challenging aggressive and self-injury behaviours' and 'Struggling with intense non-physical challenging behaviours'. The two viewpoints reveal distinct differences in both how the school staff experience the behaviours that the students display at school and how the school staff are handling this behaviour. These findings accord with information presented in 'On the road to success with SMS' (Haas-Givler & Finucane, 2014), regarding the type of behaviours the students display in schools. It seems that it is the more intense non-physical behaviours that is more demanding for the school staff to handle. Investigating the disagreement patterns, it seems as if the staff experience that not all the students display the same type of behaviour, at least not in school. The specific intense non-physical behaviours include various forms of challenging behaviours, such as pushing buttons, picking on things, being demanding, lack of concentration, lack of cooperation and lack of ability to work by themselves and talking all the time. In contrast, in viewpoint one, the staff experience behaviours that are described as more acting out and self-injury behaviours. Both types of behaviours have been described in the literature (Poisson et al., 2015; Wilde et al., 2016), but the behaviours experienced in viewpoint one is more commonly described regarding SMS (Poisson et al., 2015; Wilde et al., 2016). In earlier international investigations (PISA 2000, 2003) (OECD, 2000, 2003). Norway was out on the top on the lists regarding challenging behaviours in school. This has changed in a positive direction in the last two PISA investigations, and Norway have similar results as the other Nordica countries and OECD countries regarding challenging behaviours in school (PISA 2012, 2015) (OECD, 2012, 2016). In a report regarding violence in the schools in Oslo, there are noted an increase of violence both towards schools staff and other students (Utdanningsetaten Oslo kommune, 2018). Norway also comes out on the top on list of how much resources are spent on schools (Eriksen, 2008). This indicates that it is not necessarily a lack of resources, but maybe a lack of knowledge and focus. Some studies have shown that teachers perceive what is defined as mild behaviour challenges, such as off-task behaviour and verbal disruptions, as the most frequent and problematic in schools (Alter et al., 2013; Rosenberg & Jackman, 2003). It is natural to think that working with students who self-injure or who are physically acting out, is one of the most challenging tasks in schools, but both the study from Alter et al (2013) and our results indicate the opposite. There have been a focus on the use of restrictive practises in schools in Norway the last years (Højmark, 2016). There are no laws or regulations regulating the use of restrictive practises in schools, which means they are not allowed to be used in schools. School leadership have had a focus on other strategies, than restrictive practises, to handle challenging behaviours. This increased focus on challenging behaviours and alternative interventions may have led to more support for school staff working with students with aggressive and acting out behaviours (Hansen & Østvold, 2015). The results in this study regarding children with SMS in schools may also reflect that the schools may be advised in how to handle aggression, but since the non-physical challenges for school staff have not been addressed before, there may be a lack of focus on managing students' non-physical challenging behaviours. Another point is looking at the background of the participants. Most of the participants with a special education background loaded on viewpoint 1, while those participants who loaded on viewpoint 2 had different backgrounds. The 'relationship between having special education background and viewpoint 1' is an interesting finding but, due to the lack of studies in this topic, it is difficult to generate discussion on this finding. However, one could hypothesize that, perhaps, school staff with special education training does have a better prerequisite to handle the level of challenging behaviours related to factor 1 (i.e. aggressive and self-injury behaviours). In this regard, there have been several reports the last years in Norway concerning the lack of formal special education competence in school staff providing support to students with special educational needs (Barneombudet, 2017; Nordahl & et. al, 2018). One the other hand, one study found that special education certification programs were more like to have addressed behaviour management than general education certification programs (Flower, McKenna, & Haring, 2017). However, this hypothesis needs to be confirmed by further research involving more participants than those included in our work'.

In addition, there were distinct patterns of agreement and disagreement. The consensus statements revealed that most of the school staff in this study experience it as positive to work with students with SMS and that they experience these students as polite and having good humour. It does not seem as if the challenging behaviours has a negative impact on the teacher-child relationship, as suggested by Haas-Givler (2014) and proposed in the work by Pianta and Hamre (2001). Although the staff experience considerable challenging behaviour, they seem to have positive relationships with the students. They also agree that they experience these students as having challenges with their emotional development and impulse control. These findings are also similar to the results found in earlier research (Haas-Givler & Finucane, 2014; Poisson et al., 2015). Pianta has studied the effect of teacher-student relationship on both academic and social gain in the classroom (Pianta & Hamre, 2009; Pianta & Stuhlman, 2004). There is not necessarily a clear association between the child's development and the teacher-child relationship (Pianta & Stuhlman, 2004), but the fact that it seems to be a positive relationship between students with SMS and school staff could be something that influence the development of both academic achievement and social gain in the school setting. In this study, we found that the school staff seemed to have a positive relationship with the students with SMS, despite the students' challenging behaviour. It is interesting to find a description of such difficult behaviours and positive attitudes towards the students in the same study. Studies have found that children's misbehaving can influence and result in less positive interactions with their teachers (Patterson & Fisher, 2002). Children's ability to form a strong relationship with their teachers is an indicator of positive school adjustment (Hamre & Pianta, 2005). One key component in the positive view of these students may be that the school staff also recognize that these students have some positive traits, such as politeness and humour, which in both viewpoints were placed on the agreement side of the grid. The results of this study may also indicate that challenging behaviours demand extra attention and a close monitoring of the children with SMS so that the children and teachers develop a close relationship, which was identified as crucial by Pianta and Hamre (2005).

In both viewpoints, it appears that it was difficult to inform other school staff about the student and how to handle the challenging behaviour. One participant actually put this statement on least like (-5) explaining that since students with SMS often take some time before they display the challenging behaviour, it is difficult to justify the strict adherence to the planned rules regarding the student. This finding indicates that there may be some challenges in how the school organizes and supports staff that work in such challenging environments. Oliver et al (2010), argue that understanding of an individual can be enhanced when we can trace the individual differences back to a specific genetic disorder. Therefore, providing information to school staff regarding the disorder and how it turns to specific behaviours when the children interact with specific environmental demands at school, may be an important step in supporting the staff working directly with the student with SMS'.

Several of the statements regarded the school staff's experiences of handling the challenging behaviour. It was especially three statements that differed between the viewpoints; in viewpoint two, the staff scored slightly higher than viewpoint one on 'being tired of their work' and 'experiencing it as demanding to be alone with the student' and scored lower than viewpoint on 'thinking that it is challenging in a good way to work with the student'. Both viewpoints scored quite similar on the statements regarding how to handle the challenging behaviours and knowing what to do in situations of both acting out behaviours and self-injury and that it is positive to work with these students. It seems as if the school staff are experiencing a challenging work day with these students, but they are coping well with it in general. We observe that in viewpoint two, the school staff express more issues with coping, probably based on the intensity and type of the behaviour. It seems as if the non-physical and intense behaviours is more challenging to cope with than the acting out behaviour. Coping is important in how we manage stressful situations (Drageset, 2014; Lazarus & Folkman, 1984). The school staff requires information and knowledge of how to handle the different types of challenging behaviours in students with SMS. By adding these, their perceptions about these students may change, as well as their self-efficacy when addressing the challenging behaviours and the needs of their students, thus giving teachers more tools for a more effective coping process'.

This is, as far as I know, the first Q studies performed regarding SMS. It seems like a useful method in exploring school staffs' viewpoints in managing challenging behaviours in the schools. Some of the results may have been difficult to reveal in an interview setting. This study explores school staff's views and is limited to the participants who performed the Q sort. The findings cannot be generalized to all staff who work with SMS children, but the study provides insight into some views that may be present in a larger study. There may be a limitation in using Q sorts sent in the mail to the participants, and not performing the sort face-to-face. In a face-to-face setting misunderstanding may be solved and body language may also be studied. However, studies have shown that Q sort sent in the mail or performed using a computer has no difference in reliability or validity than interview based (face-to-face) Q sorts (Reber et al., 2000; Van Tubergen & Olins, 1979).

In this study we have established that students with SMS have a variety of challenging behaviours in school. It specifically looks like students with intense non-physical challenging behaviours are more of a challenge to work with probably due to lack of knowledge how to deal with this type of challenging behaviours. More specific information regarding the type and intensity of challenging behaviours are needed for school staff and others working with persons with SMS. And more research regarding interventions and how to handle these behaviours are also needed. The lack of knowledge may be risk factors for stress (Lazarus & Folkman, 1984). It also looks like special education trained teachers are better prepared to handle the challenging behaviours in school, but due to this small sample this needs to be further investigated. Further research is also needed in how to support school staff working with students with SMS.

The conclusion of this study is that there must be a greater emphasis on education and advising and supporting school staff's work with the non-physical challenging behaviours aspects of teaching children with SMS, as well as a continued focus on challenging aggressive behaviour.

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Disclosure

The authors declare that they have no competing interests.

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Table 1. Fisher balanced block design, N=40

	Extrovert	**Introvert**	**School Staff Experiences**
Alone	6 statements	6 statements	12 statements
With other students	6 statements	6 statements	
Various	4 statements		

Table 2. School staff characteristics

Staff Code
1 - Special Education Teacher Female student
2 – Teacher Assistant Male student
3 – Unknown profession Female student
4 – Special Education Teacher Male student
5 – Special Education Teacher Female student
6 – Teacher Female student
7 – Teacher Assistant Female student
8 – Teacher Assistant Male student
9 – Special Education Teacher Male student
10 – Special Education Teacher Male student
11 – Unknown profession Male student
12 – Other school staff Male student
13 – Other school staff Male student
14 – Special Education Teacher Female student

Table 3. Statements and factor scores, including consensus and distinguishing statements

No	**Statements**	**Factor arrays**	
		1	**2**
1*	*The student has a mature emotional development (behaves similar to other people his/her own age).*	-4	-5
2	The student often sits quietly by him/herself.	0	-4
3*	*The student has a lot of creativity.*	0	0
4*	*The student has good humour.*	3	3
5*	*The student is rarely sleepy and tired during the day.*	-2	-2
6	The student self-injures by hitting, scratching, or biting.	4	1
7*	*The student self-stimulates or has repetitive behaviour (spins or pushes on things repeatedly or asks the same questions repeatedly).*	4	3
8*	*The student is polite and social.*	3	2
9	Sometimes, I am really tired of my job.	-4	2
10*	*The student is good at technical things.*	1	2
11*	*The student has mastered 'conquer and divide' techniques.*	1	1
12	*The student does not care for other people (students or adults).*	-2	-1
13*	*I think it is just ok to be spat on.*	-3	-1
14	I think it is demanding to be alone with the student.	-1	4
15*	*The student has good impulse control.*	-3	-4
16*	*The student does not help other people.*	0	0
17	The student has bad and insulting language.	0	-2

18	The student works well with other students of the same age.	0	-2
19	I have not experienced the student getting angry at school.	-5	-1
20	The student can be experienced as very intense.	2	5
21*	*The student knows which 'buttons' to push.*	3	4
22*	*The student is rarely afraid in new situations and when meeting new people.*	0	0
23	I never get provoked by the student.	-1	2
24*	*I experience that it is difficult to divert the student's attention to something else.*	1	1
25*	*The student has a stable mood.*	-2	-3
26	The student works with and concentrates on academic work over time.	1	-4
27	The student never gets angry with screaming, kicking and hitting.	-5	-3
28	The student cooperates well with other students in group activities.	-1	-3
29	I am not sure what to do in difficult situations.	-2	0
30	Working with the student is challenging in a good way.	5	1
31	The student is not preoccupied with adults.	-3	-5
32	I know what to do when the student self-injures.	2	-1
33*	*I think it is difficult to watch when the student self-injures.*	1	1
34	*The student never destroys things at school.*	-1	-2
35	*The student has good long-term memory.*	2	0
36*	*It is positive to work with the student.*	5	4

37*	*The student demands constant attention.*	2	5
38*	*I know what to do if the student displays challenging behaviour.*	4	3
39	I think it is scary when the student gets angry or loses control.	-4	0
40*	*I think it is easy to explain to the other teachers about the student and how to handle situations.*	-1	-1

Note. Statements in italics are consensus statements non-significant at $p<0.01$, and statements also flagged with an * are also non-significant at $p<0.05$) (statements not in italic are distinguishing statements (variance across factor z-scores)

Table 4. Factor loadings with an X indicating a defining Q sort

Staff Code	**Factor 1**	**Factor 2**
1SETF	0.2673	0.5504X
5SETF	0.7113X	0.3926
4SETM	0.7095X	0.2106
3UF	0.4205	0.7488X
2TAM	0.8568X	0.2148
6TF	0.6856X	0.4224
7TAF	0.2871	0.6258X
8TAM	0.8100X	0.2290
9SETM	0.7064X	0.1447
10SETM	0.8043X	0.2794
11UM	0.8387X	0.2205
12OM	-0.0207	0.7438X
13OM	0.6232X	0.3637
14SETF	0.6788X	0.1271

Note. SET: Special Education Teacher, T: Teacher, TA: Teacher Assistant, O: Other school education, U: Unknown profession,

F: Female student with SMS, M: Male student with SMS

Table 5. Characteristic statements and scores for the two viewpoints

	Factor 1: Managing challenging aggressive and self-injury behaviours in school	**Factor 2: Struggling with intense non-physical challenging behaviours**
Positive side of the grid	30. Working with the student is challenging in a good way (+5)* 36. It is positive to work with the student (+5) 6. The student self-injures by hitting, scratching, or biting (+4)* 7. The student self-stimulates or has repetitive behaviour (spins or pushes on things repeatedly or asks the same questions repeatedly) (+4) 38. I know what to do if the student displays challenging behaviour (+4)* 21. The student know which 'buttons" to push (+3) 4. The student has good humour (+3) 8. The student is polite and social (+3)	20. The student can be experienced as very intense (+5) 37. The student demands constant attention (+5) 21. The student knows which 'buttons' to push (+4) 36. It is positive to work with the student (+4) 14. I think it is demanding to be alone with the student (+4) 7. The student self-stimulates or has repetitive behaviour (spins or pushes on things repeatedly or asks the same questions repeatedly) (+3) 4. The student has good humour (+3) 38. I know what to do if the student displays challenging behaviour (+3)

Negative side of the grid	13. I think it is just ok to be spat on (-3) 15. The student has good impulse control (-3) 31. The student is not preoccupied with adults (-3)* 1. The student has a mature emotional development (behaves similar to other people his/her own age) (-4) 39. I think it is scary when the student gets angry or loses control (-4)* 9. Sometimes, I am really tired of my job (-4)* 19. I have not experienced the student getting angry at school (-5)* 27. The student never gets angry with screaming, kicking and hitting (-5)*	25. The student has a stable mood (-3) 27. The student never gets angry with screaming, kicking and hitting (-3) 28. The student cooperates well with other students in group activities (-3) 26. The student works with and concentrates on academic work over time (-4) 2. The student often sits quietly by him/herself (-4) 15. The student has good impulse control (-4) 1. The student has a mature emotional development (behaves similar to other people his/her own age) (-5) 31. The student is not preoccupied with adults (-5)

Note. Asterisk (*) indicates significant at p<.01, for the distinguishing statements for factor 1.

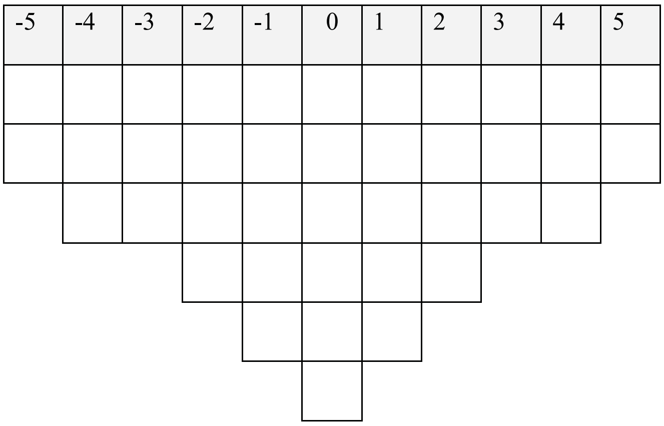


Figure 1. Grid used during Q sorting.

Paper V

School Staff's Experiences of Working with Smith-Magenis syndrome in Schools –How Do They Handle the Behaviours?

Abstract

The aim of this study is to explore how school staff are handling the challenging behaviours of students with Smith-Magenis syndrome (SMS) in schools. SMS is a rare genetic syndrome. Fourteen school staff of students with SMS in Norway participated in the study by sorting 40 statements according to Q methodology. The sorting was analysed utilising by-person factor analysis. Four viewpoints were revealed, 1) In control, 2) struggling, 3) Strugglers relying on parents and 4) Support dependent. Several of the consensus statements were regarding academic work with the students with SMS. We found consensus around the fact that academic work is not a priority for these students. To be in control and feel safe when working with student with SMS, school staff are dependent on support from the school's leadership and colleagues, in addition to cooperation with parents. In this study we found that school staff working with females with SMS are struggling more than school staff working with males with SMS. We also found that there is more focus on challenging behaviours than focus on academic work with students with SMS.

Keywords: Smith-Magenis syndrome, challenging behaviours, school, school staff, Q methodology, handling behaviours.

Introduction

According to Kokkinos (2007) managing students misbehaving is one issue that has the most impact on teachers stress and burnout.

Challenging behaviours in schools put a great demand on the staffs capacities such as their competency, motivation, and values (Roland, Øverland, & Byrkjedal-Sørby, 2016). Working with challenging behaviours in schools demands special skills in the team, and it looks like there is a general lack of resources in cases regarding students with challenging behaviours (Roland et al., 2016).

In phenomenological study of teachers experience of teaching children with ADHD the main themes that was discovered were; lack of information, child’s behaviours is disruptive, lack of resources, lack of support, and burden having the child in the class (Harazni & Alkaissi, 2016).

Students with SMS

SMS is a rare, neurodevelopmental disorder caused by haploinsufficiency of the retinoic acid induced 1 (RAI1) gene due to either a deletion of chromosome 17 (17p11.2), or a mutation in RAI1 (Slager, Newton, Vlangos, Finucane, & Elsea, 2003; A. C Smith et al., 1986). Common characteristics include cognitive impairment sleep disturbance, self-injury, stereotypies and aggressive behaviours (Greenberg et al., 1996; A. C. Smith, Dykens, & Greenberg, 1998). The incidence of SMS is estimated to be 1:15 000–1:25 000 births (Greenberg et al., 1991). In Norway, Frambu Resource Centre for Rare Disorders know of approximately 40 persons with SMS. The disorder is under diagnosed and delayed diagnosis is common (Gropman, Duncan, & Smith, 2006).

Students with Smith-Magenis Syndrome (SMS) have a neurobehavioural phenotype that has been characterized as challenging for both parents and teachers. The educational functioning in students with SMS is significantly influenced by the neurobehavioural phenotype based on both physical, cognitive and medical symptoms (Haas-Givler & Finucane, 2014). These problems include sleep disruption, behavioural and psychiatric symptoms, stereotypic behaviours, sensory integration issues and variable levels of. Intellectual disabilities (ID) (De Leersnyder et al., 2001; Gropman et al., 2006; Laje et al., 2010; Madduri et al., 2006; Martin, Wolters, & Smith, 2006; Poisson et al., 2015). Many persons with SMS also meets the criteria for Autism spectrum disorders (ASD) (Laje et al., 2010; Martin et al., 2006).

Recent research has reported a reversed gender difference regarding SMS and ASD, favouring the females (Nag, Nordgren, Anderlid, & Naerland, 2018).Variable levels of cognitive impairment have been demonstrated in SMS, ranging from lower levels of the normal population to severe ID, most of them having mild to moderate ID (Poisson et al., 2015). The substantial challenging behaviours and impaired adaptive function leads to a lower perceived cognitive level for many individuals with SMS (Neira-Fresneda & Potocki, 2015). This may interfere disproportionately with learning and school performance and affect the educational performance (Haas-Givler & Finucane, 2014). It has also been described that individuals with SMS need more assistance than expected based on their level of intellectual functioning (Udwin, Webber, & Horn, 2001).

Recent research have stated that students with SMS display both challenging aggressive and self-injury behaviours and non-physical challenging behaviours in schools (Nag, Øverland, & Nærland, In review). The study by Nag et al (In review) also revealed that the school staff are coping differently according to the type of behaviours experienced as problematic by the staff (e. g. aggressive vs non-physical challenging behaviours).

Challenging behaviours often has a negative impact on the persons learning performance, but also prove a challenge for the learning environment (Roland et al., 2016). According to Neira-Fresneda (2015) both educational and behavioural intervention for students with SMS are extremely challenging.

Challenging behaviours in schools put a great demand on the staffs capacities such as their competency, motivation, and values (Roland et al., 2016). Working with challenging behaviours in schools demands special skills in the team, and it looks like the is a general lack of resources in cases regarding students with challenging behaviours (Roland et al., 2016).

Aim of the study

The aim of this study was to explore the experiences of how the school staff handle the challenging behaviours in students with Smith-Magenis syndrome.

Methods

This study follows the same design and data was collected at the same time as a previous study regarding the challenging behaviours of SMS and school staffs' coping (Nag et al., In review).

William Stephenson introduced and developed Q methodology in 1935 (Stephenson, 1935). According to Stephenson (1953) is a person's subjectivity a communicative behaviours, something that can me measures and studied. Q methodology is designed to explore patterns in what people say to themselves and to others. A by-person factor analysis reveal the subjectivity by identifying unique viewpoints revealed as factor structures (Brown, 1986; Stephenson, 1953). There are commonly five steps used in Q studies (Van Exel & de

Graaf, 2005). a) Definition of concourse, b) developing the Q set, c) defining the participants, d) the Q sort and analysis and e) interpretation.

Definition of concourse

The concourse is defined as the universe of available communication about a specific topic (Thorsen & Allgood, 2010). The term comes from Latin *concursus,* meaning «a running together," as when ideas run together in thought (Brown, 1993). The content of the concourse can come from different sources such as interviews, conversations, social media, magazines or literature (Brown, 1980). In this Q study open-ended questionnaire and standardized questionnaires (Developmental Behaviour Checklist and Vineland Adaptive Behavior Scales) were completed by the parents of persons with SMS as part of a larger study (Nag et al., 2018 and ongoing work) and literature regarding SMS (Haas-Givler & Finucane, 2014; Neira-Fresneda & Potocki, 2015) were used to identify the concourse.

Developing the Q set

The statements from the concourse were systematically reduced from 150 to 40. Fisher's balanced block design was used to create a balanced and structured set of statements (Fisher, 1960; Stephenson, 1953). In this study a 3 x 2 block design were used. Three main dimensions (methods, cooperation, guidance/knowledge) on one side and two main dimensions (behaviours and academic) on the other side were used to ensure coverage of a wide range of statements. An additional statements category of "school staff feelings" was also added (Table 1). The statements were reduced by grouping them in similar groups. From the statements that addressed the same issue, one statement was selected, or statements were combined. The statements were printed on separate cards and numbered arbitrarily, and the generated statements are known as the Q set (Coogan & Herrington, 2011; Van Exel & de

Graaf, 2005). The final selection of statements is presented in Table 4 (translated from Norwegian to English).

Table 1.

Fishers balanced block design, N=40

	Behaviours	Academic	School Staff Feelings
Methods	6 statements	6 statements	5 statements
Cooperation	5 statements	4 statements	
Guidance/Knowledge	7 statements	7 Statements	

Participants (P set)

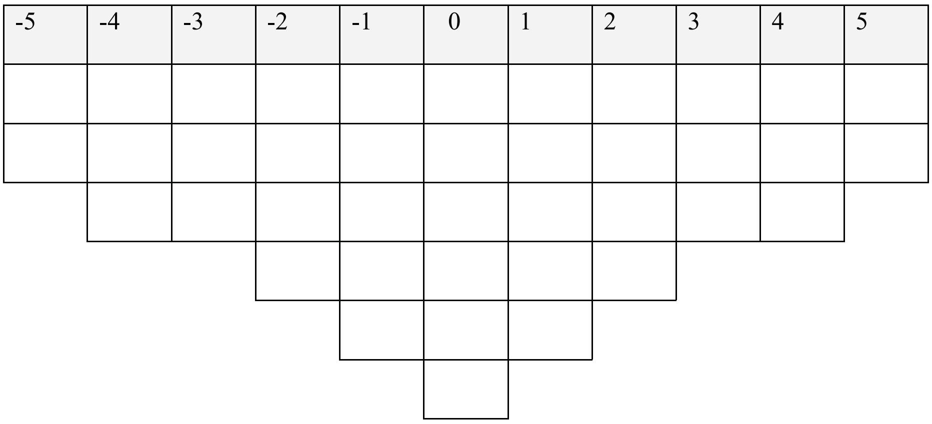
In this study the P set consist of school staff currently working with a student with SMS. Principals of the 10 Norwegian schools (we know of only 10 students in grade school in Norway) were contacted and asked to distribute the Q sort to up till three of their staff members currently working with a student with SMS. Approximately 50 % (14) volunteered to participate in the study and completed and returned the Q sort. One reminder email was sent to the principals after one month. The participants' position in the schools are indicated in Table 2. Most of the participants (eleven) worked in regular education settings, only three worked in special education schools. More than half of the participants (eight) did not have any special education training. None of the participants had worked with a student with SMS prior to the students they had now. Six of the participants had worked with the students with SMS for 1-2 years, five had worked with the student with SMS for 4-5 years, and one had worked with the students with SMS for 7 years. Two of the participants did not indicate how long they had worked with the students with SMS.

Q sorting

The students with SMS are spread geographically all over Norway therefore a face-to-face Q sorts would be very ineffective and expensive. The Q sorts were sent in the mail and the participants sorted it on their own. Studies have shown that Q sort sent in the mail or performed using a computer has no difference in reliability or validity than interview based (face-to-face) Q sorts (Reber, Kaufman, & Cropp, 2000; Van Tubergen & Olins, 1979). The participants sort the cards from most like to most unlike to their experience working with this student with SMS. In this study a distribution grid with 11 categories (from +5 to -5) was created to fit 40 statements cards (Fig. 1). The package also included information about the study and written instructions. After they finished sorting the cards, the participants were instructed to write down the numbers of the statements in the correct place in the grid. We also asked them to provide a rationale for the placement of the two cards placed on the far right (+5) and far left (-5) sides of the grid. A form was provided for this written rationale.

Figure 1.

The grid used during Q sort



Data Analysis and Interpretation

All the Q sorts were plotted and analysed using the PQ Method Programme (Schmolck, 2002). In the analysis process the degree, or level of dissimilarity and similarity of points between the individual sorters are calculated. Then, factor analysis is performed to examine how many groupings of similar Q sorts there are. The factors or views consist of people with similar views (sorts) (Van Exel & de Graaf, 2005). Factor scores are essentially weighted z-scores for each statement in the Q-sample; these scores can be converted into an array of scores (factor array) that correspond to the plus 5 to minus 5 values in the original Q-sort continuum (McKeown & Thomas, 1988). The factors were interpreted based on the characteristic statements of each factor, as well as distinguishing and consensus statements. In addition, written statements regarding why the participants placed the statements on either end of the scale were used to enlighten the results.

Ethical Considerations

This study was part of a larger study regarding SMS. This study was approved by the Norwegian Ethical Committee (2015/1026). The participants signed an informed consent form.

Results

In the principal component analysis, the programme calculated eight unrotated factors with eigenvalues from 7.29 to 0.36 and explained variances from 52 to 3 %. Using varimax rotation, four factors were extracted. One Q sort was confounded, which means it loaded similar on more than one factor. Five participants loaded on factor 1, three on factor 2, three on factor 3 and two participants loaded on the fourth factor (Table 2). Only two participants loaded on the fourth factor, but we needed to calculate with four factors to get three distinct factors (Table 2). In the principal component analysis, the fourth factor had an eigenvalue less than 1 (0.91) but in the investigation of the factors, factor four had some distinct differences from the three other factors and we have therefore decided to present all four factors. These four factors explained 28, 17, 17 and 14 % variances respectively. All four factors were correlated (Table 3), indicating an overlap between them. But all four factors also have some distinct differences.

Table 2.

Factor loadings with an X indicating a defining Q sort

Staff Code*	Factor 1	Factor 2	Factor 3	Factor 4
1SET F	0.0603	0.2158	0.7750X	0.4302
2TA M	0.8490X	0.0420	0.1349	0.1485
4SET M	0.4880	0.3829	0.3033	0.1459
3U F	0.3449	0.8222X	0.0944	0.0151
5SET F	0.3449	0.2000	-0.0779	0.8450X
6T F	0.2975	0.1581	0.4541	0.6755X
7TA F	-0.0516	0.8474X	0.2095	0.1941
8TA M	0.7099X	0.2096	0.2357	0.3610
9SET M	0.5238	0.3532	0.6378X	0.0377
10SET M	0.6391X	0.4363	0.3668	0.2837
11U M	0.7408X	0.2674	0.1892	0.3456
12O M	0.2565	0.5779X	0.2173	0.3914
13O M	0.8595X	0.0797	0.1919	0.1495
14SET F	0.3803	0.1510	0.8238X	-0.0950

Note: * SET: Special Education Teacher, T: Teacher, TA: Teacher Assistant, O: Other school education, U: Unknown profession, F: Female student with SMS, M: Male student with SMS

Table 3

Correlation among factors

	Factor 1	Factor 2	Factor 3	Factor 4
Factor 1	-	r = 0.43	r = 0.59	r = 0.61
Factor 2		-	r =0.48	r = 0.43
Factor 3			-	r = 0.38

Table 4 gives an overview of which statements that have loaded high and low on each of the four factors. The statements on the extreme ends of the sorting grid (+5, +4, -4 and -5) characterise the factor and are used to interpret and understand the meaning of the factors.

[Table 4 here]

Interpretation of factors

Factor 1: In control. The view in this factor is that they have gotten guidance and information regarding SMS and are handling the work well. They enjoy their work and feel safe even though the students display challenging behaviours. Two of the participants explain the placement of statement 25 (Table 5), I am afraid when the student gets angry and is screaming, kicking, or hitting, on least like this way:

> *I have worked with this student for many years. I feel that I know this student well. I think it is an advantage to know each other. He also knows my boundaries.*
>
> *I know what to do in the different situations, and I mostly know what sets of the behaviours.*

They are also working well with the parents. One of the participants comments the cooperation with the parents this way:

> *Cooperation with the parents are instructive and good. They are grateful for the job we are doing, and they often tell us that. I experience them as comfortable that we are taking care of their child in a good way.*

Factor 2: Struggling. In this factor the view is that it is hard to work with students with SMS because of the challenging behaviours, and they struggle because they need to do things a little differently than they are doing with other students. One participant describes the placement of statement 20 (Table 5), I think it is hard to work with this student because if the behaviours, on most like this way:

> *The behaviours are the most challenging regarding working with this student. There is a lot of planning and adaptation behind every day with this student, and still it may end up with challenging behaviours.*

They also think it is hard to inform parents of challenging behaviours because of the parents' feelings but they are working well with the parents. In this group they also lack support from the leadership at the schools and from other colleagues.

Factor 3: strugglers relying on the parents. In this view the school staff have gotten their information and guidance from the parents and not so much from the school leadership and guidance from other than the parents (pedagogical centres for example).

> *I am having a good dialog with the parents. They know their child the best. We can discuss solutions and challenges.*

In this factor they also struggle with academic focus due to the challenging behaviours, but like their work with these students.

> *A lot of the time at school are used for breaks, rewards, acting out, repetition, slower progressions etc.*

Factor 4: support dependent. In this view they have gotten guidance and training regarding SMS and have support from the school leadership and colleagues.

> *We have gotten close and good guidance and follow up from the habilitation centre. They have been available for any questions, extra visits etc when need because of increase in challenging behaviours.*

They have not gotten a lot of information from the parents and are not working a lot with the parents. One of the participants explain the placement of statement 34 (Table 5), it is the parents that have taught us how to handle the behaviours, on least like this way:

> *The parents are exhausted and do not have the energy to contribute to help us in the school day.*

Distinguishing statements

Eighteen of the statements distinguish between the four different factor views. The scores on all statements and distinguishing statements are presented in Table 5.

[Table 5 here]

Five of the statements are distinguishing for factor 1 (in control). Those holding this view believe that the challenging behaviours does not stop them from having an academic focus, they are, together with those holding factor 3 (strugglers relying on parents), not afraid when the students gets angry, and they are clearly looking forward to their work days because of these students' charm, humour and love. The school staff holding this position believe that knowledge regarding SMS is not as important when planning adaptations for the student as the school staff representing the three other factor views. On the other side the staff holding this view believe that knowledge regarding SMS is important when planning academic adaptation.

Six of the statements distinguishing for factor 2 (struggling). The school staff holding this position think it is difficult to work with these students because of their challenging behaviours, believe it is hard when parents gets upset because of the feedback from the school, they lack support both from colleagues and the leadership at school and do not receive sufficient follow up after bad episodes.

Seven of the statements distinguish factor 3 (strugglers relying on parents) from the other factors. School staff holding this view receive god support from the parents and do not lack training regarding SMS, but they have not received any training in how to handle the challenging behaviours. Still, they do not get afraid when the students get angry.

Four statements distinguish factor 4 (support dependent) from the other factors. Staff holding this view do get support after difficult episodes and have no problem following the recommendations regarding the challenging behaviours. The staff holding this view have not been trained by the parents and value the cooperation with the parents less than the staff in the other three factors.

Consensus statements

Fourteen statements do not distinguish between any pair of the factors (Table 5). In all the factors the staff agree that both preparation for the student a head of the activities and to be ahead of difficult situations are important for students with SMS. In all the factors they also agree that they do have a structure that prevent challenging behaviours. Several of the consensus statements are regarding academic work with the students with SMS and they have scores in the middle (+2 to -2). It looks like it is consensus around the fact that academic work is not a priority for these students.

Differences based on gender of student and the school staffs' role

Five out of eight school staff working with male students loaded on factor 1: In control. None of the school staff working with female student loaded on factor 1. School staff working with female students were evenly spread out loading on factor 2 (struggling), 3 (strugglers relying on parents) and 4 (support dependent). None of the school staff working with male students loaded on factor 4: Support dependent.

Most of the teachers (four out of six) loaded on factor 3 and 4 (strugglers relying on parents and support dependent). All participants loading on factor 4 were teachers. The teacher assistant all loaded on factor 1 and 2 (in control and struggling). The rest were spread out on factor 1, 2 and 3.

Discussion

The purpose of this study was to explore school staff's experiences of handling the challenging behaviours students with SMS display in schools. We found four distinct viewpoints in how the school staff handle the challenging behaviours in schools. The four viewpoints mainly differ in how they perceive support from school leadership and cooperation with the parents.

Cooperation with parents and support from the school leadership

One of the most important findings in this study was how the school staff perceived on one side the support and cooperation with parents. On the other side, support from the school leadership and colleagues. How the four different factors perceive those two dimensions are illustrated in Figure 2.

Figure 2.

Factors divided on parent support and leadership support

		Support from school leadership and colleagues	
		Positive	Negative
Parent support and cooperation	Positive	Factor 1, in control	Factor 3, strugglers relying on the parents
	Negative	Factor 4, support dependent	Factor 2, struggling

It does seem like that to be in control and feel safe working with students with SMS you especially need support from school leadership and colleagues. Those loading on viewpoint 2 (struggling) seems to be in a particularly difficult position experiencing lack of support from school leadership and colleagues and experiencing it as challenging to work with the parents. Support and cooperation with parents are also an important piece, but if you only have that and not support from the leadership and colleagues it seems like it is hard to handle the challenging behaviours in the schools. In a qualitative study of teachers teaching students with ADHD a similar results emerged and one of the major accusations was lack of support from the school leadership (Harazni & Alkaissi, 2016).

Gender difference

Another interesting finding in this study is that the school staff of male and female students seems to handle the challenging behaviours differently. Most of the staff working with a male student seems to load on viewpoint 1: in control. In this viewpoint it seems like they are both working well with the parents and get support from the school leadership and colleagues. This finding may indicate that it is more challenging to work with females with SMS than males. Other studies have found some gender differences in SMS, with both more autism spectrum symptomatology, hypersensitivity and frustration with communication in females (Edelman et al., 2007; Laje et al., 2010; Nag et al., 2018). It is especially in the social domain that females have more problems than males (Nag et al., 2018). It may be that problems in the social area provides the school staff with more challenges than other behavioural challenges. Nag et al (2018) also found lower levels of ID in females and lower score on adapted behaviour measured by Vineland Adaptive Behaviour Scale, but neither were significant. In general, it seems like some of the important skills needed in schools, such as social competence, adaptive behaviour and cognitive level are lower in females than males. It may be therefore it seems like school staff working with males are handling the challenging behaviours better than school staff working with female student with SMS.

Lack of focus on academic work

The third important finding in this study is the focus on academic work with students diagnosed with SMS. One of the immediately observations is that neither of the statements regarding academic work really end up on either ends of the grid. They are almost all concentrated from -3 to +3 in all four viewpoints, with a few exceptions. It seems like the school staff have very strong opinions or attitudes toward the challenging behaviours, cooperation with parents or support from school leadership and colleagues. When it comes to the academic work those strong opinions or attitudes seems to fade away. On the statement stating: "I know how to work academically with this student" (statement 11, table 4) the scores on the different viewpoints are: 1, 2, 0 and 2, respectively. Udwin et al (2001) found a lack of progress in educational achievement from childhood to adulthood. They also found low abilities in other areas such as independence in daily living skills and occupational achievement. This discrepancy between different abilities and the cognitive level have been attributed to the behavioural challenges (Udwin et al., 2001). Udwin et al (2001) posed a question if this lack of abilities could be a function of limited educational input or a ceiling in abilities regarding individuals with SMS. Our study indicates, at least, a limited focus on academic or educational focus. It seems like the challenging behaviours do capture at lot of the focus regarding this disorder in school. Recent research (Nag & Nærland, In process) indicates a relation between daily living skills and challenging behaviours. Therefore, an effort to shift the focus from just handling the challenging behaviours toward a focus on learning and educational outcomes could provide a decrease in challenging behaviours. In general education, one of the suggestions for preventing challenging behaviours in school is to provide explicit and engaging academic instruction (Alter, Walker, & Landers, 2013). It would be interesting to see how this suggestion would impact the challenging behaviours overall regarding SMS, but especially the challenging behaviours in schools.

In conclusion to be in control and feel safe when working with student with SMS, school staff are dependent on support from the school's leadership and colleagues, in addition to cooperation with parents. School staff working with females with SMS are struggling more than school staff working with males with SMS. We also found that here is more focus on challenging behaviours than focus on academic work with students with SMS.

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The authors declare that they have no competing interests.

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Table 4.

Characteristic statements and scores for the four viewpoints

	Factor 1 – in control	Factor 2 - strugglers	Factor 3 – strugglers relying on parents	Factor 4 – support dependent
The positive side of the grid	30. I have been given guidance in how to handle this student's challenges and feel safe (+5) 33. I am looking forward to every day because of this student's charm, humour, and love (+5) 40. The most important knowledge on how to work with this student have I gotten through my own practise in the school (+4) 12. We may avoid many problems by being ahead of possible tricky situations (+4) 36. Knowledge about this disorder is important regarding the academic adaptation (+4) 31. Cooperation with the parents are important regarding the adaptation in the school (+3) 23. I get good support from the parents and like to talk with them (+3) 3. Regular pedagogical/special education tools work well with this student (+3)	12. We may avoid many problems by being ahead of possible tricky situations (+5) 14. It needs clear boundaries to be able to get any academically work done (+5) 20. I think it is hard to work with this student because if the behaviours (+4) 2. I often use individual support conversations before and after challenging behaviours (+4) 26. It is hard when the parents get upset regarding feedback about the student's act (+4) 36. Knowledge about this disorder is important regarding the academic adaptation (+3) 23. I get good support from the parents and like to talk with them (+3) 31. Cooperation with the parents are important regarding the adaptation in the school (+3)	12. We may avoid many problems by being ahead of possible tricky situations (+5) 23. I get good support from the parents and like to talk with them (+5) 31. Cooperation with the parents are important regarding the adaptation in the school (+4) 2. I often use individual support conversations before and after challenging behaviour (+4) 40. The most important knowledge on how to work with this student have I gotten through my own practise in the school (+4) 36. Knowledge about this disorder is important regarding the academic adaptation (+3) 15. It is the challenging behaviours that makes it difficult to have focus on the academic (+3) 33. I am looking forward to every day because of this student's charm, humour, and love (+3)	36. Knowledge about this disorder is important regarding the academic adaptation (+5) 30. I have been given guidance in how to handle this student's challenges and feel safe (+5) 12. We may avoid many problems by being ahead of possible tricky situations (+4) 18. I get a good follow-up after difficult situations with this student (+4) 28. I receive good support from the school leadership, so I can focus on the academic work (+4) 2. I often use individual support conversations before and after challenging behaviours (+3) 14. It needs clear boundaries to be able to get any academically work done (+3) 21. I know what I should do if the student self-injures (+3)

The negative side of the grid	8. I lack training regarding this disorder (-3) 15. It is the challenging behaviours that makes it difficult to have focus on the academic (-3) 6. We are lacking a structure that could prevent challenging behaviours (-3) 22. The parents are difficult to cooperate with because the student behaves different at home and at the school (-4) 20. I think it is hard to work with this student because if the behaviours (-4) 1. This student does not need preparation before an activity to make it work (-4) 24. I think it is challenging to cooperate with the parents of this student (-5) 25. I am afraid when the student gets angry and is screaming, kicking, or hitting (-5)	6. We are lacking a structure that could prevent challenging behaviours (-3) 13. I can work academically with this student the same way I do with all other students (-3) 8. I lack training regarding this disorder (-3) 28. I receive good support from the school leadership, so I can focus on the academic work (-4) 24. I think it is challenging to cooperate with the parents of this student (-4) 29. It is not important to me that my colleagues and leadership at the school gives me praise and value my work (-4) 19. It is not important to have knowledge regarding this disorder in order make adaptation to this student (-5) 1.This student does not need preparation before an activity to make it work (-5)	1. This student does not need preparation before an activity to make it work (-3) 30. I have been given guidance in how to handle this student’s challenges and feel safe (-3) 37. It is easy to put academic demands on this student (-3) 25. I am afraid when the student gets angry and is screaming, kicking, or hitting (-4) 17. The parents have too high expectations regarding the academic work and that makes the cooperation with them difficult (-4) 19. It is not important to have knowledge regarding this disorder in order make adaptation to this student (-4) 24. I think it is challenging to cooperate with the parents of this student (-5) 22. The parents are difficult to cooperate with because the student behaves different at home and at the school (-5)	20. I think it is hard to work with this student because if the behaviours (-3) 39. There is a lot of knowledge regarding this disorder and academic functioning (-3) 6. We are lacking a structure that could prevent challenging behaviours (-3) 19. It is not important to have knowledge regarding this disorder in order make adaptation to this student (-4) 1. This student does not need preparation before an activity to make it work (-4) 13. I can work academically with this student the same way I do with all other students (-4) 10. It is difficult to follow the recommendations we have been given regarding the behaviours (-5) 34. It is the parents that have taught us how to handle the behaviours (-5)

Table 5.

Statements and factor scores, including consensus and distinguishing statements

Statement #		Factor 1	Factor 2	Factor 3	Factor 4
1*	*This student does not need preparation before an activity to make it work.*	-4	-5	-3	-4
2	I often use individual support conversations before and after challenging behaviours.	1**	4	4	3
3	Regular pedagogical/special education tools work well with this student.	3	0	-2*	2
4**	*The school staff can give guidance and help parents regarding behaviours.*	0	1	1	-1
5**	*It is easy to make choices regarding the academic work.*	0	-1	0	-1
6**	*We are lacking a structure that could prevent challenging behaviours.*	-3	-3	-2	-3
7*	*The parents are more preoccupied with the social rather than the academic.*	0	-1	2*	-1
8	I lack training regarding this disorder.	-3	-3	2**	-2
9	It is the parents that have given us information regarding this disorder.	1	-2	2	-2
10	It is difficult to follow the recommendations we have been given regarding the behaviours.	-1	1	0	-5**
11**	*I know how to work academically with this student*	1	2	0	2

12**	*We may avoid many problems by being ahead of possible tricky situations.*	4	5	5	4
13*	*I can work academically with this student the same way I do with all other students.*	-2	-3	-1	-4
14	It needs clear boundaries to be able to get any academically work done.	2	5	1	3
15	It is the challenging behaviours that makes it difficult to have focus on the academic	-3*	2	3	1
16**	*I have good tools to prohibit the challenging behaviours.*	1	-1	0	0
17	The parents have too high expectations regarding the academic work and that makes the cooperation with them difficult.	-2	1	-4	-1
18	I get a good follow-up after difficult situations with this student.	2	-2*	1	4*
19	It is not important to have knowledge regarding this disorder in order make adaptation to this student.	-1**	-5	-4	-4
20	I think it is hard to work with this student because if the behaviours.	-4	4*	0*	-3
21	I know what I should do if the student self-injures.	2	0	-1	3

22	The parents are difficult to cooperate with because the student behaves different at home and at the school.	-4	-1	-5	-2
23	I get good support from the parents and like to talk with them.	3	3	5**	1
24	I think it is challenging to cooperate with the parents of this student.	-5	-4	-5	-1
25	I am afraid when the student gets angry and is screaming, kicking, or hitting.	-5**	1	-4*	0
26	It is hard when the parents get upset regarding feedback about the student’s act.	-1	4*	-2	0
27*	*It is hard to get a function cooperation around homework.*	0	2	-1	0
28	I receive good support from the school leadership, so I can focus on the academic work.	1	-4*	1	3
29	It is not important to me that my colleagues and leadership at the school gives me praise and value my work.	0	-4*	0	1
30	I have been given guidance in how to handle this student’s challenges and feel safe.	5	0*	-3**	5
31	Cooperation with the parents are important regarding the adaptation in the school.	3	3	4	0*

32**	*I think it is hard to work with parents in despair and full of worry.*	-1	0	-2	0
33	I am looking forward to every day because of this student's charm, humour, and love	5**	2	3	1
34	It is the parents that have taught us how to handle the behaviours.	2	0	2	-5**
35*	*Visual tools do not work at all in the learning situations.*	-2	-1	-1	-2
36*	*Knowledge about this disorder is important regarding the academic adaptation.*	4	3	3	5
37	It is easy to put academic demands on this student.	0	-1	-3	0
38	It is hard to follow the academic demands from the reports and IEP.	-2	-2	1	2
39*	*There is a lot of knowledge regarding this disorder and academic functioning.*	-1	-2	-1	-3
40	The most important knowledge on how to work with this student have I gotten through my own practise in the school.	4	1	4	2

Note. Statements in italics are consensus statements, statements flagged with * are non-significant at $p<0.01$, and statements flagged with ** are also non-significant at $p<0.05$ (statements not in italic are distinguishing statements (variance across factor z-scores). Factor scores flagged with * are significant distinguishing at $p<0.05$ and factor scores flagged with a ** are significant distinguishing at $p<0.01$.

9 Appendix 1-7

Appendix 1 – Factor loadings for the Q studies

	Study 1- Experience and coping of challenging behaviours		Study 2 - How to handle the challenging behaviours			
Staff Code	Factor 1 – Managing	Factor 2 - Struggling	Factor 1 – In control	Factor 2 - Struggling	Factor 3 – Strugglers relying on parents	Factor 4 - Support dependent
1TF	0.2673	0.5504X	0.0603	0.2158	0.7750X	0.4302
5TF	0.7113X	0.3926	0.3449	0.2000	-0.0779	0.8450X
4TM	0.7095X	0.2106	0.4880	0.3829	0.3033	0.1459
3UF	0.4205	0.7488X	0.3449	0.8222X	0.0944	0.0151
2TAM	0.8568X	0.2148	0.8490X	0.0420	0.1349	0.1485
6TF	0.6856X	0.4224	0.2975	0.1581	0.4541	0.6755X
7TAF	0.2871	0.6258X	-0.0516	0.8474X	0.2095	0.1941
8TAM	0.8100X	0.2290	0.7099X	0.2096	0.2357	0.3610
9TM	0.7064X	0.1447	0.5238	0.3532	0.6378X	0.0377
10TM	0.8043X	0.2794	0.6391X	0.4363	0.3668	0.2837
11UM	0.8387X	0.2205	0.7408X	0.2674	0.1892	0.3456
12OM	-0.0207	0.7438X	0.2565	0.5779X	0.2173	0.3914
13OM	0.6232X	0.3637	0.8595X	0.0797	0.1919	0.1495
14TF	0.6788X	0.1271	0.3803	0.1510	0.8238X	-0.0950

Note. T: Teacher, TA: Teacher Assistant, O: Other school education, U: Unknown profession, F: Female student with SMS, M: Male student with SMS

Appendix 2 – Translated statements Q study 1 (Article IV)

No	Norwegian	English
1	Eleven har en moden emosjonell utvikling (oppfører seg som andre på sin alder).	The student has a mature emotional development (behaves similar to other people his/her own age).
2	Eleven sitter ofte for seg selv og i ro.	The student often sits quietly by him/herself.
3	Eleven har mye kreativitet.	The student has a lot of creativity.
4	Eleven har god humor.	The student has good humour.
5	Eleven er sjelden trøtt og sliten på dagtid	The student is rarely sleepy and tired during the day.
6	Eleven skader seg selv ved å slå seg, klore seg eller bite seg.	The student self-injures by hitting, scratching, or biting.
7	Eleven selvstimulerer eller har repeterende atferd (spinner eller trykker på ting gjentatte ganger, gjentatte spørsmål).	The student self-stimulates or has repetitive behaviour (spins or pushes on things repeatedly or asks the same questions repeatedly).
8	Eleven er høflig og sosial	The student is polite and social.
9	Av og til er jeg veldig lei jobben min.	Sometimes, I am really tired of my job.
10	Eleven er god på tekniske ting.	The student is good at technical things.
11	Eleven er god på «splitt og hersk» teknikker.	The student has mastered “conquer and divide” techniques.
12	Eleven viser ikke omsorg for andre (elever og voksne).	The student does not care for other people (students or adults).
13	Jeg opplever det som helt greit å bli spyttet på.	I think it is just ok to be spat on.
14	Jeg synes det er krevende å være mye alene med eleven.	I think it is demanding to be alone with the student.
15	Eleven har god impulskontroll.	The student has good impulse control.
16	Eleven er dårlig på å hjelpe til.	The student does not help other people.
17	Eleven har en stygg og krenkende ordbruk.	The student has bad and insulting language.

18	Eleven fungerer godt sammen med jevnaldrende.	The student works well with other students of the same age.
19	Jeg har ikke erfart at eleven har blitt sint på skolen.	I have not experienced the student getting angry at school.
20	Eleven kan oppleves som veldig intens.	The student can be experienced as very intense.
21	Eleven vet hvilke «knapper» de skal trykke på.	The student knows which “buttons” to push.
22	Eleven er sjelden redd i nye situasjoner og i møte med nye mennesker.	The student is rarely afraid in new situations and when meeting new people.
23	Jeg blir aldri provosert av eleven.	I never get provoked by the student.
24	Jeg opplever at det er vanskelig å avlede eleven over til noe annet.	I experience that it is difficult to divert the student’s attention to something else.
25	Eleven har veldig jevnt humør.	The student has a stable mood.
26	Eleven jobber konsentrert med skolearbeidet over tid	The student works with and concentrates on academic work over time.
27	Eleven blir aldri sint og skriker, sparker eller slår.	The student never gets angry with screaming, kicking and hitting.
28	Eleven samarbeider veldig godt med andre elever på gruppeoppgaver.	The student cooperates well with other students in group activities.
29	Jeg er usikker på hva jeg skal gjøre i vanskelige situasjoner.	I am not sure what to do in difficult situations.
30	Det er utfordrende på en god måte å arbeide med denne eleven.	Working with the student is challenging in a good way.
31	Eleven er lite opptatt av de voksne.	The student is not preoccupied with adults.
32	Jeg vet hva jeg skal gjøre når eleven skader seg selv.	I know what to do when the student self-injures.
33	Jeg synes det er vanskelig å se på når eleven skader seg selv.	I think it is difficult to watch when the student self-injures.
34	Eleven ødelegger aldri ting på skolen.	The student never destroys things at school.
35	Eleven har god langtidshukommelse.	The student has good long-term memory.

36	Det er positivt å arbeide med denne eleven.	It is positive to work with the student.
37	Eleven krever konstant oppmerksomhet.	The student demands constant attention.
38	Jeg vet hva jeg skal gjøre om eleven viser utagerende atferd.	I know what to do if the student displays challenging behaviour.
39	Jeg synes det er skremmende når eleven blir sint eller mister kontrollen.	I think it is scary when the student gets angry or loses control.
40	Jeg synes det er enkelt å forklare andre lærere hvordan eleven er og hva de skal gjøre.	I think it is easy to explain to the other teachers about the student and how to handle situations.

Appendix 3 – Translated statements Q study 2 (Article V)

No	Norwegian	English
1	Denne eleven trenger ikke forberedelser for at aktiviteten skal fungere.	This student does not need preparation before an activity to make it work.
2	Jeg bruker ofte individuelle støttende samtaler i forkant og etterkant av utfordrende atferd.	I often use individual support conversations before and after challenging behaviour.
3	Vanlig pedagogikk/ Spesial-pedagogikk fungerer godt på denne eleven.	Regular pedagogical/special education tools work well with this student.
4	Vi på skolen kan veilede og hjelpe foreldrene i forhold til atferd.	The school staff can give guidance and help parents regarding behaviour.
5	Det er lett å vite hva vi skal velge ut når det gjelder skolefag.	It is easy to make choices regarding the academic work.
6	Vi mangler struktur som kunne være med på å forebygge atferdsvansker.	We are lacking a structure that could prevent challenging behaviour.
7	Foreldrene er mer opptatt av det sosiale enn det faglige.	The parents are more preoccupied with the social rather than the academic.
8	Jeg mangler opplæring om diagnosen.	I lack training regarding this disorder.
9	Det er foreldrene som har gitt oss informasjon om diagnosen.	It is the parents that have given us information regarding this disorder.
10	Det er vanskelig å følge de anbefalingene vi har fått vedrørende atferden.	It is difficult to follow the recommendations we have been given regarding the behaviour.
11	Jeg vet hvordan jeg skal arbeide med det faglige med denne eleven.	I know how to work academically with this student
12	Ved å ligge i forkant av mulige vanskelige situasjoner kan vi unngå mange problemer.	We may avoid many problems by being ahead of possible tricky situations.
13	Jeg kan arbeide med fag for denne eleven på samme måte som med alle andre elever.	I can work academically with this student the same way I do with all other students.
14	Det kreves tydelige grenser for å kunne få til noe faglig.	It needs clear boundaries to be able to get any academically work done.
15	Den utfordrende atferden gjør det vanskelig å ha fokus på fag.	It is the challenging behaviour that makes it difficult to have focus on the academic

16	Jeg har gode verktøy for å forhindre atferdsvanskene.	I have good tools to prohibit the challenging behaviour.
17	Foreldrene har for høye forventninger til skolefaglig fungering og det gjør samarbeidet vanskelig.	The parents have too high expectations regarding the academic work and that makes the cooperation with them difficult.
18	Jeg får god oppfølging etter vanskelige situasjoner med eleven.	I get a good follow-up after difficult situations with this student.
19	Kunnskap om diagnosen er ikke spesielt viktig for å tilrettelegge for denne eleven.	It is not important to have knowledge regarding this disorder in order make adaptation to this student.
20	Jeg synes det er vanskelig å arbeide med denne eleven på grunn av atferden.	I think it is hard to work with this student because if the behaviour.
21	Jeg vet hva jeg skal gjøre om eleven skader seg selv.	I know what I should do if the student self-injures.
22	Foreldrene er vanskelige å samarbeide med fordi eleven oppfører seg ulik hjemme og på skolen.	The parents are difficult to cooperate with because the student behaves different at home and at the school.
23	Jeg får god støtte fra foreldrene og liker å snakke med dem.	I get good support from the parents and like to talk with them.
24	Jeg synes det er krevende å samarbeide med foreldrene til denne eleven.	I think it is challenging to cooperate with the parents of this student.
25	Jeg er redd når eleven blir sint og skriker, sparker eller slår.	I am afraid when the student gets angry and is screaming, kicking, or hitting.
26	Det er vanskelig når foreldrene blir lei seg for tilbakemeldinger om elevens fungering.	It is hard when the parents get upset regarding feedback about the student's act.
27	Det er vanskelig å få til et samarbeid rundt lekser.	It is hard to get a function cooperation around homework.
28	Jeg får god hjelpe å støtte fra ledelsen til å kunne fokusere på det faglige.	I receive good support from the school leadership, so I can focus on the academic work.
29	Det er ikke så viktig for meg at andre kollegaer og ledelsen verdsetter/skryter av arbeidet mitt.	It is not important to me that my colleagues and leadership at the school gives me praise and value my work.

30	Jeg har fått veiledning i hvordan jeg skal håndtere vanskene til eleven og opplever det som trygt.	I have been given guidance in how to handle this student’s challenges and feel safe.
31	Samarbeidet med foreldrene er viktig for tilretteleggingen.	Cooperation with the parents are important regarding the adaptation in the school.
32	Jeg synes det er krevende å jobbe med fortvilte og bekymrede foreldre.	I think it is hard to work with parents in despair and full of worry.
33	Eleven viser mye sjarme, humor og kjærlighet så jeg gleder meg til hver dag.	I am looking forward to every day because of this student’s charm, humour, and love
34	Det er foreldrene som har lært oss hvordan vi skal håndtere atferden.	It is the parents that have taught us how to handle the behaviour.
35	Visuelle hjelpemidler fungerer veldig dårlig for læring.	Visual tools do not work at all in the learning situations.
36	Kunnskap om diagnosen er viktig for å tilrettelegge det faglige.	Knowledge about this disorder is important regarding the academic adaptation.
37	Det er enkelt å stille faglige krav til eleven.	It is easy to put academic demands on this student.
38	Det er vanskelig å følge de faglige kravene fra sakkyndig vurdering/IOP.	It is hard to follow the academic demands from the reports and IEP.
39	Det finnes mye kunnskap om skolefaglig fungering angående denne diagnosen.	There is a lot of knowledge regarding this disorder and academic functioning.
40	Den viktigste kunnskapen om hvordan arbeide med denne eleven har jeg fått gjennom egen praksis i skolen.	The most important knowledge on how to work with this student have I gotten through my own practise in the school.

Appendix 4 – Background information form for school staff

Norwegian	English
Navn på skolen:	Name of the school
Stilling på skolen (assistent, kontaktlærer, spes.ped, etc):	Position at school (teaching assistant, teacher, special education teacher, etc)
Kjønn (på den som fyller ut skjemaet):	Gender (of the person filling the form)
Utdanningsnivå (høyskole, fagbrev, etc):	Educational level (High School Diploma, university, etc)
Alder/klassetrinn for eleven med SMS:	Grade level/age of the student with SMS
Hvor lenge har du arbeidet med/kjent akkurat denne eleven:	How long have you known/worked with this student?
Kommentarer	Comments

Appendix 5 – Easy to read consent form (Norwegian)

SMS Kartleggingsstudie Mars 2015

Vil du være med i en undersøkelse?

”Smith-Magenis` syndrom – En kartleggingsstudie”

Hva skal vi undersøke?
Frambu vil finne ut mer om Smith-Magenis` syndrom. Det er derfor vi sender deg dette brevet.

Hva skjer i undersøkelsen?
Alle som blir med vil få en pakke med spørreskjemaer. Disse skjemaene er det foreldrene dine (eller andre som står deg nær) som skal fylle ut. I tillegg ber Frambu om tillatelse til:

- hente inn informasjon om diagnosen din fra sykehuset der den ble stilt og informasjon fra andre helseinstitusjoner der du har vært.
- At en lege og en fysioterapeut undersøker deg og hva du kan om du kommer til Frambu på kurs.
- ta en blodprøve fra armen din om du kommer til Frambu på kurs(for å finne ut om du har noen helseproblemer eller har risiko for å få det)
- ta prøver av spyttet ditt om du kommer til Frambu på kurs.

Du vil også få tilsendt et lite apparat som du skal feste på armen din i 14 dager (som en klokke). Apparatet måler hvor mye du går og beveger deg når du er hjemme og hvordan du sover. Etter de 14 dagene, sender du apparatet til Frambu. Du får med en konvolutt med adresse og frimerke når du mottar apparatet.

Fordeler og ulemper
Mesteparten av undersøkelsen er det foreldrene dine som må gjøre noe med.

Blodprøven vi tar er helt vanlig med et stikk i armen. Noen ganger får man et lite blåmerke der stikket har vært. Spyttprøvene tas ved at du får en bomullspinne inn i munnen. Dette gjør ikke vondt. I undersøkelsen av lege og fysioterapeut blir du bedt om å gjøre forskjellige ting (løpe, hoppet, etc), dette vil ikke gjøre vondt.

Hvordan passer vi på det vi får vite om deg?
Det vi får vite om deg i undersøkelsen vil ikke bli brukt til andre ting enn å finne ut mer om Smith-Magenis` syndrom.
All informasjon og prøvesvar blir tatt vare på uten navn, så ingen andre enn de som gjør undersøkelsen kan finne ut hvem de handler om.

Etter at undersøkelsen er ferdig, skriver vi om hva vi fant ut i ett eller flere blader. Vi skriver ikke hvem som har vært med i undersøkelsen..

Du velger selv om du vil være med eller ikke
Det er frivillig å delta. Hvis du blir med, kan du ombestemme deg når du vil uten noen spesiell grunn. Uansett hva du bestemmer, vil det ikke forandre andre tilbud fra Frambu. Hvis du vil være med i undersøkelsen, må du skrive under på skjemaet "Jeg vil være med i undersøkelsen Smith-Magenis` syndrom - En kartleggingsstudie".

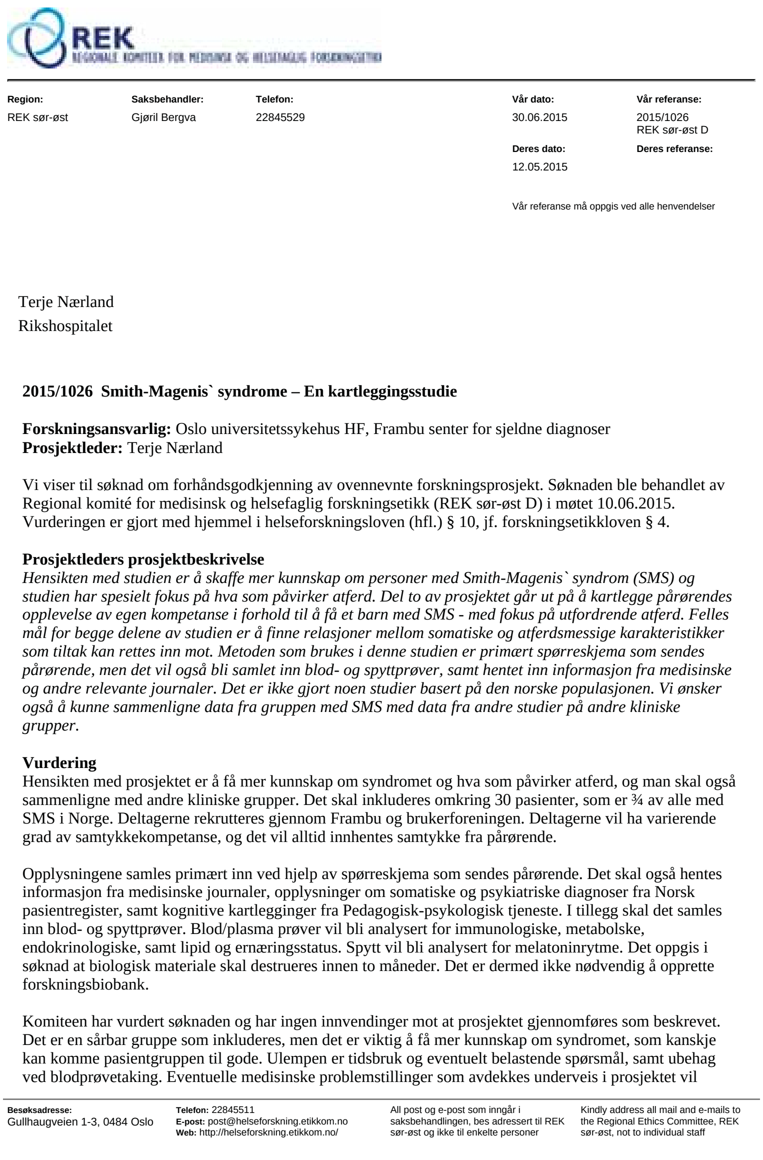
Vil du vite mer?

- Kontakt Frambu ved Spesialpedagog Heidi E. Nag på e-post hel@frambu.no eller telefon 64 85 60 00 eller NevSom ved Psykolog Terje Nærland, e-post ternae@ous-hf.no
- Du kan du lese mer om undersøkelsen i vedlegget som heter "Forespørsel om deltakelse i forskningsprosjektet"

Hvis du vil være med i undersøkelsen, må du fylle ut og sende til Frambu skjemaene:

- Jeg vil være med i undersøkelsen « Smith-Magenis` syndrom - En kartleggingsstudie »

Appendix 6 – Regional Ethical Committee (1)



REK
REGIONALE KOMITEER FOR MEDISINSK OG HELSEFAGLIG FORSKNINGSETIKK

Region:	Saksbehandler:	Telefon:	Vår dato:	Vår referanse:
REK sør-øst	Gjøril Bergva	22845529	30.06.2015	2015/1026 REK sør-øst D
			Deres dato:	**Deres referanse:**
			12.05.2015	

Vår referanse må oppgis ved alle henvendelser

Terje Nærland
Rikshospitalet

2015/1026 Smith-Magenis` syndrome – En kartleggingsstudie

Forskningsansvarlig: Oslo universitetssykehus HF, Frambu senter for sjeldne diagnoser
Prosjektleder: Terje Nærland

Vi viser til søknad om forhåndsgodkjenning av ovennevnte forskningsprosjekt. Søknaden ble behandlet av Regional komité for medisinsk og helsefaglig forskningsetikk (REK sør-øst D) i møtet 10.06.2015. Vurderingen er gjort med hjemmel i helseforskningsloven (hfl.) § 10, jf. forskningsetikkloven § 4.

Prosjektleders prosjektbeskrivelse
Hensikten med studien er å skaffe mer kunnskap om personer med Smith-Magenis` syndrom (SMS) og studien har spesielt fokus på hva som påvirker atferd. Del to av prosjektet går ut på å kartlegge pårørendes opplevelse av egen kompetanse i forhold til å få et barn med SMS - med fokus på utfordrende atferd. Felles mål for begge delene av studien er å finne relasjoner mellom somatiske og atferdsmessige karakteristikker som tiltak kan rettes inn mot. Metoden som brukes i denne studien er primært spørreskjema som sendes pårørende, men det vil også bli samlet inn blod- og spyttprøver, samt hentet inn informasjon fra medisinske og andre relevante journaler. Det er ikke gjort noen studier basert på den norske populasjonen. Vi ønsker også å kunne sammenligne data fra gruppen med SMS med data fra andre studier på andre kliniske grupper.

Vurdering
Hensikten med prosjektet er å få mer kunnskap om syndromet og hva som påvirker atferd, og man skal også sammenligne med andre kliniske grupper. Det skal inkluderes omkring 30 pasienter, som er ¾ av alle med SMS i Norge. Deltagerne rekrutteres gjennom Frambu og brukerforeningen. Deltagerne vil ha varierende grad av samtykkekompetanse, og det vil alltid innhentes samtykke fra pårørende.

Opplysningene samles primært inn ved hjelp av spørreskjema som sendes pårørende. Det skal også hentes informasjon fra medisinske journaler, opplysninger om somatiske og psykiatriske diagnoser fra Norsk pasientregister, samt kognitive kartlegginger fra Pedagogisk-psykologisk tjeneste. I tillegg skal det samles inn blod- og spyttprøver. Blod/plasma prøver vil bli analysert for immunologiske, metabolske, endokrinologiske, samt lipid og ernæringsstatus. Spytt vil bli analysert for melatoninrytme. Det oppgis i søknad at biologisk materiale skal destrueres innen to måneder. Det er dermed ikke nødvendig å opprette forskningsbiobank.

Komiteen har vurdert søknaden og har ingen innvendinger mot at prosjektet gjennomføres som beskrevet. Det er en sårbar gruppe som inkluderes, men det er viktig å få mer kunnskap om syndromet, som kanskje kan komme pasientgruppen til gode. Ulempen er tidsbruk og eventuelt belastende spørsmål, samt ubehag ved blodprøvetaking. Eventuelle medisinske problemstillinger som avdekkes underveis i prosjektet vil følges opp, og deltagere som opplever ubehag ved utfylling av spørreskjemaer og får reaksjoner kan kontakte prosjektmedarbeider. Komiteen finner at prosjektet er forsvarlig å gjennomføre.

Komiteen har noen kommentarer til informasjonsskrivet:
-Kapittel A om spørreskjema i skrivet til pårørende er veldig detaljert. Det kan i stedet angis antall spørreskjema og hva de handler om, i stedet for detaljert gjennomgang av alle.
-Komiteen gjør oppmerksom på at barn under 16 år ikke er samtykkekompetente i lovens forstand. Det skal derfor ikke legges opp til at disse selv samtykker. De skal kun få informasjon. Skrivene må revideres i henhold til dette.
-Det står i skrivet til pårørende at avidentifiserte opplysninger vil bli lagret i ytterligere 5 år for videre forskning. Hva denne videre forskningen går ut på er ikke beskrevet, hverken i søknad eller protokoll. Komiteen ber derfor om at setningen strykes. Komiteen tillater at avidentifiserte opplysninger oppbevares i 5 år etter prosjektslutt, men dette er av dokumentasjonshensyn og for etterkontroll.

På denne bakgrunn setter komiteen følgende vilkår for godkjenning:
-Informasjonsskrivene skal revideres i tråd med komiteens kommentarer og sendes inn til orientering.

Vedtak
Med hjemmel i helseforskningsloven § 9 jf. 33 godkjenner komiteen at prosjektet gjennomføres under forutsetning av at ovennevnte vilkår oppfylles.

I tillegg til vilkår som fremgår av dette vedtaket, er godkjenningen gitt under forutsetning av at prosjektet gjennomføres slik det er beskrevet i søknad og protokoll, og de bestemmelser som følger av helseforskningsloven med forskrifter.

Tillatelsen gjelder 31.12.2019. Av dokumentasjonshensyn skal opplysningene likevel bevares inntil 31.12.2024. Forskningsfilen skal oppbevares avidentifisert, dvs. atskilt i en nøkkel- og en opplysningsfil. Opplysningene skal deretter slettes eller anonymiseres, senest innen et halvt år fra denne dato.

Forskningsprosjektets data skal oppbevares forsvarlig, se personopplysningsforskriften kapittel 2, og Helsedirektoratets veileder for «Personvern og informasjonssikkerhet i forskningsprosjekter innenfor helse og omsorgssektoren».

Dersom det skal gjøres vesentlige endringer i prosjektet i forhold til de opplysninger som er gitt i søknaden, må prosjektleder sende endringsmelding til REK.

Prosjektet skal sende sluttmelding på eget skjema, senest et halvt år etter prosjektslutt.

Klageadgang
REKs vedtak kan påklages, jf. forvaltningslovens § 28 flg. Klagen sendes til REK sør-øst D. Klagefristen er tre uker fra du mottar dette brevet. Dersom vedtaket opprettholdes av REK sør-øst D, sendes klagen videre til Den nasjonale forskningsetiske komité for medisin og helsefag for endelig vurdering.

Vi ber om at alle henvendelser sendes inn på korrekt skjema via vår saksportal: http://helseforskning.etikkom.no. Dersom det ikke finnes passende skjema kan henvendelsen rettes på e-post til: post@helseforskning.etikkom.no.

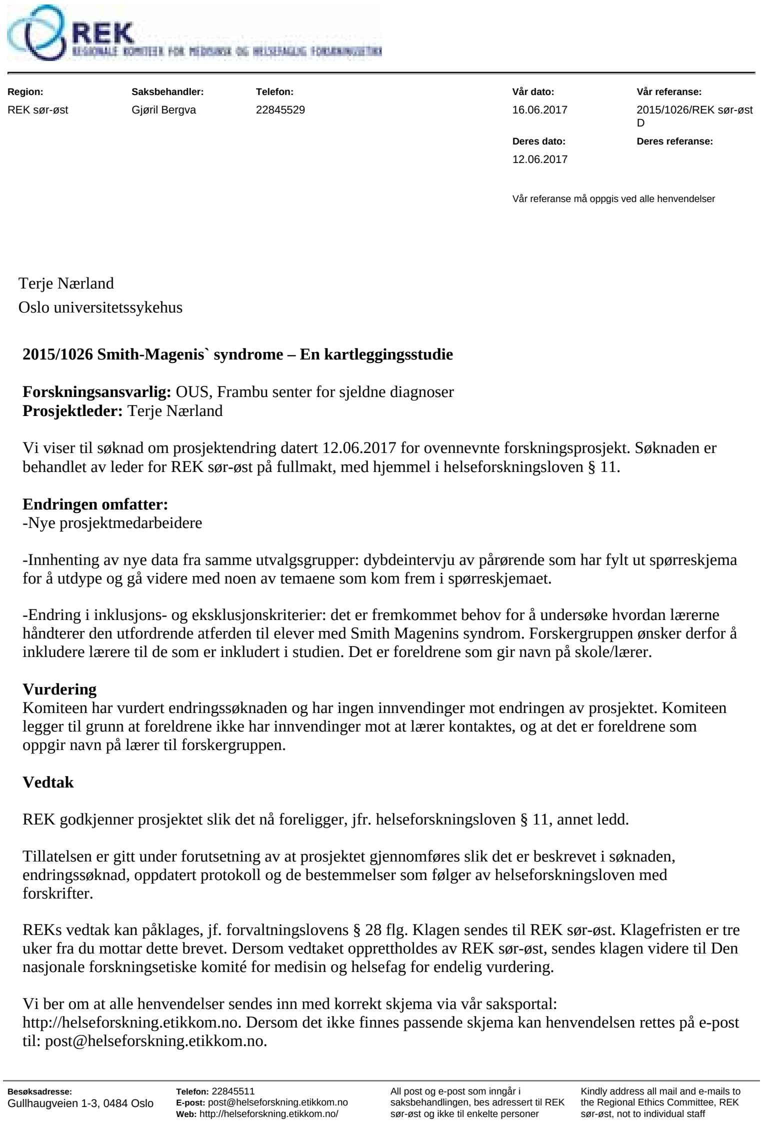
Vennligst oppgi vårt referansenummer i korrespondansen.

Med vennlig hilsen

Finn Wisløff
Professor em. dr. med.
Leder

Gjøril Bergva
Rådgiver

Appendix 7 – Regional Ethical Committee (2)



REK
REGIONALE KOMITEER FOR MEDISINSK OG HELSEFAGLIG FORSKNINGSETIKK

Region:	**Saksbehandler:**	**Telefon:**	**Vår dato:**	**Vår referanse:**
REK sør-øst	Gjøril Bergva	22845529	16.06.2017	2015/1026/REK sør-øst D
			Deres dato:	**Deres referanse:**
			12.06.2017	

Vår referanse må oppgis ved alle henvendelser

Terje Nærland
Oslo universitetssykehus

2015/1026 Smith-Magenis` syndrome – En kartleggingsstudie

Forskningsansvarlig: OUS, Frambu senter for sjeldne diagnoser
Prosjektleder: Terje Nærland

Vi viser til søknad om prosjektendring datert 12.06.2017 for ovennevnte forskningsprosjekt. Søknaden er behandlet av leder for REK sør-øst på fullmakt, med hjemmel i helseforskningsloven § 11.

Endringen omfatter:
-Nye prosjektmedarbeidere

-Innhenting av nye data fra samme utvalgsgrupper: dybdeintervju av pårørende som har fylt ut spørreskjema for å utdype og gå videre med noen av temaene som kom frem i spørreskjemaet.

-Endring i inklusjons- og eksklusjonskriterier: det er fremkommet behov for å undersøke hvordan lærerne håndterer den utfordrende atferden til elever med Smith Magenins syndrom. Forskergruppen ønsker derfor å inkludere lærere til de som er inkludert i studien. Det er foreldrene som gir navn på skole/lærer.

Vurdering
Komiteen har vurdert endringssøknaden og har ingen innvendinger mot endringen av prosjektet. Komiteen legger til grunn at foreldrene ikke har innvendinger mot at lærer kontaktes, og at det er foreldrene som oppgir navn på lærer til forskergruppen.

Vedtak

REK godkjenner prosjektet slik det nå foreligger, jfr. helseforskningsloven § 11, annet ledd.

Tillatelsen er gitt under forutsetning av at prosjektet gjennomføres slik det er beskrevet i søknaden, endringssøknad, oppdatert protokoll og de bestemmelser som følger av helseforskningsloven med forskrifter.

REKs vedtak kan påklages, jf. forvaltningslovens § 28 flg. Klagen sendes til REK sør-øst. Klagefristen er tre uker fra du mottar dette brevet. Dersom vedtaket opprettholdes av REK sør-øst, sendes klagen videre til Den nasjonale forskningsetiske komité for medisin og helsefag for endelig vurdering.

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Vennligst oppgi vårt referansenummer i korrespondansen.

Med vennlig hilsen

Finn Wisløff
Professor em. dr. med.
Leder

Gjøril Bergva
Rådgiver

Kopi til: *Oslo universitetssykehus HF ved øverste administrative ledelse: oushfdlgodkjenning@ous-hf.no; Frambu ved øverste administrative ledelse: info@frambu.no; UXODBA@ous-hf.no; Kjetil.Orbeck@frambu.no*