The health and socio- emotional problems of parents with children with sex chromosome aneuploidies, in a social- ecological point of view.

Summary:

This master thesis consists of two papers: the first is theoretical and the second is empirical. The introduction section presents a theoretical background and an explanation of methodology decisions with rationales, and the thesis concludes with a final summary. A brief description of the two articles:

Paper 1: Understanding of disability and coping strategies of parents with children with disabilities, in a social- ecological perspective

Perceptions of what constitutes the term “disability” and the description of who are considered disabled will vary from one historical era to another, and also from one community to another within the same era. This paper presents some of the welfare- state developments which as influenced ways of thinking and assessing understandings of “disability” and “impairment”. In light of Antonovsky’s theory of salutogenesis, this paper explores the stress dimensions and factors that parents of children with disabilities are exposed to, and how ‘sense of coherence’ and coping strategies are key strategies in managing difficult life situations. Life situations of parenting a child with a disability may have a negative impact on parents’ sense of coherence. At the same time, different coping strategies may help retain parents’ health, and prevent the occurrence of stress and diminished psychological health.

Paper 2: The health and socio- emotional problems of parents with children with sex chromosome aneuploidies

The purpose of this empirically-based study is to augment our knowledge about psycho-social health among parents of boys with sex chromosome disorders. Using a quantitative methodological approach, this paper explores and examines some of the challenges parents and children with sex chromosome aneuploidies have. Previous research of boys with sex chromosome aneuploidies (SCA) shows a wide range of problems, both socially, cognitively and psychologically. With the first paper as a theoretical theory foundation, this second paper shows that parenting a child with SCA may affect parents’ sleep quality, and that there is a correlation in poor social and emotional functioning in children as well as diminished mental health in parents. Poor sleep quality is also connected with low personal well-being scores, high scores in health complaints and a high number of days with health complaints. Findings also show that mothers report lower scores regarding health, satisfaction in life and sleep quality than fathers. Mothers of boys with SCA especially are at greater risk in experiencing stress symptoms, reporting less satisfaction of life and lower life quality measures.

Preface:

This thesis is part of a two- year Master program in Disability and Society at the Department of Social Work and Health Sciences (ISH) at the Norwegian University of Science and Technology (NTNU) in Trondheim. Paper II in the thesis is written in collaboration with Frambu, a national competence center for rare disorders. I would like to give special thanks to psychologist Krister Fjermestad at Frambu for providing me with the opportunity to join the research there concerning social-emotional difficulties in children with sex chromosome aneuploidies and their parents. Thank you for your patience, your feedback and encouragement.

I would like to thank all the informants who have taken the time to participate in this study, and shared so much of their lives.

Thanks to my supervisor Kristjana Kristiansen for your support and feedback, especially in the final phases. I would also like to give my thanks to my friends and family for the support and encouragement, and wonderful help with SPSS and EndNote among other challenges I have had during this thesis- work.

Overview

Summary: iii

Paper 1: Understanding of disability and coping strategies of parents with children with disabilities, in a social- ecological perspective iii

Paper 2: The health and socio- emotional problems of parents with children with sex chromosome aneuploidies iii

Preface: v

Introduction: 1

About disability 1

About sexual chromosome aneuploidy: 2

Research questions 3

Theoretical background 4

Logical positivism 4

Social constructivism. 5

How real is the reality in research? 6

Salutogenesis: 6

Pre- understanding: 7

Methodological discussion 9

Research sample 9

Questionnaires 10

Validity and reliability 11

References: 12

Paper I: 15

Understanding disability and coping strategies of parents with disabled in a socio- ecological perspective 15

Ecological theory 18

A change in disability history 19

Normalization 19

What is a ‘disability’? 20

Quality of life 22

A vulnerable adolescence 23

Stigma and devaluation in a vulnerable situation 24

Parent’s coping 25

Social services and support 26

Finding a sense of coherence 29

Stressors and coping 30

Final comments 32
References: 33
Paper II: 35
The health and social- emotional problems of parents with children with sex chromosome aneuploidies 35
Introduction 37
Antonovsky’s sense of coherence 37
Sex chromosome aneuploidy 38
Klinefelter syndrome 39
Parents’ coherence 41
Main questions 42
Methods: 43
Sample: 45
Discussion 49
References: 52
Challenges and ethical considerations 55
Final comments 57
Attachments: 59

Introduction:

A family is not an isolated entity, but a complicated system with several components that work together. These components change and develop together with interactive and mutual influences. In this thesis, I look at the family as such a system, and scrutinize what happens when one of the components does not work as one might have expected. This line of thinking is inspired by ecological theory, based on general system theory, and developed with dynamic and humanistic dimensions (Klefbeck & Ogden, 1995). An ecological perspective concerning families with disabled children means that the family has an active and important role in the development of children and throughout adolescence.

In the following part, I present the theoretical background for this thesis, including professional and personal perspectives. This background information is intended to be a theoretical foundation for both papers, as well as a framework to create coherence between the two papers Additionally, I will explain my choices of methodological approaches and theory-choices. Finally, in the last section, I will summarize my results and offer some concluding remarks.

About disability

Disability is an umbrella term, resulting from the social consequences of impairments of a physical, sensory, emotional, developmental, or cognitive nature, or a combination of these. Impairments are a problem located in bodily functions or structures. An impairment may be congenital (occurring at or before birth), or acquired later in life (such as through disease or accident). The typical way of assessing the degree of impairment ranges from 'mild to moderate, severe, or profound'. When diagnosed with a combination of impairments, one often uses terms such as "multiply- impaired/disabled".

Subsequent limitations in activity and participation restrictions are these days generally considered a disability as a result of actual impairments. An activity limitation is a constraint encountered by someone when executing a task or activity, while a participation restriction is a problem experienced by an individual in involvement in life situations (World Health Organization, 2001).

In Norway, the understandings and definitions of disability have developed and changed synchronously with welfare state developments. One advantage of being diagnosed and labelled as impaired or disabled is that such classifications are coupled with special rights, such as financial benefits, practical assistance, and technical aids. On the other hand, the word "disability" also relates to issues of discrimination and societal prejudice. The use of the words “impaired” or “disabled” includes persons with reduced functioning that is both located in the individual, as well as created and maintained by societal or physical obstacles and barriers for marginalized groups (Tøssebro, 2009).

About sexual chromosome aneuploidy:

Chromosome abnormality is in many cases a condition that causes certain needs, or behaviors that create what we might call “expectation gaps” between the child and the majority of children, and that reduces the child's functioning in physical, mental or social ways.

Sex chromosome aneuploidy (SCA) is the term used to describe a group of chromosomal disorders in which individuals are born with an atypical number of sex chromosomes (X and Y),(Visootsak & Tartaglia, 2013). Normally, one’s genetic material is distributed across 46 chromosomes, 44 autosomes and 2 sex chromosomes. For girls, it is described as 46 XX, and 46 XY for boys. A missing or additional sex chromosome leads to a variety of conditions. When an oocyte (egg) is fertilized with an additional X chromosome causing for example a combination of 47 XXY, Klinefelter Syndrome will occur. The extra X chromosome can come from either the sperm or the egg (Sokol, 2012).

SCA conditions are estimated to occur in 1 in 400 births, making them the most common chromosomal abnormalities in humans (Visootsak & Tartaglia, 2013). In comparison, Down syndrome occurs in about 1 in 690 births (National Association of Down Syndrome).

About 80% of men have the most common combination 47, XXY, known as Klinefelter syndrome. The remaining 20% have other combinations with more X or Y’s (Makowski, 2005). The different combinations result in different symptoms, varying from individual to individual.

Because of the variety in symptoms, many men are not diagnosed until approaching or reaching adulthood, or never at all. For people with Klinefelter syndrome, decreased production of testosterone is a common sign in diagnosis, and many men are diagnosed when attempting yet failing to reproduce. Many men with SCA show various physical conditions, such as increased body height, wider hips and narrow shoulders. Poorer coordination, dexterity, running and motor abilities, lower muscle tone and strength, synkinetic movements, and tremors are often present and usually identified in early childhood (Sokol, 2012). In paper II, I describe the different karyotypes and symptoms more closely, and examine some results of the functioning of children with SCA and their parents.

In paper I in this thesis, the content of the term “disability” is based on the definition from the World Health Organization, and include both persons with physically or psychological impairments of any kind. There is an enormous variety in types of disabilities, but common for all, is that parents’ feelings, reactions and coping strategies when having a disabled child are very similar. Most parents have to reorganize their lives, establish contacts with social supports, and provide comprehensive care for a child with poor health or impairments, regardless of a diagnosis. Paper II examines these issues, specifically for parents of children with sexual chromosome aneuploidies.

Research questions

The fundamental issue for this thesis is to discover and scrutinize how parenting an impaired child affects parents, and explore and examine which factors affect parent’s function and their perception of quality of life. The thesis consists of two papers that with different perspective and methods will elucidate these questions.

Paper I; “Understanding of disability and coping strategies of parents with children with disabilities, in a socio- ecological perspective”, is a theoretical literature study based on articles found on NTNU’s online library search, and literature provided from NTNU library. I also have found inspiration for sources to search for after reading articles and other papers on these subjects. This article focus on how a child’s impairment and resultant disability affect parents’ health and daily life functioning, and how parents manage the family life situation.

Paper II; “The health and social- emotional problems of parents with children with sex chromosome aneuploidies” is an empirical study about parents of boys with sexual chromosome aneuploidies. I have used a quantitative method to obtain information about parents and their life situations. By using quantitative methods, it is easier to compare the experiences of families with boys with SCA, and provide clearer insights as to whether there are differences within groups of chromosomal abnormalities, the parents, as well as variables such as education, age, or geographical location. The research questions in this study have been:

- How do parents of boys with SCA rate their subjective health, sleep quality and quality of life?
- Is the parents’ mental and physical health related to the functioning of their child?

My explanation for choice of methodology and analysis tools for Paper II will be presented in its own methodology chapter, as well will choice of theory to support Paper II be presented in an own theory chapter. Furthermore, Paper I is considered a theoretical foundation for Paper II.

I will start this thesis with a theoretical introduction about science in disability care, how we understand and discover find science, and important guidelines for scientists in social work

Theoretical background

Logical positivism

The logical positivism that occurred in Vienna in the 1920s initiated the start of the field of modern philosophy of science. Logical empiricists emphasized that science begins with observations of reality, and their mission is then to renovate all of philosophy and convert it to a new scientific philosophy. Scientific knowledge is thus a set of general statements or theories generalized from observations in empirical research (Ringdal, 2013).

The positivist will focus on explaining social phenomena. Like the physicist, one should look for reasons. Explanations should be shown in terms that indicate cause and effect, and knowledge should be able to be expressed in regularity and 'laws' of nature. Within this naturalistic, positivistic view of science, knowledge is only based on the positive provable. Naturalism asserts that nature, and everything in it, is the way it appears to be by observation. The positivistic research-stance is objective, and avoids influence from unscientific assessments (Garsjø, 2001). That makes religion, myths and beliefs something humans construct to explain what we do or do not understand.

The "scientific method" is considered fundamental to the scientific investigation and eventual discovery of new knowledge based on physical evidence, so-called empiricism. This scientific method involves many techniques for investigating phenomena, finding new knowledge or coordinating and integrating existing knowledge (Ringdal, 2013).

The knowledge perspective based in critical theory involves a diversion from positivism, and that only results which can be verified empirically can be considered valid truths. All objectivity is not rejected in critical theory, but since the production of knowledge is situational and context-dependent, one must see knowledge development in historical, philosophical and political contexts, and is therefore not a neutral process. Knowledge of social phenomena in particular is considered greatly socially-constructed and value-laden. (Askeland, 2006).

There are two main steps in the scientific method. The first is to collect data through observation and experimentation, and the second to formulate and test hypotheses. This may be followed by more observations or experiments if one needs more data. An important part of such an approach is reporting the results. One of the most common reporting procedures is writing peer-reviewed articles in scientific journals (Ringdal, 2013).

Social constructivism.

The core of social constructivism is that the social reality we know of is constructed and reconstructed through actions and interaction among people. Money, titles, political institutions and organizations are all examples of social constructions. Also morals, cultural norms and religion can be seen as social constructions. This results in a relative reality for each one of us, only reachable for us through our constructed images of reality. The" truth" is relative and culture dependent (Ringdal, 2013).

Natural sciences mainly study physical objects, while the focus of social science research consists of humans as thinking and acting social creatures. Human beings are able to reflect upon their actions and make choices between various options. Nevertheless, humans' actions during the day are largely characterized by routines, and actions are regulated by social procedures, sanctions and norms, morality and legislation. This makes social phenomena to act almost as if they are natural phenomena, as the routines are so habituated in us that reflection often feels unnecessary. It is still important to be aware of not confusing these regular actions as natural phenomena, because humans at any point can decide to act differently and conflict with social laws (Ringdal, 2013).

There is no clear distinction between theory and method: they are rather an integrated duality (Helbæk, 2008). The question of how scientific knowledge and concepts form the foundation for social action and change is central. Scientific theory can help us to clarify the consequences of our earlier, our present and our future practices. It can increase our ability to break with traditionally assumptions and provide openness to new perspectives in social work and disability care (Garsjø, 2001).

Whether it is deliberate or not, disability care have always been colored by different theories and values, and been influenced by prevailing social views. Professionals should constantly question why we act and think as we do, on which assumptions we base our actions and attitudes, and how they exist. The insight it gives, makes it possible to change the practice so that it is in accordance with theories and values we claim as fundamental (Fook, 1996; Morley, 2004). Being a critically reflective professional may lead to changes in attitudes and actions in relation to other people. This may again cause repercussions and eventually contribute to social change (Askeland, 2006).

How real is the reality in research?

Philosophers such as Popper and Skjervheim have delivered convincing arguments that an objective science based on the logical positivistic pattern is not be possible because the researcher will always be affected by the historical period, personal values and theoretical starting points. Still, the scientist does his or her utmost to uncover the circumstances of reality, without considering interest in own benefits, by following recognized instructions and documenting their research. This is a by some considered a necessary condition for scientific research (Ringdal, 2013).

Casual explanation has its place in social science as well as natural science. The expression “casual” belongs in theories and models. Predictions or hypotheses derivating from casual models contribute to build bridges over the gap between theory and reality (Ringdal, 2013).

It is somewhat a paradox that the researcher is the one who controls the data by choosing the variables he or she finds interesting. In that way he or she can manipulate, or act to create a casual explanation, based on former notions or ideas they had in advance. There also might be times it is desirable to find an answer someone considers “correct”. This trend may happen unconsciously for the researcher, and therefore reflection is an important tool to stay true to your research (Ringdal, 2013). The researcher may risk being affected for example by cultural upbringing, beliefs, pressure from employer, personal expectations or other factors that color one’s ways of thinking and analyzing what one observes.

Salutogenesis:

While the psychosocial view presented in paper II focuses on stressors as an inhibitory factor for health, the salutogenesis theory, presented in paper I, focuses on factors promoting health. Medical science has attempted to answer the question “what makes us sick” for a long time. By turning the question around, the Israeli-American sociologist Antonovsky gave us a new view on how good health expresses itself. Good health is not necessarily absence of all that is pathogenic, nor does poor health always relate to conditions of diagnosis and disease. In changing the gaze from the pathogenic and health-risks, Antonovsky focused on personal individuality about resilience and good health (Tamm, 2012).

In a salutogenic perspective, stressors are described as constantly present, yet personal resources and capacity to manage the stressors are considered important. The answer Antonovsky found after empirical research, is that the common characteristics for humans with good health is what is now known as a *sense of coherence* (SOC). This is described as an attitude or a way to approach life (Tamm, 2012).

How we face the small and larger challenges in our daily life is closely combined with how we perceive ourselves. This is also called our self-image or self-perception. How we assess ourselves define who we are, or who we think we are when we face our surroundings (Grue, 2013). When you believe in your strengths, facing problems seems much more possible to approach and overcome. Our surroundings can also have deep impacts in how we develop the image of ourselves. We want to be a part of a group or several groups in society, and we typically adapt our behavior when meeting others, within the cultural norms and situational expectations deemed appropriate. The feedback we get from our significant others and the expectations they have, or may have towards us, also influence our behavior and for example choice of words. The image one has of oneself can change according to what group you compare yourself to, known as your reference group (Garsjø, 2001).

Pre- understanding:

A clarification of my personal and professional pre-understanding is also a necessary part of the background for this thesis. A pre-understanding consists of a person's sum of experiences, beliefs, hypotheses, perspectives, approaches and theoretical reference frameworks, which combined constitute the motivation and engagement for a research project. What follows is some of my pre- understandings and prerequisites for this research master-study.

My younger brother lives with a genetic disability called Duchennes muscular dystrophy, which causes serious physical impairments. This has given me an extended experience in how an impairment affects the family as a whole, and how important objectives such as universal design and individual adaptions is for participation in society. I have also witnessed and experienced the importance of a good support from public services, and communication with its employees.

My educational background started with bachelor degree in social work, focusing on people who need help and support in difficult life situations. This bachelor degree provides basic knowledge and skills for attempting to prevent or solving social problems for families and individuals in neighborhood communities. I have 10 years of experience working with disabled children and adults, and am familiar with the challenges, physically and emotionally related to people with impairments. I have also 2 years of experience with working in the Norwegian Child Welfare Service *('barnevernstjenesten')*, which presented me with close working experiences with families parents in difficult and various life situations. A post-graduuate year of studying "Law and social sciences" at the University-college in Molde made me interested in protecting the legal rights of children and disabled people, and made me choose the master program "Disability and Society" at NTNU. Carrying out social research was new to me when I started this master program. I quickly became interested in the combined way to approach social problems and disability issues, using individual-based explanations combined with socially-constructed and historical explanations. Writing the master thesis in papers and using quantitative methods, is a chance to highlight ways to understand society, disability, and individual inequalities, while using quantitative research makes it possible as a social worker to recognize patterns in a large group of people, and propose measures to improve the situation on an individual level and in social structures.

My interest for social work has been present since I in high school and learned about the legal rights-system and about discrimination of disabled people. My interests have since then developed into how to assist disabled people in the best possible ways to become as independent as possible, and how to support and guide parents through the tangled jungle of social support system, including legal entitlements. In this context, I contacted Frambu, a national resource center for families where someone has a rare impairment, and asked if they had any planned or on-going research projects I could join in connection with my master thesis. Psychologist Krister Fjermestad at Frambu responded positively, and provided me with the opportunity to join a recently-started project. While Fjermestad and his colleagues mainly focus on the children in their comprehensive research regarding various aspects of physical and emotional health for those with sexual chromosome aneuploidies, my focus was to be mainly on their parents and their mental and physiological health.

With this as set of pre-understandings, I consider my professional and personal background perspectives to be adequately covered.

Methodological discussion

The choice between qualitative or quantitative research strategies can be a practical choice, but more typically a choice of scientific standpoint beliefs, that is: how one positions oneself in understanding how and where knowledge develops and what is ‘real’. Today, the two research approaches are considered more like complementary methods than contradictions (Holbæk, 2001).

Quantitative research strategies are based on gathering information across the breath of a population or a sample selection of the population, where closeness to responders is not possible. The main goal is to collect structured information that is possible to convert into numbers and often for comparative purposes. The collected enumerative data is analyzed with using various statistical techniques (Ringdal, 2013).

A survey- questionnaire is one systematic method to collect data from a selection of persons to obtain a statistic description of a population. Such questionnaires are standardized, so that all responders are asked the same questions in the same way. Surveys are the most commonly used data-collecting method in documenting official statistics (Ringdal, 2013).

Regarding content concerning coping and life-quality which are theoretically-based social constructions, they are so abstract that they are extremely difficult impossible to objectify and measure. However, we can observe conditions or expressions that we may interpret as evidence of these terms. These conditions or signs and expressions can be used as indicators of something that is not observable, combined with the uncertainty this entails.

Research sample

The SCA sample is comprised of 25 boys, recruited from one of two settings. Seven participants were recruited from the annual meeting of the Norwegian Klinefelter Syndrome (KS) Association in 2012, where information about the study was provided and families were invited to participate. It is not known how many families were present at the meeting, which leaves the exact response rate is unknown. The remaining sample, 18 participants, was recruited through the database of Frambu, a national resource center for rare disorders. It is possible for families to self-refer, and registration in the user database is voluntary. At the time of recruitment, 44 individuals with SCA under age 18 years were registered in the database.

Six families from the database were among the participants recruited at the KS meeting. Thus, the response rate from the database was 47% (18 of 38 cases new to the study). In both settings, parents were given an envelope with the questionnaires with a stamped return envelope. The analyses are based on responses from one of the parent (20 mothers and 5 fathers). The final SCA sample comprised 25 boys. In terms of karyotype, the sample comprised 13 boys with KS, six boys with 47,XYY, three boys with 48,XXYY, and three boys with 48,XXXY. Karyotype details were parent-reported, and double-checked in medical records for participants recruited from the resource center (72% of the sample). There were no discrepancies between parent reports and medical records (Frambu v/Krister Fjermestad).

Questionnaires:

The parents were handed out the following six questionnaires. (See attachment for more details).

- Background Sheet – parents, This questionnaire was developed for this study, and consist of questions about education and employment, marital status, drug and medication use, number of children, age of children etc.
- Strength and Difficulties Questionnaire (SDQ, Goodman et al., 2000). SDQ is a 25 -unit questionnaire to be answered on a 3- point scale (0 = not at all, 1 = somewhat, 2 = a lot). The answers provide partly scorer on five factors: Depression, friend problems, behavioral problems, hyperactivity, and personal strengths.
- Personal Wellbeing Index - Parents (PWI, International Wellbeing Group, 2005). Questionnaire related to a quality of life domain, among them: standard of living, health, safety, future security. A rate from 0-10 describes the level of satisfaction.
- Pittsburgh Sleep Quality Index - Parents (PSQI; Buvsse et al., 1989). PSQI is a questionnaire with ten questions on self-perceived sleep quality. The questions relate to usual sleep habits during the past month.
- Subjective health complaints - parents (SCH, Eriksen et al., 1999). Questionnaire which 29 somatic symptoms, rated on a four - point scale according to how much you have experienced them last 30 days. SHC in a standardized instrument which measures of health issues.
- Behavioral Inhibition Questionnaire. BIQ; (Bishop, Spence, & McDonald, 2003)
  A parent-reported measure for the assessment of frequency of child’s behavior. Parents reported the frequent occurrence a certain behavior or action, and range it on a scale from 1-7, where “7” indicates “almost always” in frequency.

Validity and reliability

Validity and reliability are essential ingredients of research quality. Reliability indicates how stable and consistent observations and resultant findings are, such that measuring something repeatedly with the same method and with the same or nearly similar results, the method can be considered to have a high reliability. Reliability in social science research is equivalent to precision. Validity is a measure of whether the data collected corresponds to what ones want to measure. A method has high validity if it measures what one actually wants to investigate and measure. Validity in social science research corresponds to accuracy in scientific research (Helbæk, 2011).

The surveyed group of participants consisted of almost all parents of boys with KS nationwide, collected in a common place when the forms were dealt out. Parents may have filled out the forms together, and it can be difficult to find differences between the sexes in a set of parents. This can cause a sort of couple-conformity, and less variety in the answers. Conformity can be understood as an action to search for similarities or affiliation. One can therefore adapt attitudes, beliefs and behaviors regarding what individuals perceive as ‘normal’ within a group. This does not necessarily happen deliberately, but can have a social impact on attitudes, thoughts or behavior (Myers & Lamm, 1975)

References:

Askeland, G. (2006) Kritisk reflekterende- mer enn å reflektere og kritisere. http://www.idunn.no/ts/nsa/2006/02/kritisk_reflekterende_mer_enn_a_reflektere_og_kritisere?mode=print&skipDecorating=true&textSize=

Fook, Jan (ed) (1996) The Reflective Researcher: Social Workers' Theories of Practice Research, Sydney, Allen & Unwin.

Garsjø, O. (2001) Sosiologisk tenkemåte: En introduksjon for helse- og sosialarbeidere. Oslo: Gyldendal akademisk.

Grue, L. & Rua, M. (2013) To skritt foran : om funksjonshemning, oppvekst og mestring / Lars Grue og Marte Rua. Oslo : Gyldendal akademisk.

Klefbeck, J. & Ogden, T. (c1995) Nettverk og økologi : problemløsende arbeid med barn og unge / Johan Klefbeck og Terje Ogden. (Oslo) : TANO.

Makowski, G. (2005) Advances in Clinical Chemistry, vol. 40 Gulf Professional Publishing

Morley, Christin (2004) Critical Reflection as a Response to Globalization, International Journal of Social Welfare, Vol 13, 297–303.

Myers, D. G., & Lamm, H. (1975). The polarizing effect of group discussion. Am Sci, 63(3), 297-303.

National Association of Down Syndrome; http://www.nads.org/pages_new/facts.html, 04.05.2014

Ringdal, K. (2013) Enhet og mangfold : samfunnsvitenskapelig forskning og kvantitativ metode / Kristen Ringdal. Bergen : Fagbokforl..7

Sokol, R. Z. (2012). It's not all about the testes: medical issues in Klinefelter patients. Fertil Steril, 98(2), 261-265. doi: 10.1016/j.fertnstert.2012.05.026

Tamm, M. (cop. 2012) Psykosociala teorier vid hälsa och sjukdom / Maare Tamm. Lund : Studentlitteratur.

Tøssebro, J..(. (c2009) Funksjonshemming : politikk, hverdagsliv og arbeidsliv / Jan Tøssebro (red.). Oslo : Universitetsforlaget.

Visootsak, J., & Tartaglia, N. (2013). Autism and other neurodevelopmental disorders R. L. Hanson & S. J. Rogers (Eds.), (pp. 306).

WHO, 2011 http://whqlibdoc.who.int/hq/2011/WHO_NMH_VIP_11.01_eng.pdf?ua=1

Paper I:

Understanding disability and coping strategies of parents with disabled in a socio- ecological perspective

Words: 6514

Author: Jannike Sunde

Paper I

The purpose of this paper is to discuss disability as a constructed phenomenon, and yet still a very real one, and how society and attitudes together with physical barriers affect the meaning and content of the word “disability”. Although some fundamental arguments rightly claim that disability is constructed, impairments are also very real and can place tough strains on parents. This paper discusses some challenges parents meet when having a child with impairments, and some strategies to manage these challenges.

Studies and surveys report or imply great challenges for families with disabled children. They also reveal great determination, much love and concern, and great will and ability to adapt. It may seem that if the child’s impairment is accepted and if the family receives adequate and continuous help, that the greatest concerns are already eliminated. Statistically, family patterns in families with disabled children are not that different from other families. When it comes to number of children, parents working outside the home, divorces and break-ups, the numbers are not remarkable compared to other families. In some families, parents have also reported that having a disabled family member has brought them closer together (Tøssebro & Lundeby 2002).

Today the research trend is more directed to issues of resilience rather than the ‘problem’ itself. This explains how most families manage relatively well psychologically despite the stress and strains they experience. The concept resilience has its origin in the fields of developmental psychology, and refers to an inner resistance to external influences. This way of thinking is related to research on families with disabled children, when discovering that many families do quite well despite many challenges (Lundeby, 2008).

This paper has a social ecological perspective. This means that one’s choice of actions or feelings is never caused by a single reason, but by the complexity of the structural opportunities and constrains in the context of the time one lives in. When we approach the notion of disability with including the importance of environmental conditions, we are better able to see the constructed contextual dimensions and the social understanding of disability, including how definitions and classifications are socially constructed.

Ecological theory

Ecological theory is based on general system theory. This theory is developed with dynamic and humanistic dimensions making it applicable in disciplines such as pedagogics and physiology (Klefbeck & Ogden, 1995). How one perceives and interprets people depends on their surroundings. People are both created by their society and are creators of their society at the same time, because of a complex mutual influence, or duality (Wendelborg, 2010). Because the structural opportunities and constraints are anthropogenic, one has an option to choose differently, and contribute to changing some social structures. This theoretical approach understands people as competent and also with having awareness of their own actions.

Humans develop and are socialized in interaction with others. The American psychologist Urie Bronfenbrenner is considered the creator of the ecological model. This model consists of circles that rank the importance of the environments that surround an individual (Klefbeck & Ogden, 1995). Bronfenbrenner was mainly concerned with child psychology and primarily constructed this theory to explain children's identity. Originally, Bronfenbrenner placed a child in the center of the circles, to understand which systems effect children's development in growing up. Bronfenbrenner describes the environment as concentric circles where one environment is a circle that is incorporated into an even larger environment. Russian dolls are often used as a metaphor-image for how Bronfenbrenner envisions the different environments the individual is interacting with, where a small doll fits inside larger ones, and where the individual is at the center. This image shows that the distance between the environments and the individual is varying. Bronfenbrenner separates four levels of environmental systems: micro-, meso-, exo- and macro systems (Wendelborg, 2010). These systems are bounded in size and importance of interactions. Families are an example of a part of the microsystem.

Parents interact with several systems at these different levels. In this case, I will focus on their health and life-quality. Parents are thusly considered to be in the middle of the ecology model, especially when looking at factors that affect parental identity and coping. With an ecological perspective about families with disabled children, it is assumed that the family plays an active and important role in children's developmental conditions including during adolescence, despite that family's attendance is sometimes limited by access to different arenas in life because of a child's impairment.

A change in disability history

A new direction of thinking in the social- politics in the 1960s changed how we understand, define and value impairments and disability. Disability had earlier been considered an individual problem, and a medical understanding ruled the descriptions and responses. Because of sickness or disease, disabled people were deemed incapable to enter and participate in the society like others (Grue & Rua, 2010).

Basic welfare state ideology has been a contributing factor to disabled people being acknowledged as a natural part of society, and underpins the objectives of social integration. In the Norwegian government White paper nr. 88 (1966-67) (St.meld. nr. 88, 1966-67) regarding development of care for disabled people, the right to normal living conditions for disabled people was highlighted for the first time. Debates about normalization, integration and socialization started, including fundamental arguments about extending human rights for disabled people. These debates have made significant changes in how we regard people with various ‘mental disorders’, including recent consensus that disabled people should be entitled to the same standards of living and freedom to plan their lives as others (Grue & Rua, 2010).

The understanding and politics in the disability field in Norway was to some extent influenced by Sweden where the trend began 20 years earlier, following political efforts to evaluate and provide recommendations for guidelines for “living a good life”. One subsequent result in Norway was the establishment of two main principles for children and families, as formulated in White paper 88 (1966-67). The first was that the disabled children should grow up at home, and the other was that public services in general should be organized and provided in regular kindergartens and neighborhood schools. The reason for the first step was located in the belief that mothers and family-life were considered to provide better care than institutional settings, and thus better predictors of well-being for child development. The second principle can primarily be attributed to the ideology of inclusion and normalization, and the downsizing of the large institutions (for ex, Tøssebro & Lundeby 2002).

Normalization

The debate about the concept of normalization has led to fundamental changes in social politics in many countries, and positive changes in the lives of marginalized groups in society. The result of the debate is a new understanding concerning service, equality, needs, rights and quality in social work. Normalization was first mentioned in a public document in White Paper No. 88 (Kristiansen, 1993).

Normalization was fundamental for achieving an inclusive life for disabled people by facilitating better environmental conditions (Grue & Rua, 2013). Introducing the term "normalization" was a sign of a change from the segregative ways of thinking which had long characterized politics disability politics (Grue & Rua, 2010).

Kristiansen cites Wolfensberger who did the first attempt to make "normalization" a universal term and a theory. Normalization in social politics meant that people with a disability would have supported opportunities to follow normal daily routines, including such as normal yearly happenings or religious events. Normalization also included the idea that disabled people should have the possibility for relationships, independent economy and culturally-typical living conditions. Respect for individual choices and needs were also compatible with principles of normalization (Kristiansen, 1993). Disabled people would have an improved life-quality, and greater autonomy in designing their own lives.

In Norway today it is assumed that most disabled children grow up at home together with their siblings and parents. At the same time, there is an expectation that social services and support agencies will provide assistance and help. The intended objective is that the overall situation of the disabled child should not be fundamentally different from other children (Tøssebro & Lundeby, 2002), and that this is related to public welfare policy responsibilities. The historical changes in understanding disability are enormous and complicated, but have clearly had an important influence on the rights of disabled people and their social status.

With the two ground pillars from St.meld.88, the principles for a more equitable adolescence between disabled and 'normal' children were established. However, disabled children also usually have other needs for special attention and care than most children. The last 40 years has been characterized by an increased responsibility for the welfare state, both economically and in terms of practical help and support (Söderström & Tøssebro, 2010).

What is a 'disability'?

Having a congenital or acquired impairment often leads to a person becoming a client of social welfare services, or receiving certain special attention in daily life by using medication or aids, practical or personal assistance for participating in daily life. By using this description we see that the term 'disabled' affects a large number of people.

I will now present three different approaches of understanding the term "disability". The major part of definitions and classifications are well-established today, arising from an understanding characterized by the medicalization of disability which has prevailed the last two hundred years (for ex, Grue, 2004). Classifications with diagnoses and resultant responses represent an essential or individual understanding of what a disability *is*. This means that the understanding of a disability is reduced to a description and classification such as disease or malfunctioning body-part. Thus, disability is connected to an objective and identifiable characteristic located in the person (the actual impairment). This approach leads to understandings that medical and curative treatments are needed to improve or change the person's life situation. This way of defining a disability without any social or contextual conditions represents an understanding where disability is reduced to a personal tragedy, and those who are affected are innocent victims of a cruel destiny. The understanding disregards the importance of environmental conditions. Today, we call this understanding the "medical model" (for ex. Grue, 2004).

From the 1970s, the understanding of disability was extended and supplied by another view, called the "social model". The social model emerged from the political-activist arguments of the "Union of Physically Impaired Against Segregation" (UPIAS) in England. The social model views the problems faced by disabled people as the result of societal oppression and exclusion, rather than being located in their individual deficits. This approach emphasizes that the physical and social barriers a disabled person meets contribute to or determine the extent of how impaired and disabled a person is in a situation, and not a result of the actual impairment itself (Shakespeare, 2013). The social model defines disability as oppression, and a major consequence has been that measures to struggle for society to remove or reduce the barriers that hinder disabled people to participate in society (see for ex. Grue & Rua, 2010).

The third model is known as the "relational model". This is the dominant perception in the Nordic countries, and the one used in this thesis. In the relational model, one studies the interaction between the impaired individual and the societal/environmental contexts to assist one's understanding of the processes that create disability. It is neither *only* the individual nor *only* the context which is essential. Disability is not a result of individual characteristics or of social barriers, but is the result of anthropogenic construction in a complex ongoing interaction between the environment and the individual (Wendelborg, 2010). Time, place and context determine if one is disabled in a current situation (Grue, 2004). This way of understanding disability is consistent with the social ecological perspective on families used in this paper.

Quality of life

The term “life-quality” is deficient and debated, and continues to lack definition-consensus. It may be however safe to say that quality of life is primarily subjective, and that it relates to factors such as close relationships, sense of belonging, security, and having faith in future prospects. Most would contend that it is not possible to observe or measure degrees of life quality. Surveys regarding “life-quality” have had a growing interest during the past few years. Life-quality is an individual understanding of the sum of negative and positive feelings and experiences that you are aware off (NOU:1999). World Health Organization defines life-quality as “Individuals’ perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns.” (WHO, 1997). In order to perceive an image of life-quality, persons are often asked several different questions in matters that are considered important when it comes to daily life, such as sleep, health, future prospects and so on.

People with disabilities and their families face many barriers when they attempt to determine and increase their life-quality, acquire close friends and attempts to be self-sufficient. To have a safe and more or less predictable outlook that many people take for granted can be challenging. For many people, quality of life relates to identity and the fulfillment of various roles in society. Having the opportunity for self-actualization is the uppermost level in Maslow’s hierarchy of needs (Brümmer & Sarot, 1996). This perspective claims that one cannot start developing ‘higher-order’ needs such as esteem or aesthetic necessary to attain the highest level of self-actualization, until one’s basic needs are fulfilled. Physical needs, safety and social needs are examples of basic needs (Brümmer & Sarot, 1996). According to Maslow’s theory, self- realization or actualization is one’s ability to become the best you can be, based on your own assumptions. If the environment is appropriate, people will grow up straight, beautiful and societally successful, actualizing the potentials they have inherited (Simons et. al., 1987). It is about maximally using one’s talents to the outermost, and become all that one can and wants to be.

Self- realization typically costs time, energy and resources. For many people with disabilities, the time dimension is a major hindrance. Activities often take more time and energy including both planning and eventual performance. Additionally, often the need of assistance to carry out tasks becomes a question of resource-use. Many activities modified for disabled people are associated with special supports for individual needs. Resources such as technical aids, personal assistance and adapted settings are efforts that can improve opportunities for active participation and self-fulfillment. Here, I am referring to activities that one does based on own personal interests, rather than basic human needs. One's personal interests divide one from others, and separate you as an individual person.

A vulnerable adolescence

Humans exist in tension between being compliant and being unique. Our individuality develops in interaction with other people and our larger societies. We learn to distinguish our personal feelings, experiences and needs from other people. How we see ourselves as a person in relation to others has been discussed by many social science researchers, perhaps especially George Mead with his social mirroring theory and Erving Goffman's theory of stigma (Garsjø, 2001). Within a sociological perspective, stigma or stigmatization is seen as a process where some individuals place other individuals in a particular category by perceiving or attributing groups of people societally-devalued and negative characteristics because of appearances, behaviors, backgrounds, and so on. A stigma is a generalization where a person's specific pattern of behavior or characteristics defines a person's general character. The negative social status caused by the stigmatization takes precedence over their other features, and typically results in marginalization for the impaired person (Garsjø, 2001). Stigmatization is thus part of the disablement process.

Puberty is an extremely vulnerable time. It is a time of physiological changes, with major resultant impacts on psychological, emotional and social functioning. Lars Grue offers two possible ways for puberty to affect people psychologically. The first is how the biological changes in puberty affect a person's behavior, and second, regarding psychological and social adaption to one's surroundings. He further proposes two models to explain these changes (Grue, 1999).

The first one is the "direct-affect model". This model disregards all explanation of cultural, historical and environmental factors and only focuses on biological determination. The psychological changes in human growth are singularly explained by increased levels of hormones, and consequently how hormones affect emotions and behavioral functioning. Raised hormone-levels are seen to be the cause of sexual and aggressive impulses during the teen-years (Grue, 1999). Richards et al (1993:29, cited in Grue, 1999) refer to comprehensive research with roots in the direct-affect model, and found the model incomplete. Though most persons experience increased hormone levels due to puberty, people experience and deal with this phase in different ways.

Searching for more extended understandings and explanations of the changes during puberty, the “indirect effect-model” was developed, where both individual and contextual factors are considered as playing central roles in how physiological changes are experienced, interpreted and managed by youth and others. The school one attends, friends, activities, interests, family’s support and attitudes, and other factors impact how a person experiences changes during puberty and what consequences the physiological transformation may have in given cultural contexts (Grue, 1999).

Grue refers to Neugarten’s (1979) hypothesis about the social clock, or the “deviation hypothesis”. This line of thinking implies that humans have a biological clock determined for when certain happenings occur. If these “events” occur earlier or later than expected during the life-span, one can experience being different compared to those who develop according to what is considered age-appropriate (Grue, 1999). Teenagers who enter puberty early or late (or not at all) can be considered physiologically divergent from others and are likely to experience negative consequences from this divergence from what is expected. Adolescence is a critical time in developing one’s own identity, and appearance and behavior represent an image and invitation to other people, perhaps especially peers. Stereotypes about what constitutes ‘physically attractive’ girls and boys set high societal standards. These social signals also affect psycho-social changes that happen at this vulnerable age (Grue, 1999).

Puberty presents an individual with challenging new roles, views and ways of thinking. However, obtaining belonging to the group known as ‘teenagers’ requires feelings of belongingness and similarity, and at times an impairment causes a visible inequality (Grue, 2004). The body as part of an individuals’ experience of self- identity is increasingly more important for the modern human (Giddens 1997, cited in Kassah, 2009). Previously, the common idea was that one was a human *with* a body, whereas today to a greater extent we *are* our body. One clear example is in social media where one is always reminded of how dress, hairstyle, make-up and how you smell contribute to the image of self-presentation, to the outside world and also to yourself. Focus on diets, training and healthy appearances are perceived as more important than ever in efforts to live up to society’s ideas about beauty and perfection. Yet, attaining the perfect body is a difficult or impossible task for very many.

Stigma and devaluation in a vulnerable situation

An important concept is social role. It can be defined as a socially accepted pattern of behavior, responsibilities, expectations and privileges that are usually defined with intent to fulfill a specific function, which is assigned to a group or an individual and reflects the social status of this person or group. A severe devaluation happens when we collectively or individually devalues others as a group. Another word for attributing characteristics to people based on expectations or judgments of their appearance, is stigma (Kristiansen, 1993).

There are different ideals in cultures and societies that determine who gets devalued or stigmatized. By looking at the qualities that are highly valued in the society, we implicitly learn something about the qualities considered degraded. People with qualities or characteristics that are the opposite of what society considers positive, will be at greatest risk of being placed in devalued roles (Kristiansen, 1993).

A serious consequence about being devaluated is the risk of being rejected. Rejection from family, neighbors or society as a whole, causes an experience segregation. Segregation means being positioned distant from the majority of society. Segregation is not voluntary, and happens at a social level as well as a physical level. Using words as "us" and "them" about groups of people are examples of subtle ways to segregate. Another result of being devaluated is an experience of loss of autonomy, meaning that one may feel less control over own identity and situation and less freedom of choice. A loss of autonomy may occur in both large and small scale. Restricting laws and regulations are one example, someone making personal decisions for you is another. A person who is experiencing devaluation often develop a devaluated self-image causing a feeling of being inferior. (Kristiansen, 1993).

The devaluing labeling and imagery of persons who are different or diverging from "normality" often spreads to the family as a whole. Parents' reactions and social limitations of having a disabled child are often affected by negative societal reactions, such as fear of negative feedback limiting families from attending activities and events (Lundeby, 2008).

Stigma is a relational concept, a perceptual interaction one might say. People feel stigmatized in interaction with other people's actions, attitudes or statements. For many people, 'accepting' the stigma is the easiest way out.

Parent's coping

The introduction to the challenges regarding normalization, stigma, and a disabled body address the problems disabled people experience as they grow up, and socialize outside the childhood home. These struggles also affect parents. Seeing and experiencing their child struggle and trying to adapt into an increasingly marginalizing society is often a major stress and concern for many parents. Many parents can experience similar feelings to grief when they are told that their child is unhealthy or “different”. They feel that they have lost the child of their dreams, and need to re-orient themselves about their new family situation. Some even describes this as a crisis, and total chaos rules. When the shock and grief phase is over, the processing and acceptance phase begins. The crisis will never truly be over, and usually follows the family throughout life (Tøssebro & Lundeby, 2002).

A diagnosis can be a door-opener to practical help and benefits, but is also stigmatizing and straining. Tøssebro and Lundeby cite Cullberg who describes the same processes or phases when becoming aware or being informed that their child has an impairment. The phases consist of feelings such as shock, denial, anger, grief, and acceptance. Not everyone goes through all the phases, and the sequence of these feelings varies from individual to individual (Tøssebro & Lundeby, 2002). Parents often worry about how friends and the extended family will handle news about their impaired child to a greater extent than how they will handle the challenges themselves.

Social services and support

A diagnosis works like a tool in dealing with the news of an impairment. It is an answer to what is “wrong” with the child, and an explanation for certain behaviors or appearance. A diagnosis, or the lack of it, is often the beginning of a long working partnership between parents and public services. Parents are seeking information, help, economic compensation and support. Often this means writing applications and for some, getting these rejected. Services should be coordinated and responsibility allocated, yet many parents describe obstacles for orientation through the public systems and establishing help. Often the process of getting adequate help is criticized. Some parents find it offensive to ask for help because they feel they need to underestimate themselves and exaggerate their child’s needs in order to acquire needed supports. It can be a large burden to focus on all the negative aspects about their child, and their daily-life family struggles to obtain assistance. From Lundeby’s (2008) research, parents explain the difficulties in applying for services. They often experienced rejections, and had to cross some personal lines to access and arrange needed help. One repeated clear example that parents find especially difficult is describing in detail how much “extra-work” having a disabled child is, such as time used for dressing, eating, and transport. Many parents also find it shameful or hurtful to compare their child with “normal” children, in convincing public services that they need support. Parents find this process unfair, especially when the distribution of help and support seems somewhat randomized (Lundeby, 2008). Parents emphasize the importance of good interactions with public services. Especially the first impression with the service system is important to feel attended to, understood and supported. Trust is an important keyword. Maintaining social life, work responsibilities, and taking care of relationships are other concerns these parents are having. In order to achieve that, another important keyword is emphasized; flexibility. All families are different and expects different and customized solutions to their life situation while also depend on predictability, this requires a high degree of flexibility of social services. This is often a major challenge in the bureaucratic system of social services (Lundeby, 2008).

Common for most parents is that the way they were informed about the impairment and that communication with professionals is crucial. Choice of words is important, with a large influence on subsequent feelings about having an impaired child (Tøssebro & Lundeby 2002). Communication with specialists, economical status, educational background, and marital status are all factors that contribute to the explanation of the psycho-social health situation of these parents and how manage their overall family life situation. It is no secret that having an impaired child is a strain for most families, but the individual reactions and coping strategies are numerous and various (Lundeby, 2008).

A general challenge for these families is the opportunity to get out of the house on weekends, evenings and vacations. Many leisure-time activities are difficult because of practical challenges. In families with other children, siblings are often affected by limitations in daily life caused by the impaired child or teenager. Parents often try to compensate for this. Because of that, in addition to nurturing their own interests and needs, many parents experience pressures related to time and the extras tasks involved (Tøssebro & Lundeby, 2002). This might be meetings, respite stays, short-term institutional care, physiotherapy or other treatments, that lead to less time for other family activities. Sometimes the impairment itself leads to limitations to an active social lifestyle. Some children are less mobile and it is then more challenging to bring the child on excursions or organized activities. Parents often fear that the child may experience negative reactions. Some find it more difficult to avail themselves of babysitters: parents of children with additional needs face a greater problem than others when it comes to entrusting their child to others (Tøssebro & Lundeby 2002). Overall, parents often feel they lack the time and energy to do regular activities which other families often take for granted (Tøssebro & Lundeby 2002).

A challenge that applies to both siblings and parents seems to be having concern for the impaired child. Siblings could have a high level of worry and concern regarding the disabled child's well-being, such as poor health or being bullied. Some also report being afraid that their sibling would feel lonely, or not be taken good enough care of (Connor & Stalker, 2003).

As the child grows up and the challenges change, the worries of parents change as well. Realizing that one's child is going to grow old with impairments and limitations, and even maybe actually outlive them is a concern for many parents. Questions as "who will take care of him/her when I'm gone?", or "How will his/her life be without me?" are typical worries for many. Parents worry about getting older themselves and becoming too weak to help or supervise their child's care and safety. Worries about their child's future living conditions, educational challenges, going through puberty, and boyfriend/girlfriend questions are also difficult for many parents (Zahl, 2011).

Gallagher's study (op.cit.) confirmed that parents caring for a child with developmental disabilities report poorer sleep quality, more stress and less social support compared to other parents. These are all factors that may cause depression symptoms and lower a person's sense of coherence. Sleep quality in an important aspect of well-being and is strongly related to overall quality of life (Gallagher, Phillips, & Carroll, 2010)

Beresford cites Schilling, Gilchrist & Schinke (1984) who define three levels of social support, all of importance at different levels. The first level of support derives from close family members and friends, the second level extends to include neighbors and friends that are more distant, and the third level of support is the least intimate one, consisting of infrequent support such as formal and institutional supports. These three levels combined provide support with and for various functions (Beresford, 1994). Some need practical advice from professional or personal support groups. For some, help from a neighbor is necessary for daily chores, or perhaps emotional support from a partner or close friend is most helpful. To have outlets for personal creativity, social activities and generally experiencing feelings of 'normality', help in reducing the stress that caring for a impaired child involves (Grue, 2004).

However, sometimes, support systems have the opposite effect than their intentions and become a stressor instead of a coping resource (Beresford, 1994). Surveys show that many find interactions with professional and their service systems stressful. Many parents are also unsatisfied with the follow-up from their support services. This is also consistent with my findings concerning parents with SCA-disorders (see Paper II), where there is a correlation of negative life satisfaction, subjective health reporting, and negative satisfaction with support systems.

Finding a sense of coherence

In recent years, there has been an enormous amount of research devoted to explore the stressors associated with caring for a child with an impairment, and how these stressors may affect parents' well-being. Researchers have lately changed the focus away from describing the stressors experienced to instead investigate resilience and coping strategies of these parents (Beresford, 1994).

The modern theorist Aaron Antonovsky focuses on good health and well-being rather than illness and morbidity. He asks why some people, despite all the suffering they experience in life, still are in good health? His approach is known as a salutogenic model. Antonovsky contends that people should be seen in their total life- context. In Antonovsky's view, people are active and self-determinate and able to decide what is important and valuable in his or her life. The feeling of being empowered is considered important for a meaningful life. The model Antonovsky created is named "Sense Of Coherence" or SOC. It consists of a person's ability to understand various situations in life, to feel like we have the power to manage them, and find it meaningful to handle the challenges one meets (Tamm, 2012).

According to Antonovsky, a person is neither "sick" nor "healthy", but in a constant tug-of-war between these two poles. Our resources and use of coping strategies (SOC) define where we are in this struggle at any time. Instead of asking "what makes us sick?" Antonovsky asks: "what keeps us healthy"? (see for ex. Tamm, 2012). Beresford (1994) has analyzed how parents with impaired children manage the challenges they are given in life. The child may seem like a tragedy, but one way to manage the subsequent daily-life struggles, is to employ a salutogenic perspective. To find salutogenic strategies, one seeks for strategies that effectively minimize problems, reduce emotional stress, and make greater sense of meaning in one's existence (Beresford, 1994).

The term "coping" presupposes that a situation causing stress or strain exists. Personal coping resources are both physical and psychological ones, including physical health, moral and ideological beliefs, and previous coping experiences. These resources exhibit themselves in for example general parenting and home-making skills, cognitive abilities, and other personality characteristics. Research has proven that personality variables and personal qualities are an important coping resource in themselves, and that they potentially affect the availability of other personal and social-ecological coping resources (Beresford, 1994). A relational understanding to disability, may have led to an increased possibility to cope and manage stressful conditions (Grue, 2004).

Stressors and coping

Stress is typically combined with feelings of short-coming in interactions with others: feelings of not fulfilling the expectations of others, or from the environment. Everyone can at any time experience stressful periods, when there is a mismatch between someone's resources and expectations and demands. According to Antonovsky, stressors can be chronic or acute. The acute stressors are discrete and time limited, such as a divorce or a sudden death of a close friend or relative. In some cases, an acute stressor can alter a person's life so that it becomes a chronic stressor. A chronic stressor is a generalized and long-lasting life situation, a condition or characteristics that influence many parts of a person's life situation. Long-lasting feelings of loss or deprivation, inadequate resources, or deprivation of opportunities of a role over time are examples of chronic stressors (Olsson & Hwang, 2002).

Coping is a management strategy for handling stress. Coping means mobilization of efforts and energy in several ways, both practically but also in meeting behavioral and cognitive challenges. Coping helps one handle inner and outer troubles and burdens that one encounters in interactions with the larger environments (Lazarus & Folkman, 1984). Coping serves two main functions: to overcome or change the problem one faces and to regulate emotional responses to the experienced difficulties (Grue, 2004).

There are two ways to cope with a stressful event. One is to change the factors that cause the stress or the discomfort regarding the stress, or alternatively one can change and adapt to live with the strains and stress. These coping strategies vary from family to family. Some find relief and strength in their religion, some find emotional support from friends and close family important, while others prefer practical help (Grue, 2004).

The solution to any stressor or vulnerable situation is resources and coping strategies, but stress and vulnerability can also become a vicious circle. Stress may deplete coping resources and increase vulnerability. For example, an important coping strategy is physical energy. Caring for a child with impairments may lead to sleep deprivation and resultant lower energy, increasing parental vulnerability and general abilities to handle with the stresses of caring (Beresford, 1994). Sleep quality is an important aspect of well-being and is strongly related to overall quality-of-life and life-satisfaction both subjectively and according to more objective surveys.

Studies show that parents of children with autism, have been reported to have poorer sleep quality and more daytime dysfunction compared to control groups (Meltzer & Mindell, 2006). Parenting a child with impairments increases the risk for poorer sleep quality, and higher stress symptoms. Meltzer and Mindell (2006) found that parents caring for children with cystic fibrosis and ventilator-assisted children were characterized by poor sleep quality and associated depression. Stress among parents caring for children with development disabilities has also been shown to vary with social support, and social support has long been regarded to mitigate distress (Meltzer & Mindell, 2006).

Physical health is an important coping resource for parents of disabled children, as caring can be physically demanding. For example, lifting a heavy or resistant, uncomfortable child may be daily routines. Repeatedly awakenings during the night by the child, and can be mentally challenging as lack of sleep reduces parents' endurance and energy (Beresford, 1994).

Beresford (1994) refers to Bregman, which in 1980 conducted a unique study regarding which coping strategies parents found most helpful. Three strategies to manage everyday life were identified. The first one was to "take each day as it comes". By focusing on the present, parents planned and completed interesting and exciting activities for the children while they still were able to enjoy them (Beresford, 1994). The second most important strategy is to "maintain as normally lifestyle as possible". By doing that, children would be able to maintain social contact with other (healthy) children as long as possible. They secured that their child would be treated normally by people in the neighborhood, and by modifying the house and children's clothing they would be able to help themselves as long as possible (Beresford, 1994). Normalization is an important strategy for parents' coping, as well as a central key for the political approach to include disabled into the society. To live a life as normal as possible includes that the child participates in normal activities, but it also means that the parents have a normal life situation with space for socialization and activities.

The last strategy for coping mechanism, is "reducing the risk of crisis". This was done by keeping themselves well-informed about the child's situation and seeking out for the best options for the child's health care, education and aids and monitoring the standard of services (Beresford, 1994). As for the mental state, a common cognitive coping strategy for parents of disabled children are " reminding self about how much worse it could be", according to Brown and Hepple's (1989) (in Beresford, 1994).

Final comments

The term “disability” involves an extended and somewhat diffuse debate, including a wide range of what is considered an impairment, how we understand interaction with social and other environmental conditions, and how society, families and individuals react and respond. Explanation of concepts and different phenomena in social work can help us to identify and understand notions of reality and social constructions in a different perspective than our own. An understanding of disability as a social and cultural phenomenon. A “disability” appears (or not) in certain situations dependent on time, place and culture, and is thus relatively situation-dependent and socially-constructed. Impaired and then disabled people and their families face many daily challenges: physical hindrances, systemic structural obstacles, and more personal psychological, physiological or socially strains.

Despite many decades of radical changes in social work, influenced by the debate about normalization and human rights, discrimination and stigmatization are still topical issues. Research has documented how these challenges affect the children and their path to adolescence and adulthood, and also at times very challenging and difficult for parents and siblings. Despite various forms of public assistance, respite alternatives, economic supports and other help from formal and informal services, many parents still develop symptoms of stress and depression.

Stress is an individual feeling, and depends on individual characteristics. To predict how someone might cope or adjust to stressors depends somewhat on how and why an individual views his/herself and their surrounding society and world. Antonovsky’s theory emphasizes the importance of personality factors which contribute to good health and well-being as opposed to stress factors. Personal resistant resources contribute to making stressors understandable and more manageable for the individual encountering stressful situations.

A social- ecological understanding implies that the disability does not only affect the individual but the entire family. Social services should reflect that, and offer help directed to the family has a whole, not only the individual. The support should aim to mitigate parents’ symptoms of stress, as well as taking steps to contribute a normal life situation for the family as possible.

References:

Beresford, B. A. (1993) Resources and strategies: How parents cope with the care of a disabled child. J. Child Psychiat. Vol. 35, No 1, pp 171-209

Bishop, G., Spence, S. H., & McDonald, C. (2003). Can parents and teachers provide a reliable and valid report of behavioral inhibition? Child Dev, 74(6), 1899-1917.

Brümmer, V. & Sarot, M (1996) Happiness, well- being and the meaning of life. A dialogue of social science and religion.

Connors, C. & Stalker, K. (2003) The views and experiences of disabled children and their siblings : a positive outlook London : Jessica Kingsley.

Folkman, S. & Lazarus, R.S. (c1984) Stress, appraisal, and coping New York : Springer.

Gallagher, S., Phillips, A. C., & Carroll, D. (2010). Parental stress is associated with poor sleep quality in parents caring for children with developmental disabilities. J Pediatr Psychol, 35(7), 728-737. doi: 10.1093/jpepsy/jsp093

Garsjø, O. (2001) Sosiologisk tenkemåte : en introduksjon for helse- og sosialarbeidere / Olav Garsjø. Oslo : Gyldendal akademisk.

Gallagher, S., Phillips, A. C., Oliver, C., & Carroll, D. (2008). Predictors of psychological morbidity in parents of children with intellectual disabilities. J Pediatr Psychol, 33(10), 1129-1136. doi: 10.1093/jpepsy/jsn040

Grue, L. (1999) Funksjonshemmet ungdom og livskvalitet. Oslo : Ad notam Gyldendal

Grue, L. (2004) Funksjonshemmet er bare et ord. Forståelse, fremstillinger og erfaringer Oslo:Abstrakt forlag

Grue, Lars & Rua, Marte (2010), Funksjonsnedsettelse, oppvekst og habilitering, NOVA, Norsk institutt for forskning om oppvekst, velferd og aldring. Rapport nr 19/2010

Grue, L. og Rua, M. (2013) To skritt foran: om funksjonshemning, oppvekst og mestring / Lars Grue og Marte Rua. Oslo: Gyldendal akademisk.

Kassah, A.K. & Kassah, B.L. (2009) Funksjonshemning : sentrale ideer, modeller og debatter, Bergen : Fagbokforl..

Klefbeck, J. og Ogden, T. (1995) Nettverk og økologi : problemløsende arbeid med barn og unge. Oslo: TANO.

Kristiansen, K. (1993) *Normalisering og verdsetjing av sosial rolle.* Oslo : Kommuneforlaget.

Lundeby, H (2008) Foreldre med funksjonshemmete barn:En studie av familiemønster, yrkesaktivitet og møter med hjelpeapparatet. NTNU, Norges teknisk-naturvitenskapelige universitet. Fakultet for samfunnsvitenskap og teknologiledelse. Institutt for sosialt arbeid og helsevitenskap

Lundeby, H. og Tøssebro, J. (2002) Å vokse opp med funksjonshemming: de første årene Oslo: Gyldendal Akademisk.

Meltzer, J. Lisa, & Mindell, A. Jodi. (2006). Impact of a Child's Chronic Illness on Maternal Sleep and Daytime Functioning American Medical Association, 166(1), 1749-1755.

NOU 1999: 2 Livshjelp, Behandling, pleie og omsorg for uhelbredelig syke og døende (http://www.regjeringen.no/nb/dep/hod/dok/nouer/1999/nou-1999-2.html?id=141460. 03.05.14)

Olsson, M. B., & Hwang, C. P. (2002). Sense of coherence in parents of children with different developmental disabilities. J Intellect Disabil Res, 46(Pt 7), 548-559.

Shakespeare, T (2013) The social model of disability. The disability studies Reader. Davis, Lennard J., 2013. Routledge, New York.

Simons, Janet A., Irwin, Donald B. Drinnien and Beverly A. (1987) Maslow's hierarchy of needs. from Psychology - The Search for Understanding.West Publishing Company, New York

Söderström, S. & Tøssebro, J. (2010) Innfridde mål eller brute visjoner? Noen hovedlinjer I utviklingen av levekår og tjenester for utviklingshemmede. NTNU, Samfunnsforskning AS, Mangfold og inkludering.

St.meld.nr.88 (1966-67). Om utviklingen av omsorgen for funksjonshemmede. Oslo: Sosialdepartementet.

Tamm, M. (cop. 2012) Psykosociala teorier vid hälsa och sjukdom. Lund : Studentlitteratur.

Wendelborg, C, 2010. Å vokse opp med funksjonshemming i skole og blant jevnaldrene. Philosophiae doctor avh. NTNU, fakultet for samfunnsvitenskap og teknologiledelse.

WHO (1997) World Health Organization: WHOQOL, Measuring Quality of Life. (http://www.who.int/mental_health/media/68.pdf. 03.05.14)

Zahl, K (2011) Skal vi velge livet? Juritzen forlag, Oslo

Paper II:

The health and social- emotional problems of parents with children with sex chromosome aneuploidies

Words: 5041

Author: Jannike Sunde

Co-author: Krister Fjermestad

Fjermestad’s contribution will be referred to in the text.

The data for this study was collected and provided by Krister Fjermestad. Fjermestad have contributed with comments and feedback on this paper.

Introduction

The purpose of this study is to increase the knowledge of psychosocial health among parents of boys with sex chromosome disorders. The knowledge about the psychosocial health of this group is very limited. An increased awareness about these parents can recognize stressors and coping strategies for parents in a vulnerable situation. Finding in correlation between children's and parents' problems in this study, may help prevent developing symptoms of stress and depression in terms of early intervention. Using Antonovsky's theory of "sense of coherence" is valuable in understanding individual differences in psychological adaption in parents of children with a disability (Olsson & Hwang, 2002).

Antonovsky's sense of coherence

It has been over 30 years since the American- Israeli medical sociologist Aaron Antonovsky introduced his salutogenic theory "sense of coherence" as a global orientation. Antonovsky claimed that the way people view their life has an influence on their health. Sense of coherence explain why people stay well and are able to improve their health in stressful situations (Lindstrom & Eriksson, 2006). Instead of focusing on what makes us sick, Antonovsky looks at factors promoting health, despite pathogenesis. Antonovsky's health study is called salutogenesis (Malterud & Walseth, 2004).

What is experienced as stress is depended by the characteristics in the person who is exposed by it. Antonovsky suggests that it is impossible to foresee what consequences a specific stressors has on an individual's health without knowing something about that person's view of her/himself and the world (Olsson & Hwang, 2002). People have to understand their lives and be understood by others in order to perceive that they are able to manage a situation, and most importantly is that this must be perceived as meaningful enough to find motivation to continue to manage the situation (Lindstrom & Eriksson, 2006).

Antonovsky's "sense of coherence" comprises three components; "understanding", "manageability" and "meaningfulness". "Understanding" is defined as the ability to comprehend stimuli as consistent and structuralized information. "Manageability" is the amount of resources one holds to meet different demands, and the last factor, "meaningfulness", is the level of how meaningful you find it to handle or solve your stressors (Grønholt, Nordhagen, & Heiberg, 2007).

Parents of children with intellectual disability with low sense of coherence, experience higher level of stress and depression than parents of children who develop normally. The life situation of parenting a child with intellectual disability may have a negative impact on parents' sense of coherence (SOC) levels, which, in turn will make them more vulnerable to experiencing stress and depression (Olsson & Hwang, 2002). The SOC theory is a good instrument in understanding the individual differences in coping and psychological adaption for parents with children with intellectual disability.

A study by Gallagher confirmed that parents caring for children with developmental disabilities report poorer sleep quality, more stress and less social support, all recognized as factors that may cause depression symptoms and reduce a person's sense of coherence. Sleep quality is an important aspect of well- being and is strongly related to overall quality of life (Gallagher et al., 2008). The burdens of raising a child with disabilities may cause parents to be in a psychological state of stress, which can be manifested in poor psychological health (Olsson & Hwang, 2002). Meltzer and Mindell (2006) found in their research that parents caring for children with cystic fibrosis and ventilator-assisted children were characterized by poor sleep quality and depression (Meltzer & Mindell, 2006).

Parents of disabled children experience a different life situation than others. Some parents describes days as brutal. Sleepless night characterizes everyday life. Some families have a strong need for routines to make it through the day. Parents also need confirmation in their expertise in being parents to feel appreciated (Johnsen, Fegran, & Kristoffersen, 2012).

In the salutogenic model, experiencing life as meaningful and having a positive way of thinking is health promoting. Despite the challenges in care giving to a child with impairments, parents also need to be aware of the healthy and normal in their child. Studies show that parents who are able to consider their child as a resource to personal growth, joy, maternity, or realization had better outcome regarding coping management, and increased belief in managing their situation. They experienced care giving meaningful, and that life had a new dimension by realizing what they consider important in life (Johnsen et al., 2012).

Sex chromosome aneuploidy

Sex chromosome aneuploidy (SCA) is the term used to describe a group of chromosomal disorders in which individuals are born with an atypical number of sex chromosomes (Visootsak & Tartaglia, 2013). The missing or extra sex chromosome lead to variety of conditions. Having an extra or missing X or Y- chromosome(s) is associated with increased risk for both physical and socio-emotional difficulties, including psychiatric and behavioral disorders (Visootsak & Graham, 2006). As a group, SCA conditions are estimated to occur in

1 in 400 births, making them the most common chromosomal abnormalities in humans, and are more common than Fragile X- syndrome and Down's syndrome. SCA are also known as sex chromosome abnormalities, sex chromosome variants or sex chromosome anomalies (Visootsak & Tartaglia, 2013).

Considerable documentation show increased risk for physical and socio-emotional difficulties in all SCA variations, but there are some important differences between karyotypes (N. Tartaglia, Ayari, Howell, D'Epagnier, & Zeitler, 2011). Somatic and cognitive development are more likely to be affected as the number of X chromosomes increases. Each extra X chromosome is associated with an IQ decrease in approximately 15-16 points, with language most affected, particularly expressive skills (Linden, Bender, & Robinson, 1995). Medical conditions that affect the lives of boys and men with SCA, includes tumors, vascular disease, endocrine, metabolic and autoimmune diseases, and cognitive and behavioral dysfunction (Sigman, 2012). The symptoms are individual and are expressed in different ages.

This study is comprised of boys with four of the most common sex chromosome aneuploidies. Klinefelter syndrome 47, XXY (KS), is the most commonly occurring combination in SCA disorders. KS occurs in up to 1:650 males. Other combination which also is referred to in this thesis, is 47, XYY, occurring in 1:1000 males, 48, XXYY, occurring in 1:18 000 males, and 48, XXXY, occurring in 1:17 000 males (Cordeiro, Tartaglia, Roeltgen, & Ross, 2012; N. R. Tartaglia, Ayari, Hutaff-Lee, & Boada, 2012). Based on a comparison of the number of cases ascertained by clinical genetic testing to the prevalence of SCA 1 in 400 births, it is estimated that no more than 25% of individuals with SCA are diagnosed in their lifetime (Visootsak & Tartaglia, 2013). Only about 10 % are diagnosed before puberty. A British study estimates similar frequency of diagnosis. A mild phenotype and a great variation in symptoms might be the main reason why so many men goes through their lives without a diagnosis (Groth, Skakkebaek, Host, Gravholt, & Bojesen, 2013).

Klinefelter syndrome

Over 50% of the boys in our sample is diagnosed with Klinefelter syndrome. Parents may often observe abnormal development in their child, without recognizing any specific pattern to confirm a feeling of "something wrong". Language skills are often delayed, with first words spoken around 18 to 24 months, versus normally 12 months. The language development persist during childhood, and leads to challenges and frustration when difficulties in formulating sentence structure and producing coherent narrative occurs. Deficits occur in comprehension when the language is complex or abstract. Because of problems in the language and communication, parents may often experience behavioral outbursts of frustration. During the first few years of life, when the need for testosterone is low, most males with the XXY karyotype do not show any obvious differences from typical male infants and young boys. Some may have slightly weaker muscles, resulting in late ability to sitting up, crawling, and walking later than average. In example, baby boys with KS do not start walking until age 18 months on average. (Simpson et al., 2003).

Decreased production of testosterone is a common expression for diagnosis and affects various physical conditions. Small testes and hypergonadotropic hypogonadism are key findings in Klinefelter syndrome. The hypogonadism may lead to changes in body composition and a risk of developing metabolic syndrome and type 2 diabetes (Groth et al., 2013). Enlarged breast tissue (gynecomastia) is a relatively common symptom for all boys in puberty (Herlihy et al., 2011). As are slightly increased final height and slightly wider hips with narrower shoulders. The increased final height may be caused by abnormally long legs (Wattendorf & Muenke, 2005). The physical symptoms of Klinefelter syndrome depends on the testosterone levels in the body. The degree of symptoms is based on the amount of testosterone that is needed for a specific age or developmental stage, and the amount of testosterone available for the body (Simpson et al., 2003).

Klinefelter syndrome is the most common cause of primary testicular failure and infertility (Herlihy et al., 2011; Sigman, 2012). Some men are not diagnosed until grown age, when facing reproducing problems (Sigman, 2012). Poor coordination, dexterity, running ability, poor muscle tone and strength, synkinetic movements and tremor are usually identified in early childhood (Sokol, 2012).

Although intelligence in general can be within the normal range, a reduced cognitive functioning will be recognized, as boys with Klinefelter has a reduced capability with reading and writing in combination with reduced psychological impetus. Social difficulties may be experienced as a consequence of language based learning difficulties (van Rijn, Swaab, Aleman, & Kahn, 2008). Many boys may benefit of speech therapy and special education (Groth et al., 2013).

In a study by Rijn (2008) men with Klinefelter syndrome reported more distress during specific social situations and were characterized by increased levels of autistic features across all dimensions of the autism phenotype (van Rijn et al., 2008). Finding of high rates of autism traits across all dimensions of the autism phenotype may indicate vulnerability for autistic features in social behavior and communication, which may also extend to other aspects of the phenotype as well. Rijn's findings suggests that it is possible that men with Klinefelter not only have an increased vulnerability for autism, but also for other psychiatric disorders.

Testosterone treatment may contribute to secure a proper masculine development of sexual characteristics, prevent osteoporosis, sufficient increase in muscle bulk, increase energy and improve mood and concentration (Groth et al., 2013). Because many men with Klinefelter are diagnosed late in life, an effective treatment are delayed. In particular, early treatment of learning disabilities and androgen deficiency are often imperative for optimal outcome (Simpson et al., 2003). A lifelong treatment is recommended, though this practice is not evidence- based (Groth et al., 2013).

Parents' coherence

Several studies implies that parents with disabled children are physically and emotionally challenged. Parents often experience a state of psychological stress because of the demands of raising a child with disabilities. The life situation of parenting a child with intellectual disability may have a negative effect on parents' sense of coherence, and make them more vulnerable to experience stress and depression. This stress can be manifested in poor psychological health (Olsson & Hwang, 2002).

Stress among parents caring for children with developmental disabilities has been shown to vary with social support, which has long been regarded to mitigate stress (Bailey, Wolfe & Wolfe, 1994). Parents of children with developmental disabilities were found to report less social support (Gallagher et al., 2008). The study by Gallagher et.al., (2008) explored whether stress, child behavior problems and social support were associated with increased risk of poor sleep quality in parental care givers.

A study of caregivers of ventilator depended children, reported caregiver sleep quality was found to mediate the relationship between child's health and caregiver's depression and fatigue (Gallagher, Phillips, & Carroll, 2010).

Caregiving for disabled is associated with health risks. Recent studies have shown that caring for disabled and impaired children can have a significant negative impact on physical health and life expectancy. Gallagher (et. al, 2010) refers to several studies supporting this theory. Caring for a child with a chronic illness goes above and beyond the responsibilities of raising a healthy child. Studies have reported higher rates of depression and anxiety in parents of children with chronic health conditions or disabilities compared with parents of healthy children (Gallagher et al., 2010).

In a study by Breslau, Staruch and Mortimer (1982), the impact of child disability on psychological distress in mothers were examined. Scores on two indexes of psychological distress was compared on 369 mothers of children with cystic fibrosis, cerebral palsy, myelodysplasia, or multiple physical handicaps, with those of 456 mothers from a randomly selected sample of families as "control" subjects. Mothers of disabled children scored significantly higher than the control group on both indexes of psychological distress. This finding persisted when the mothers' education, family income, and racial composition were controlled for. Type of disability and the diagnostic classification of the disabled children, was unrelated to the mother's level of psychological distress. In contrast, the disabled child's dependence on others in daily activities had a significant effect on both measures of psychological distress. Mothers’ distress increased with the child’s dependency (Breslau, Staruch, & Mortimer, 1982).

Main questions

There is a considerable lack of knowledge about parents of children with SCA as a group. Considering the wide range of challenges boys with SCA may experience, both mentally, socially, cognitively and psychologically, I would like to take a look at if there is any correlation in these children’s and their parents’ health situation. I aim to examine challenges the parents’ and children are struggling with, and see if there is any correlation in their problems. Because of extensive documentation in the research literature on psychosocial health is closely related to sleep quality (Pallesen et. al., 2005; (Gallagher et al., 2008). Additionally, I aim to include parents’ sleep quality as one of the objectives of psychosocial health, as well as subjective health complaints and personal well-being index.

The main questions in this study are:

a) How does parents of boys with SCA rate their subjective health, sleep quality and quality of life?
b) Are parents’ mental and physical health related to the functioning of their child?

The importance of this study’s results will be valuable for support systems when approaching parents with children with extra needs, and offer adequate help and follow-up. The information will also be important for the parents who participated in this survey, and other families in similar situations. Finally, the information will be of importance for further research and future comparison of this group.

Methods:

This study is conducted in collaboration with Frambu, a national competence center for rare disorders in Norway. Frambu has gathered information from 25 families with boys with sex chromosome aneuploidies, using different questionnaires to perceive and identify information from both parent and children. The parents were recruited in two stages. Seven families were recruited from the annual meeting of the Norwegian Klinefelter Association in 2012, where information about the study was provided and families were invited to participate. It is not known how many families were present at the meeting leaving the exact response rate unknown. The remaining sample (18 families) was recruited through the database of Frambu. The study and information gathering were already approved by the Regional committees for Medical and Health Research Ethics (REK) when I joined the study.

The participants consist of 38 parents from 25 families. The parents were handed out the following six questionnaires, which was completed by the mother, father or by both parents. All questionnaires are parent- reported.

The Behavioral Inhibition Questionnaire (BIQ; Bishop, Spence, & McDonald, 2003). This questionnaire contains 30 questions to be answered by parents about their children on which extent, ranged from 1 to 7, different temperament traits were present during childhood. The Behavioral Inhibition Questionnaire (BIQ) is a parent-rating scale for measuring temperamental characteristics referring to shyness, fearfulness, and withdrawal in young, preschool children. In the current study, parents of children above preschool-age was asked to rate the BIQ retrospectively, describing the child's behavior in different situations in preschool (aged 3-5). For each statement, parents were asked to consider whether their child showed a behavior "almost never," "rarely," "once a month", "sometimes", "often", "very often" or "almost always". Internal consistency for the SCA sample is α= 95

The strengths and difficulties questionnaire (SDQ; Goodman, Ford, Simmons, Gatward &Meltzer, 2003). The SDQ is a 25- item measure parent- reported questionnaire, designed to assess psychological adjustment and socio-emotional functioning in children aged 3 to 16 years. The instrument produces scores on five subscales: emotional symptoms, conduct problems, hyperactivity and inattention, peer difficulties and pro- social behavior. The participants were required to indicate either 0 (not true), 1 (somewhat true) or 2 (certainly true) for each statement, with higher scores indicative of more significant problems for each subscale, except pro-social behavior where higher scores indicate positive adjustment. The current study internal consistency for the SDQ- questionnaire is α= .70.

Personal wellbeing index. (PWI, International Wellbeing Group, 2005). The PWI scale in this paper contains seven items of satisfaction, each one corresponding to a quality of life domain as: standard of living, health, achieving in life, relationships, safety, community-connectedness, and future security. These domains are theoretically embedded, as representing the first level deconstruction of the global question: "how satisfied are you with your life as a whole?". In the current study the reliability test shows chronbach alpha lies at α.85, which indicate strong consistency.

Pittsburgh Sleep Quality Index (PSQI; Buysse, et.al.,1989). The PSQI questionnaire is a self-rated questionnaire which assesses sleep quality and disturbances over a 1 month time interval. 19 individual items generate seven "component" scores: subjective sleep quality, sleep latency, sleep duration, habitual sleep efficiency, sleep disturbances, use of sleeping medication, and daytime function. In the current study internal consistency for the PSQI- questionnaire was α= .76.

Parents are asked to range the frequency of different statements regarding their sleep quality during the last month. Our sample comprises 14 of the originally 19 questions, whereas the four latter questions are used for clinical information only, and the last question in the originally PSQI- questionnaire are excluded from this sample. Scores on items range from 0 to 3, where 0 is no difficulty and 3 is severe difficulty.

By summing the components scores, a total sleep quality score is obtained that ranges from 0 to 15, 0 meaning good sleeping quality, while 15 refers to "bad sleepers".

Subjective health complaints (SCH; Eriksen et. al., 1999). The SHC- questionnaire is based on subjective statements about encountered problems and will by definition provide answers to the amount of pain experienced the last month. The questionnaire consist of 29 somatic symptoms, and parents were asked to range on a four-point scale pain symptoms in head, shoulders, migraine, allergies, dizziness and anxiety. The parents were also asked to estimate number of days they experienced the health problem the last 30 days. The internal consistency for the SHC- questionnaire in our sample is α = .84

Background questionnaire

In addition to the standard questionnaires, participants in the sample received a background questionnaire developed for this study. This covered five main areas: demographic information, education and work situation, SCA diagnostic information, physical health, sleep, and experiences with health professionals.

Sample:

The analyzes of the questionnaires about the parents, are based on responses from all 38 participants, and questionnaires regarding the children are based on responses from one parent from each of the 25 families. The questionnaires regarding the children are parent- reported.

The 25 boys were in the age from 2-18 years old, mean age 11.7 (SD= 4.5). Parents report different karyotypes for the boys. The sample is comprised of the following composition;

Headline

N	Karyotype	Mean age	SD	Age range
13	47, XXY	12.4	4.5	4 – 18
6	47, XYY	13	3.2	8 – 17
3	48, XXYY	9.3	5.5	3 – 13
3	48, XXXY	8.3	5.7	2 – 13

Table 1

2. Overview parents work and education:

		Mother	Father
Education	Secondary sch.		3
	High school	6	5
	University < 3	5	3
	University > 3	9	4
	Other	0	1
	Total	20	16
Work sit.	Working	14	14
	Disabled	2	1
	Working at home	2	
	Unemployed	1	
	Other	1	1
	Total	20	16

Table 2

The mothers in our sample have relatively high education. They also have higher education than the fathers in the sample. 45% of the mothers in this sample have completed a university degree longer than 3 years, compared to 25% of the fathers.

The majority of parents are working outside the home, 70 % of the mothers and 87% of the fathers. 3 of the parents are receiving disability benefits, 2 mothers, and one father.

3. Parent- reported problems

	Informant	N	Mean	SD	T- value	P- value
SHC total	Mother	18	15.6	8.1		
	Father	16	6.3	5.1	3.93	.000
PWI total	Mother	20	55	12.5		
	Father	16	60	8.8	-1.34	.188
PSQI total	Mother	20	9.1	2.8		
	Father	16	6.7	2.2	2.78	.009
	Both	38	7.9	2.8		

Table 3

A total score of all parents’ problem questionnaires are summed, and the parent are grouped in “mothers” and “fathers” to indicate differences. Because two parents have filled out the questionnaires together a combined score at the PSQI total form results in 38 parents.

High score on the SHC- total indicate higher level of health complaints. Mothers report a much higher total score on subjective health complaints than the fathers.

The PWI questionnaire has a positive direction, meaning that high values represent high satisfaction of life. Fathers mean score are 60, meaning that they are more satisfied with life standard and quality of life, than mothers. Even though differences are observed, an independent sample test could not identify any significant differences in the two groups.

It is calculated a mean score in question 1 in the PSQI. This score is summed with the rest of the scores in the questionnaire. The total range from 0-15 indicate the sleep quality of parents, high scores represent bad sleepers. Mean score is 7.9 for both parents.

4. Comparing parents' personal wellbeing

	Informant	N	Mean	SD	T-value	P-value
Overall life satisfaction	Mother	20	6.5	1.9		
	Father	16	7.2	1.3	-1.24	.22
Life standard satisfaction	Mother	20	7.6	1.7		
	Father	16	7.9	1.3	-.44	.67
Satisfaction health	Mother	20	5.4	2.4		
	Father	16	7.3	1.9	-.39*	.01
Satisfaction life achievm.	Mother	20	7.2	2		
	Father	16	7.4	1.5	-.40	.70
Satisfaction close relat.	Mother	20	7	2.5		
	Father	16	6.9	1.9	.08	.94
Safety	Mother	20	7.7	2		
	Father	16	7.8	1.7	-.10	.92
Close with community	Mother	20	6.3	3.1		
	Father	16	7.4	1.7	-1.38	.18
Future safety perspective	Mother	20	7.1	1.9		
	Father	16	7.9	1.3	-1.37	.18

The correlation is significant at level: *P< ,05 , **P< ,001 *

Table 4

This table shows the result of personal wellbeing for mothers and fathers. High numbers at the mean score indicate high satisfaction. Results shows that fathers report higher satisfaction in every scale of the personal wellbeing index than mothers, except "satisfaction close relations", the mothers scores slightly higher than fathers at this scale. Even though we notice fathers higher overall satisfaction, only one of the scales are found significant in the comparison of mothers and fathers. This is related to satisfaction of health situation.

Correlation in parent’s- and children’s- problem

	Age	PWI total	SHC total	SHC days	SDQ emot.	SDQ conduct	SDQ hyper	SDQ peer prob	SDQ pro	SDQ total
PWI total	-.40									
SHC total	.16	-.41								
SHC days	.04	-.36	.89**							
SDQ emotion.	.48*	-.30	.44*	.43						
SDQ conduct	.15	-.20	.57**	.73**	.26					
SDQ hyper	.25	-.18	.42*	.55	.06	.75**				
SDQ peer problems	.37	-.52**	.56**	.58*	.58**	.58**	.30			
SDQ pro	-.17	.09	-.34	-.55	-.29	-.42*	-.28	-.24		
SDQ total	.41*	-.40*	.65**	.75**	.64**	.86**	.68**	.83**	-.41*	
PSQI total	.29	-.43*	.87**	.85**	.56**	.60**	.38	.50*	-.50	.67**

The correlation is significant at level: *P< ,05 , **P< ,001 *

Tabell 5

Correlation in parent-reported problems and children- reported problem and age, are shown in table 5. The boys’ age correlated significantly with both total SDQ total problems (r = .41, p < .05) and the emotional problems subscale (r = .48, p < .05).

Personal wellbeing in parents correlates negatively with the SDQ total range, indicating a correlation between negative behaviors in children, and low satisfaction in wellbeing for parents. (r= -,40, p < .05)

Parents sleep quality is strongly connected with several other factors. A poor sleep quality correlates with low personal well-being scores (r= -.43, p <.05), high scores in health complaints (r=.87, p< 0.01), high number of days with health complaints (r=.85, p< 0.01). The parent-reported sleep quality correlates with the strength and difficulties questionnaire on almost every subscale.

Discussion

Previous studies show that parents of children with psychological impairments have lower sense of coherence and poorer sleep quality than other parents. In this study, I wanted to see if the same connection applies to parents of children with SCA. Two main question were emphasized. First, I wanted to see how parents evaluate their subjective health, sleep quality and quality of life, second I wanted to see if the parents' health was connected to child's functioning.

Parents in this study reports big variety of sleep quality. Table 3 show the mean score in sleep quality which is at 6,7. This is on the upper level of our ranging, indicating poor sleep for many of our parents, especially mothers. Previous studies show that parenting a disabled child increases the risk of poorer sleep quality, and higher stress symptoms. Parents sleep quality correlates with every SDQ subscales, except hyperactivity and the pro- social scale. Poor functioning and difficulties in behavior of children indicates poor sleep quality for parents.

Sleep quality is an important aspect of well-being and is strongly related to overall quality of life and life satisfaction (Gallagher, Phillips, & Carroll, 2010). This may be interpreted to that many of the parents in this sample, mothers especially, have greater risk to experience more stress symptoms, and less satisfaction of life and low life quality. In this thesis the importance of sleep quality has been emphasized regarding symptoms of stress and depression. Parents with extra care burden are in higher risk of poor sleep quality. This risk is also present for the parents in this study, especially the mothers.

The results from the SHC- questionnaire supports this theory in our case as well. In Table 3 mothers report higher scores on subjective health complaints than fathers, and have a severe higher result when it comes to number of days they have experienced pain during the last month. Mothers also have lower scores in satisfaction with questions regarding life quality, though the difference is not big enough to be considered significant.

In Table 3, we also see that fathers are in general more satisfied with their life situation according to the Personal Wellbeing Index questionnaire (PWI). High values represent high satisfaction, and possible top score is '10'. Table 4 shows an accurate measurement of the well-being factors. Only when it comes to satisfaction in close relations, mothers are slightly more satisfied then fathers (mean mothers=7, fathers= 6.9). The lowest scores occurs in question regarding health, and life achievements. Mothers are most dissatisfied with their health, with a mean score at only 5.4. The mean score on overall satisfaction of life quality for mothers is 6.5.

Even though mothers report severe higher amount of subjective health complaints than fathers, there is not an equivalent difference in personal wellbeing in mothers and fathers. Mothers report over twice as much health complaints in comparison to the father group, and they score significantly lower on health satisfaction than fathers. Still, they score very similar to the fathers when it comes to life satisfaction, and life quality- related questions.

The challenges of children with sex chromosome aneuploidies do not decrease as they get older, but changes and often get more perplex with age. This can be challenging for parents, especially considering that the parents are getting older as well. Increased problems as children get older was noticeably, but tests in this sample did not give any significant results and the research question were removed from the paper.

Based on the parent's reports, the study confirms that there is a correlation between parents' socio- emotional problems and children's functioning. It also shows that especially the mothers reports more health complaints and lower satisfaction on most life domains. The study does not give us any clear indication of the cause of these findings, but the correlation between the parents and children points towards that giving the children the adequate help with their difficulties, may also help the parents by reducing socio- emotional problems and improve their perception of life quality.

It is important that parents are aware of the possible risks of parenting a disabled child. Knowledge about reactions and feelings that may occur when facing the challenges and stressors which is normal when you have a child with a disability, may reduce assumptions of having abnormal feelings or reactions. Psychoeducation is a good approach in making parents aware of these risks. This method is based on teaching the participants about their situation or condition, and making them empowered to take control of their problems. In that way they and take steps to prevent developing poor socio- emotional health and sleep quality. Family counseling services is a possible instance for providing information for this group of parents.

There are several weaknesses and limitations in the current study. A small sample size is one of them. The small sample has made it difficult to matching the SCA sample to clinical sample and control groups. Another issue regarding generalizability is the fact that SCAs are heavily under-diagnosed. Our sample of diagnosed boys may not be representative for the undiagnosed SCA population. Our sample is also comprised of a wide age- range in the SCA- boys, making it difficult to perform comparisons.

Another limitation to the study is the lack of Norwegian norms for all scales and questionnaires. Also, there is only implemented one measurement, and there has not been any long-lasting follow-up by the families. Finally, the results are based on parent-report only, which may also be a limitation to the validity of this study.

References:

Bailey D, Wolfe DM, Wolfe CR. With a little help from our friends: Social support as a source of well-being and of coping with stress. Journal of Sociology and Social Welfare 1994;21:127-152.

Bishop, G., Spence, S. H., & McDonald, C. (2003). Can parents and teachers provide a reliable and valid report of behavioral inhibition? Child Dev, 74(6), 1899-1917.

Breslau, N., Staruch, K. S., & Mortimer, E. A., Jr. (1982). Psychological distress in mothers of disabled children. *Am J Dis Child, 136*(8), 682-686.

Buysse, D. J., Reynolds III, C. F., Monk, T. H., Berman, S. R., & Kupfer, D. J. (1989). The Pittsburgh Sleep Quality Index: A new instrument for psychiatric practice and research. Psychiatry Research, 28, 193- 213.

Cordeiro, L., Tartaglia, N., Roeltgen, D., & Ross, J. (2012). Social deficits in male children and adolescents with sex chromosome aneuploidy: a comparison of XXY, XYY, and XXYY syndromes. *Res Dev Disabil, 33*(4), 1254-1263. doi: 10.1016/j.ridd.2012.02.013

Goodman, R., Ford, T., Simmons, H., Gatward, R., Meltzer, H. (2003) Using the Strenght and Difficulties Questionnaire (SDQ) to screen for child psychiatric disorders in a community sample. *British Journal of Psychiatry*, **177**, 534-539

Eriksen, H. R., Ihlebæk, C, & Ursin, H. (1999). A scoring system for subjective health complaints (SHC). Scandinavian Journal of Public Health, 27, 63-72.

Gallagher, S., Phillips, A. C., & Carroll, D. (2010). Parental stress is associated with poor sleep quality in parents caring for children with developmental disabilities. *J Pediatr Psychol, 35*(7), 728-737. doi: 10.1093/jpepsy/jsp093

Gallagher, S., Phillips, A. C., Oliver, C., & Carroll, D. (2008). Predictors of psychological morbidity in parents of children with intellectual disabilities. *J Pediatr Psychol, 33*(10), 1129-1136. doi: 10.1093/jpepsy/jsn040

Groth, K. A., Skakkebaek, A., Host, C., Gravholt, C. H., & Bojesen, A. (2013). Clinical review: Klinefelter syndrome--a clinical update. *J Clin Endocrinol Metab, 98*(1), 20-30. doi: 10.1210/jc.2012-2382

Grønholt, Else- Karin, Nordhagen, Rannveig, & Heiberg, Arvid. (2007). Mestring hos foreldre til barn med funksjonshemminger. *Tidsskrift Norsk Lægeforening, 127*(nr 4), 422-426.

Herlihy, A. S., McLachlan, R. I., Gillam, L., Cock, M. L., Collins, V., & Halliday, J. L. (2011). The psychosocial impact of Klinefelter syndrome and factors influencing quality of life. *Genet Med, 13*(7), 632-642. doi: 10.1097/GIM.0b013e3182136d19

Johnsen, T. Reidunn, Fegran, Liv, & Kristoffersen, Kjell. (2012). Hverdagslivet til foreldre som har barn med utviklingsmessige funksjonshemninger. *Vård i Norden, 32*(1), 9-13.

Linden, M. G., Bender, B. G., & Robinson, A. (1995). Sex chromosome tetrasomy and pentasomy. *Pediatrics, 96*(4 Pt 1), 672-682.

Lindstrom, Bengt, & Eriksson, Monica. (2006). Contextualizing salutogenesis and Antonovsky in public health development. *Health Promotion International, 29*(1), 238-244.

Meltzer, J. Lisa, & Mindell, A. Jodi. (2006). Impact of a Child's Chronic Illness on Maternal Sleep and Daytime Functioning *American Medical Association, 166*(1), 1749-1755.

Olsson, M. B., & Hwang, C. P. (2002). Sense of coherence in parents of children with different developmental disabilities. *J Intellect Disabil Res, 46*(Pt 7), 548-559.
Sigman, M. (2012). Klinefelter syndrome: how, what, and why? *Fertil Steril, 98*(2), 251-252. doi: 10.1016/j.fertnstert.2012.05.011

Simpson, J. L., de la Cruz, F., Swerdloff, R. S., Samango-Sprouse, C., Skakkebaek, N. E., Graham, J. M., Jr., . . . Paulsen, C. A. (2003). Klinefelter syndrome: expanding the phenotype and identifying new research directions. *Genet Med, 5*(6), 460-468. doi: 10.109701.GIM.0000095626.54201.D0

Sokol, R. Z. (2012). It's not all about the testes: medical issues in Klinefelter patients. *Fertil Steril, 98*(2), 261-265. doi: 10.1016/j.fertnstert.2012.05.026

Tartaglia, N., Ayari, N., Howell, S., D'Epagnier, C., & Zeitler, P. (2011). 48,XXYY, 48,XXXY and 49,XXXXY syndromes: not just variants of Klinefelter syndrome. *Acta Paediatr, 100*(6), 851-860. doi: 10.1111/j.1651-2227.2011.02235.x

Tartaglia, N. R., Ayari, N., Hutaff-Lee, C., & Boada, R. (2012). Attention-deficit hyperactivity disorder symptoms in children and adolescents with sex chromosome aneuploidy: XXY, XXX, XYY, and XXYY. *J Dev Behav Pediatr, 33*(4), 309-318. doi: 10.1097/DBP.0b013e31824501c8

van Rijn, S., Swaab, H., Aleman, A., & Kahn, R. S. (2008). Social behavior and autism traits in a sex chromosomal disorder: Klinefelter (47XXY) syndrome. *J Autism Dev Disord, 38*(9), 1634-1641. doi: 10.1007/s10803-008-0542-1

Visootsak, J., & Graham, J. M., Jr. (2006). Klinefelter syndrome and other sex chromosomal aneuploidies. *Orphanet J Rare Dis, 1*, 42. doi: 10.1186/1750-1172-1-42

Visootsak, J., & Tartaglia, N. (2013). *Autism and other neurodevelopmental disorders* R. L. Hanson & S. J. Rogers (Eds.), (pp. 306).

Walseth LT & Malterud K.: Salutogenese og empowerment I allmenn-medisinsk perspektiv. Tidsskrift for Den norske lægeforening, 1:8, 2004.

Wattendorf, D. J., & Muenke, M. (2005). Klinefelter syndrome. *Am Fam Physician, 72*(11), 2259-2262.

Challenges and ethical considerations

There is multiple challenges in this study. Few responders is one of them. Secondly, the sample includes boys within an age difference of 16 years. This means that there is a big variety of life situations, and both parents and the boys have all different life challenges.

There will always be ethical and methodological challenges when parents are asked to portray their own children, and how their children’s functioning or apparent lack of functioning will affect the reported parents’ quality of life. Parents may have a difficult time when describing their child as a burden, or a source of deprivation of their own life quality on a piece of paper. The lack of social contact between the person sending the questionnaire and the parent removes the social aspect and opportunities for discussion, clarifications and expressions of feelings. On the other side, distance from the investigator may create a space to be more honest about their child and their own daily life functioning and feelings. This brings issues of validity and reliability into question: are we actually finding out what we wanted to know, and doing so in trustworthy ways?

Another issue regarding generalizability is the severe under- diagnosing in SCA, which causes an issue about our sample and representativeness for the undiagnosed SCA population.

It can also be discussed how accurate quantitative method is for measuring people's quality of life. A quantitative survey measures the number and frequency of events and certain phenomena, but fundamentally challenging in terms of measuring quality of life. It is problematic to place specific numbers on thoughts and feelings, and most of subjective life experiences. Using a questionnaire with limited measurement scales is not easy when trying to provide and recreate a correct depiction of one’s life quality.

Another limitation to the questionnaires is the lack of Norwegian norms for all scales and methods. There is only implemented one measurement, and the parents have not been followed up over time. Finally, the results are based on parent-report only, which may also be a limitation to the validity of this study.

Final comments

The social- ecological perspective in this paper indicates that people's actions are a result of the structural opportunities in the contexts we live in. This helps explain how definitions of disabilities has changed through times. Understanding disability as a combination of social constructions together with a personal impairment has contributed to increase the rights of disabled people and to change their social status in society.

Growing up in society today is difficult for children with impairments and resultant social disabilities. It is not easy to crack the codes for how to look and act in order to belong in a societally- valued group when impairment makes you different and unequal among your classmates etc. Often joining extra-curriculum activities is often difficult or impossible for persons with different kinds of disabilities.

Having a child with a impairment is also difficult for parents. For most parents, the disabled child is a loss of the anticipations and dreams of the child you were expecting. Parents can be shocked and react by feeling incapable of action. Others behave seemingly calm and act restless. Some may react with a need to be close and protect the child, while others feel resentment towards the child that were different than they imagined. Also typical are feelings of anxiety and guilt. These reaction phases typically occur when parents realize that the child will not develop according to what is normally expected. They grieve for the loss of the child they looked forward to. Eventually, they begin to look forward, seeking and hoping for solutions. These are some of the shock and grief phases parents may go through after the knowledge of that their child has an impairment. How parents handle these phases are individual and depends on their sense of coherence. Parental resources and levels of how meaningful they find dealing with or solving the stressors determine how they meet and tackle the challenges of parenting a child with disabilities.

Strong or weak SOC may also explain the variety of health complaints and life satisfaction in our sample in the second paper. There was a wide range of scores on most of the question regarding parents' socio- emotional health and sleeping pattern. This second paper confirms that parenting a disabled child have an impact on parents' sleep quality. Sleep quality also have strong correlation with factors such as personal well- being, and subjective health, along with more of the strength and difficulties subscales in the children.

Self- reported living condition surveys and other surveys that rapports people's experience of health conditions, uses many different factors to identify the circumstances that overall explains how people evaluate their health. The knowledge we get from these surveys may contribute to increased awareness for professionals when working with this particular group of people, and prevent risk to develop poor health.

The main implication of this study is that parents of boys with sex chromosome aneuploidies would benefit from support by social services, by contributing to prevent development of symptoms of stress and anxiety. Findings in the study shows that poor functioning in the SCA-boys correlates with low life satisfaction and poor sleep quality for parents. This indicates that parents should be incorporated in the treatment and follow- up of their children with sex chromosome aneuploidies.

All analyzes were performed in SPSS (Statistical Package for the Social Science), an analytical tool for quantitative data used for statistical analysis.

Attachments:

Attachment 1: Background Questionnaire

Attachment 2: Subjective Health Complaints (SHC)

Attachment 3: Pittsbury Sleep Quality Index (PSQI)

Attachment 4: Strength and Difficulty Questionnaire (SDQ)

Attachment 5: Personal Wellbeing index (PWI)

Attachment 6: Behavioral Inhibition Questionnaire (BIQ 2)

Attachment 7: Consent for participation and purpose of the study

Attachment 1

Skjema om bakgrunn, utdanning, arbeid, helse, sosialt liv og fysisk aktivitet – foreldre

1 Bakgrunnsopplysninger

1.01 Barnets fødselsår: __________

Nåværende bostedsfylke________________________

Postnummer________________________

1.02 Vi bor i
a ☐ hus/rekkehus
b ☐ servicebolig
c ☐ bokollektiv
d ☐ leilighet
e ☐ annen bolig

1.03 Antall barn i husholdningen: ☐

1.04 a Barnets høyde i dag __________ cm

b Barnets vekt i dag __________ kg

2 Foreldres udanning

2.01 Sett kryss for den høyeste utdanningen du har fullført:

Barneskoleutdanning 1.-7. klassetrinn ☐

☐

Ungdomsskoleutdanning (8- 9/10. klassetrinn), realskole, framhaldsskole, handelsskole (2 år), yrkesskole (inntil 2 år etter folkeskole)

Videregående utdanning ☐

Universitets- og høgskoleutdanning, lavere nivå (2-3 år) ☐

Universitets- og høgskoleutdanning ☐

høyere nivå (mer enn 3 år)

Annet ☐

Hvis du er i tvil, skriv ned hvilken utdanning du har fullført______________________

3 Foreldres arbeid:

3.01 Betrakter du deg hovedsaklig som

a yrkesaktiv………………... ☐
b student eller skoleelev…… ☐
c alderspensjonist………….. ☐
d førtidspensjonist…………. ☐
e arbeidsufør………………. ☐
f hjemmearbeidende………. ☐

g arbeidsledig……………… ☐
h annet…………………….. ☐

3.02 Yrke (yrkestittel): ______________________________

4 Barnets helse

4.01 Vi ber deg krysse av for eventuelle helseplager eller diagnoser barnet ditt har eller utredes for/ får behandling eller oppfølging for:

Symptom	Ja	Under utredning	Får behandling/ oppfølging	Ingen symptomer	Vet ikke
Hudinfeksjoner og eksem…	☐	☐	☐	☐	☐
Leggsår….……………….	☐	☐	☐	☐	☐
Overvekt…………………	☐	☐	☐	☐	☐
Lavt stoffskifte……….…….	☐	☐	☐	☐	☐
Benskjørhet……………….	☐	☐	☐	☐	☐
Metabolsk syndrom……….	☐	☐	☐	☐	☐
Diabetes type 2…………..	☐	☐	☐	☐	☐
Diabetes type 1…………..	☐	☐	☐	☐	☐
SLE (lupus)…..…………..	☐	☐	☐	☐	☐
Høyt blodtrykk……………	☐	☐	☐	☐	☐
Tannstillingsfeil…………..	☐	☐	☐	☐	☐
Tannkjøttsykdom………..	☐	☐	☐	☐	☐
Gynekomasti (brystutvikling)	☐	☐	☐	☐	☐

Cryptorchidism (ikke nedvandrede testikler)…….	☐	☐	☐	☐	☐

4.02 Andre helseplager barnet har du vil nevne?

____

____

____

4.03 Barnet har smerter i muskler eller ledd

a ☐ aldri
b ☐ en sjelden gang
c ☐ hver måned
d ☐ hver uke
e ☐ hver dag
f ☐ annet

Beskriv

____

____

Hvis du svarte ”aldri” gå til spørsmål 4.05

4.04 Når barnet har smerter, er det vondt i (her kan du sette flere kryss ved behov)

a	☐ Hodet	f	☐ Hoftene
b	☐ Nakken	g	☐ Knærne
c	☐ Skuldrene	h	☐ Føttene
d	☐ Armene	i	☐ Annet
e	☐ Ryggen		

Dersom annet, beskriv: ____

4.05 Opplever du at barnets dagsform varierer mye?

☐ ofte ☐ av og til ☐ sjelden

4.06 Enkelte har ulike behov for søvn og kan oppleve uregelmessigheter knyttet til søvn

Jeg opplever ikke at dette gjelder mitt barn ☐

Jeg opplever at barnet mitt har følgende søvnproblemer (sett gjerne flere kryss):

a ☐ vansker med å sovne

b ☐ vansker med å stå opp om morgenen

c ☐ trenger mindre søvn enn hva som er vanlig

d ☐ trenger ekstra mye søvn

e ☐ annet

Beskriv____

4.09 Tar barnet medisiner regelmessig?

☐ ja ☐ nei

Hvis ja, hvilke og hvor ofte:____

5 Diagnose

5.01 a Hvor gammel var barnet da dere fikk diagnosen Klinefelters syndrom? _______ år

b Hvor gammel var barnet da han fikk vite at han har diagnosen Klinefelters syndrom? _______ år

c Ble diagnosen tilbakeholdt av legen?

☐ ja ☐ nei

d Ble diagnosen tilbakeholdt av dere overfor barnet?

☐ ja ☐ nei

5.02 Er det andre som har Klinefelters syndrom i familien deres?

Far	☐ ja	☐ nei
Søsken	☐ ja	☐ nei
Andre	☐ ja	☐ nei

5.03 a Er det avklart hvilken kromosomfeil eller karyotype barnet har? (for eksempel 47xxy, 46xy/47xxy)

☐ ja ☐ nei ☐ vet ikke

b Hvis ja, gjengi/beskriv karyotype: ________________________________

5.04 a Får barnet testosteronbehandling?

☐ ja ☐ nei

b Hvis ja, hvor gammel var han da den startet? ____ år

c I hvilken form får han tilført testosteron?

a ☐ Plaster
b ☐ Gel
c ☐ Tabletter
d ☐ Sprøyter

d Har han hatt opphold i behandlingen?

☐ ja ☐ nei ☐ vet ikke

Hvis ja, beskriv hvorfor ____

e Hvis du opplever resultater av behandlingen; på hvilke områder gjelder dette? (sett gjerne flere kryss)

a ☐ Konsentrasjon

b ☐ Overskudd
c ☐ Søvn
d ☐ Behåring
e ☐ Libido
f ☐ Kroppsform/muskelmasse
g ☐ Humør
h ☐ Annet ________________________________

5.07 Hvem følger dere opp med hensyn til Klinefelters syndrom?

Ingen……………………….	☐
Fastlege…………………….	☐
Endokrinolog (hormonlege)	☐
Habiliteringstjenesten….….	☐
Ortoped……………………	☐
Hjertelege…………………	☐
Psykolog…….……………	☐
Psykiatrisk sykepleier.……	☐
Annet………………………	☐

Dersom andre, beskriv____

5.08 Er du fornøyd med tilbudet helsevesenet gir deg i forbindelse med barnets Klinefelters syndromdiagnose? Sett ett kryss.

Svært misfornøyd	Misfornøyd	Verken misforn. eller fornøyd	Fornøyd	Svært Fornøyd
☐	☐	☐	☐	☐

Tusen takk for hjelpen!

Attachment 2

SHC- questionnaire

Helseproblemer siste 30 døgn

På den neste siden nevnes noen vanligehelseplager. Vi vil be deg om å vurdere hvert enkelt problem/symptom, og oppgi **i hvilken grad duhar vært plaget** av dette i løpet av de siste tretti døgn, og **antall dager** du har vært plaget.

Eksempel

Hvis du føler at du har vært *en del* plaget med forkjølelse/influensa siste måned,og varigheten av plagene var *ca. en uke*, fylles dette ut på følgende måte:

Sett ring rundt tallet som passer best.

Nedenfor nevnes noen alminnelige helseproblemer	Ikke plaget	Litt plaget	En del plaget	Alvorlig plaget	Antall dager plagene varte (omtrent)
1. Forkjølelse, influenza	0	1	(2)	3	7

NB! Det er viktig at du fyller ut både *hvor plaget* du har vært, og *omtrentantall dager* du har vært plaget siste tretti døgn.

Helseproblemer siste 30 døgn

Nedenfor nevnes noen alminnelige helseproblemer (sett ring rundt tallet som passer)	Ikke plaget	Litt plaget	Endel plaget	Alvorlig plaget	Antall dager plagene varte (omtrent)
1. Forkjølelse, influensa................	0	1	2	3	…………
2. Hoste, bronkitt	0	1	2	3	…………
3. Astma	0	1	2	3	…………
4. Hodepine	0	1	2	3	…………
5. Nakkesmerter	0	1	2	3	…………
6. Smerter øverst i ryggen.............	0	1	2	3	…………
7. Smerter i korsrygg	0	1	2	3	…………
8. Smerter i armer	0	1	2	3	…………
9. Smerter i skuldre	0	1	2	3	…………
10. Migrene	0	1	2	3	…………
11. Hjertebank, ekstraslag...............	0	1	2	3	…………
12. Brystsmerter.............................	0	1	2	3	…………
13. Pustevansker	0	1	2	3	…………
14. Smerter i føttene ved anstrengelser	0	1	2	3	…………
15. Sure oppstøt, «halsbrann»..........	0	1	2	3	…………
16. Sug eller svie i magen...............	0	1	2	3	…………
17. Magekatarr, magesår.................	0	1	2	3	…………
18. Mageknip	0	1	2	3	…………
19. «Luftplager»..............................	0	1	2	3	…………
20. Løs avføring, diaré....................	0	1	2	3	…………
21. Forstoppelse..............................	0	1	2	3	…………
22. Eksem.......................................	0	1	2	3	…………
23. Allergi	0	1	2	3	…………
24. Hetetokter.................................	0	1	2	3	…………
25. Søvnproblemer..........................	0	1	2	3	…………

26.	Tretthet......................................	0	1	2	3	………….
27.	Svimmelhet................................	0	1	2	3	………….
28.	Angst...	0	1	2	3	………….
29.	Nedtrykt, depresjon....................	0	1	2	3	………….

Id nr______________ Dato_____________

PSQI – foreldre om seg selv

Følgende spørsmål har med ditt vanlige søvnmønster *den siste måneden* å gjøre. Du skal svare på hva som er mest riktig for *de fleste* dager og netter den siste måneden. Vennligst svar på alle spørsmål. Kryss av i den nederste ruten for det svaret du syns passer best. Vennligst svar på *alle* spørsmålene.

1. I løpet av den siste måneden, hvor ofte har du hatt problemer med søvnen fordi du…

(a) ikke klarer å sovne i løpet av 30 minutter?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

(b) våkner opp midt på natten eller tidlig om morgenen?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

(c) må opp for å gå på toalettet?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

(d) ikke klarer å puste ordentlig?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

(e) hoster eller snorker høyt?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

(f) føler deg for kald?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

(g) føler deg for varm?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

(h) har vonde drømmer?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

Id nr______________ Dato_____________

(i) har smerter?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

(j) andre grunner?
Vennligst beskriv:

____

____

Hvor ofte, løpet av den siste måneden, har du hatt problemer med søvnen på grunn av dette?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

2. I løpet av den siste måneden, hvordan vil du bedømme søvnkvaliteten din totalt sett?

Veldig bra	Ganske bra	Ganske dårlig	Veldig dårlig

3. I løpet av den siste måneden, hvor ofte har du tatt medisin (med eller uten resept) som hjelp til å sove?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

4. I løpet av den siste måneden, hvor ofte har du hatt problemer med å holde deg våken under bilkjøring, måltider eller når du holder på med sosiale aktiviteter?

Ikke i den siste måneden	Mindre enn 1 gang i uka	1 eller 2 ganger i uka	3 eller flere ganger i uka

5. I løpet av den siste måneden, hvor stort problem har det vært for deg å ha overskudd nok til å få ting gjort?

Ikke noe problem i det hele tatt	Bare et lite problem	Et visst problem	Et stort problem

---

Pittsburgh Sleep Quality Index

(Buysse, Reynolds III, Monk, Berman & Kupfer, 1989)

Til norsk ved Petter Franer, Inger Hilde Nordhus, Ståle Pallesen og Simen Øverland

Noe forkortet for denne studien ved K. Fjermestad, Frambu

Sterke og svake sider (SDQ- nor) Attachment 4

Barnets navn…………………………………….. Gutt/jente

Fødselsdato………………………………

	Stemmer ikke	Stemmer delvis	Stemmer helt
Omtenksom, tar hensyn til andre menneskers følelser	☐	☐	☐
Rastløs, overaktiv, kan ikke være lenge i ro	☐	☐	☐
Klager ofte over hodepine, vondt i magen eller kvalme	☐	☐	☐
Deler gjerne med andre barn (godter, leker, andre ting)	☐	☐	☐
Har ofte raserianfall eller dårlig humør	☐	☐	☐
Ganske ensom, leker ofte alene	☐	☐	☐
Som regel lydig, gjør vanligvis det voksne ber om	☐	☐	☐
Mange bekymringer, virker ofte bekymret	☐	☐	☐
Hjelpsom hvis noen er såret, lei seg eller føler seg dårlig	☐	☐	☐
Stadig urolig eller i bevegelse	☐	☐	☐
Har minst en god venn	☐	☐	☐
Slåss ofte emd andre barn eller mobber dem	☐	☐	☐
Ofte lei seg, nedfor eller på gråten	☐	☐	☐
Vanligvis likt av andre barn	☐	☐	☐
Lett avledet, mister konsentrasjonen	☐	☐	☐
Nervøs eller klengetei nye situasjoner, lett utrygg	☐	☐	☐
Snill mot yngre barn	☐	☐	☐
Lyver eller jukser ofte	☐	☐	☐
Plaget eller mobbet av andre barn	☐	☐	☐
Tilbyr seg ofte å hjelpe andre (foreldre, lærere, andre barn)	☐	☐	☐
Tenker seg om før han/hun handler (gjør noe)	☐	☐	☐

Stjeler hjemme, på skolen eller på andre steder	☐	☐	☐
Kommer bedre overens med voksne enn med barn	☐	☐	☐
Redd for mye, lett skremt	☐	☐	☐
Fullfører oppgaver, god konsentrasjonsevne	☐	☐	☐

Underskrift………………………………   Dato……………………

Mor/ Far/ Lærer / Andre (vennligst beskriv):

Tusen takk for hjelpen

Attachment 5

Personal Wellbeing Index

Part 1: Life Satisfaction Overall

Thinking about your own life and personal circumstances, how satisfied are you with your life as a whole?

Respondent’s raiting

Part 2: Domain Life Satisfaction

(0-10)

1. “How satisfied are you with your standard of living?” ____
2. “How satisfied are you with your health?” ____
3. “How satisfied are you with what you are achieving in life?” ____
4. “How satisfied are you with your personal relationships?” ____
5. “How satisfied are you with how safe you feel?” ____
6. “How satisfied are you with feeling part of your community?” ____
7. “How satisfied are you with your future security?” ____

Attachment 6

BIS 2 - Foreldre

Følgende utsagn beskriver barns atferd i ulike situasjoner i småbarnsalderen. For hvert utsagn bes du vurdere hvorvidt barnet ditt viser denne atferden «nesten aldri», «sjelden», «en gang i måneden», «noen ganger», «ofte», «svært ofte», eller «nesten alltid».

Prøv å gjøre vurderingen så godt du kan basert på hvordan du nå i ettertid ser barnet ditt sammenlignet med andre barn på samme alder. Vurder barnet ditt i forhold til hvordan han var i førskolealderen (ca. 3-5 år gammel).

1= «nesten aldri», 2= «sjelden», 3= «en gang i blant», 4= «noen ganger», 5= «ofte», 6= «svært ofte», 7= «nesten alltid».

	I førskolealder
1. Var nølende i nye situasjoner eller aktiviteter	1 2 3 4 5 6 7
2. Gikk gjerne bort til en gruppe ukjente barn for å være med i leken deres	1 2 3 4 5 6 7
3. Var svært stille når vi hadde nye (voksne) gjester hjemme hos oss.	1 2 3 4 5 6 7
4. Var forsiktig i aktiviteter som innebar fysiske utfordringer (for eksempel klatre, hoppe fra høyder)	1 2 3 4 5 6 7
5. Fant seg fort til rette når vi besøkte folk vi ikke kjente så godt	1 2 3 4 5 6 7
6. Likte å være senter for oppmerksomhet	1 2 3 4 5 6 7
7. Hadde lett for å spørre andre barn om å være med å leke	1 2 3 4 5 6 7
8. Var sjenert i møte med andre barn	1 2 3 4 5 6 7
9. Var glad og fornøyd første gang foreldre gikk fra ham i nye situasjoner (for eksempel dagmamma, barnehage)	1 2 3 4 5 6 7
10. Likte å opptre foran andre (for eksempel synge, danse)	1 2 3 4 5 6 7
11. Fant seg fort til rette i nye situasjoner (for eksempel dagmamma, barnehage)	1 2 3 4 5 6 7
12. Vegret seg for å spørre om å få være med sammen med en gruppe ukjente barn	1 2 3 4 5 6 7
13. Var trygg i aktiviteter med fysiske utfordringer (for eksempel klatre, hoppe fra høye steder)	1 2 3 4 5 6 7

14. Var avhengig	1 2 3 4 5 6 7
15. Så ut til å ha det bra i nye situasjoner	1 2 3 4 5 6 7
16. Var svært pratsom overfor voksne som han ikke kjente	1 2 3 4 5 6 7
17. Nølte med å utforske nye leker	1 2 3 4 5 6 7
18. Ble ute av seg første gang han ble overlatt til seg selv i nye situasjoner (for eksempel barnehage, dagmamma)	1 2 3 4 5 6 7
19. Var svært vennlig overfor barn han nettopp hadde møtt	1 2 3 4 5 6 7
20. Pleide heller å betrakte andre barn enn å delta i leken deres	1 2 3 4 5 6 7
21. Mislikte å være sentrum for oppmerksomhet	1 2 3 4 5 6 7
22. Var klengete når vi besøkte folk vi ikke kjente så godt	1 2 3 4 5 6 7
23. Likte seg i nye situasjoner og aktiviteter	1 2 3 4 5 6 7
24. Var utadvendt	1 2 3 4 5 6 7
25. Virket nervøs eller utilpass i nye situasjoner	1 2 3 4 5 6 7
26. Pratet gjerne med nye (voksne) gjester hjemme hos oss	1 2 3 4 5 6 7
27. Brukte mange dager på å venne seg til nye situasjoner (for eksempel dagmamma, barnehage)	1 2 3 4 5 6 7
28. Hadde ikke så lyst å opptre foran andre (for eksempel synge, danse)	1 2 3 4 5 6 7
29. Utforsket gjerne nye leker	1 2 3 4 5 6 7
30. Var stille overfor fremmede voksne	1 2 3 4 5 6 7

Original versjon Susan H. Spence (2003). Oversatt og tilpasset av Bente Storm Mowatt Haugland og Kristin Øding (UiB) og Hanne Kristensen (R- BUP Øst og Sør), 2007

Attachment 7

Forespørsel om deltakelse i forskningsprosjektet

”Psykososial helse blant personer med Klinefelters syndrom og andre kjønnskromosomforstyrrelser”

Foreldreversjon

Bakgrunn og hensikt

Dette er et spørsmål til deg om å delta i en forskningsstudie som har som mål å få økt kunnskap om livskvalitet ved Klinefelters syndrom (KS) og andre kjønnskromosomforstyrrelser. Frambu, som er et landsdekkende kompetansesenter for disse diagnosene, er ansvarlig for studien. Studien har spesielt fokus på livskvalitet og mental helse hos barn og voksne med disse diagnosene.

Frambu har ansvar for å samle, systematisere og spre kunnskap om en rekke sjeldne og lite kjente diagnoser, inkludert KS og andre kjønnskromosomforstyrrelser. Personer med kjønnskromosomforstyrrelser beskrives i ulike studier å ha varierende grad av kognitive, emosjonelle og fysiske vansker. Målet for denne studien er å kartlegge hvordan det oppleves å leve med en kjønnskromsomforstyrrelse. Spørsmålene vil blant annet dreie seg om barnets helse, mestring og læring, atferd og kommunikasjon. Frambu ønsker å foreta en slik kartlegging i håp om å få økt kunnskap om, og forståelse for, hvordan det er å være barn og voksen med slik diagnose i Norge. På bakgrunn av dette vil informasjonen og veiledningen som gis rundt slik diagnose forhåpentligvis bli bedre og mer adekvat, både til personer med diagnosene, deres foresatte og fagpersoner.

Frambu har pr. desember 2011 registrert 44 barn med ulike kjønnskromosomforstyrrelser, og du mottar denne forespørselen som følge av at du er forelder til en av disse.

Hva innebærer studien?

Studien tar for seg flere ulike livsområder for å gi en bred kartlegging av livskvalitet, mestring og mental helse. I denne studien er det ønskelig at du gir en rangering av disse områdene ved å besvare de vedlagte spørreskjemaene. I tillegg vet vi at foreldre til barn med ulike funksjonsnedsettelser oftere har psykososiale vansker, og derfor vil vi spørre deg som er foreldre om din fysiske og psykiske helse, samt søvnkvalitet, gjennom ulike skjema.

Bakgrunnsskjemaet innholder noen spørsmål som bare vil være aktuelle for de med overtallig X. Dette er spørsmål rundt testosteronbehandling, som ikke er aktuelt for de med kromosombilde 47 XYY.

Vi ber samtidig om at du fyller ut samtykkeskjemaet, som er en forutsetning for å delta i studien. Når samtykkeskjemaet og spørreskjemaene er ferdig utfylt ber vi deg om å returnere disse i ferdig frankert konvolutt som er vedlagt.

Når skjemaene er returnert til oss vil vi samle informasjonen og undersøke resultatene hos personer med KS og andre kjønnskromosomforstyrrelser sammenlignet med en kontrollgruppe, for å se om det er noen forskjeller i opplevd livskvalitet.

Mulige fordeler og ulemper

Det kan ta noe tid å fylle ut skjemaene og noen kan oppleve enkelte spørsmål som ubehagelig eller lite relevante. Vi håper imidlertid at deltakelse i prosjektet vil oppleves som en mulighet til å bidra til økt forståelse for, og kompetanse rundt, livskvalitet for barn og voksne med KS og andre kjønnskromosomforstyrrelser.

Hva skjer med informasjonen om deg og ditt barn?

Informasjonen som registreres fra de innsendte spørreskjemaene skal kun brukes slik som beskrevet i hensikten med studien. Alle opplysningene vil bli behandlet uten navn og fødselsnummer eller andre direkte gjenkjennende opplysninger. En kode knytter barnets navn til dine opplysninger gjennom en navneliste. Navnelisten oppbevares separat fra spørreskjemaene.

Det er kun autorisert personell knyttet til prosjektet som har adgang til navnelisten og som kan finne tilbake til deg. Det vil ikke være mulig å identifisere deg i resultatene av studien når disse publiseres. Datamaterialet, altså all innsamlet informasjon, vil bli slettet gjennom makulering av spørreskjemaene når studien er avsluttet, senest innen utgangen av 2013.

Informasjon om utfallet av studien

Resultatene fra studien vil bli publisert i ulike artikler. Resultatene vil også kunne bli publisert og benyttet i Frambus dokumentasjons- og informasjonsarbeid.

Frivillig deltakelse

Det er frivillig å delta i denne studien, og om du deltar eller ikke vil ikke ha noen innvirkning på fremtidig tilbud på eller fra Frambu. Dersom du ønsker å delta, undertegner du samtykkeerklæringen og sender inn denne sammen med de utfylte spørreskjemaene. Du kan når som helst og uten å oppgi noen grunn trekke ditt samtykke til å delta i studien. Dersom du har spørsmål knyttet til studien, eller på et senere tidspunkt ønsker å trekke deg kan du kontakte prosjektleder Krister Fjermestad tlf. 64 85 60 00 eller prosjektmedarbeider Simen Stokke 64 85 60 41.

Kontaktinformasjon

Spørsmål om studien kan rettes til:

Krister Fjermestad	Frambu	eller	Simen Stokke	Frambu
Telefon	64 85 60 00		Telefon	64 85 60 41
E-post	kfj@frambu.no		E-post	sis@frambu.no

Samtykke til deltakelse i studien

”Psykososial helse blant personer med Klinefelters syndrom og andre kjønnskromosomforstyrrelser”

Jeg har mottatt og lest informasjon om studien ”Psykososial helse blant personer med Klinefelters syndrom og andre kjønnskromosomforstyrrelser”, og samtykker til å besvare spørreskjema om meg selv og barnet mitt.

Jeg er villig til å delta i studien

……………………………………………………………………………………………………...

Sted **Dato** **Underskrift av prosjektdeltaker**