Anorectal malformations and Hirschsprung disease
Functional and psychosocial challenges
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Chapter 9
General discussion and future perspectives
General discussion and future perspectives

The aim of this thesis was to identify the functional and psychosocial problems patients with an anorectal malformation or Hirschsprung disease can encounter during life, and to investigate the psychosocial impact these diagnoses can have on parents. We hypothesized that a multidisciplinary approach of the treatment of children, adolescents and adults with an anorectal malformation or Hirschsprung disease is essential for both functional and psychosocial outcome. And that having a child with an anorectal malformation or Hirschsprung disease can cause psychosocial and psychological problems for parents.

Quality of Life

QoL has become an important aspect in the treatment of many chronic diseases in the last decades. In recent years, QoL of patients with an anorectal malformation or Hirschsprung disease has become a topic for many authors. A problem in QoL research is that many different questionnaires are used and different concepts are labelled as QoL. A definition of QoL must be given in order to know what was investigated. In addition a distinction has to be made between “QoL” and “health-related QoL”.

QoL is a multidimensional concept assessing function and satisfaction in all aspects of life. In other words, QoL is one’s evaluation of his/her functioning in a wide range of areas. QoL is subjective and refers to satisfaction. Health related QoL only measures those aspects that are influenced by one’s health. To measure QoL, questionnaires can be used. A questionnaire measuring all aspects of QoL includes six domains: the physical domain, the psychological domain, the social domain, environment, level of independence and spirituality. We prefer that QoL questionnaires are completed by patients themselves, however in certain circumstances, for example a very young patient, they can be completed by proxies of the patient. However, it is known from literature that parents and siblings of patients can overrate or underestimate the QoL of a patient.

Also the construction of a questionnaire has influence on the outcome of a study. A questionnaire assessing satisfaction with function, such as the WHOQOL-100, reveals different areas of concern compared to a questionnaire that inquires merely on function in different domains, such as the SF-36. A questionnaire as the WHOQOL-100 reflects the (dis)satisfaction of the patient and this does not necessarily mirror the functional limitations. Therefore using validated QoL questionnaires is an essential element in order to conduct “high quality” QoL research. Also defining what you want to measure before starting QoL research will help you use the best questionnaire for your hypothesis.
Quality of life is not a static concept. It may fluctuate over time because of certain life events. Also coping strategies of a person change over time, which can influence a person’s QoL. QoL research will be more representative when set up as a longitudinal study.

In Chapter 2 we performed a qualitative analysis of studies concerning QoL in patients with an anorectal malformation. Almost all studies performed on QoL in patients with an anorectal malformation were of “low quality” or “moderate quality”. Real conclusions on QoL therefore could not be drawn from these studies. All studies did advocate to perform longitudinal research on this topic. Therefore, we started the KLANKbord-study, a multicentre, longitudinal QoL study of patients with an anorectal malformation or Hirschsprung disease and their parents. With this study we hope to provide more results on QoL of patients and parents in a longitudinal design. Furthermore, we want to see whether it is possible to identify those moments in life where our patients or their parents could benefit from more intensive treatment, both for functional and psychosocial problems.

**Anxiety, parental stress and QoL of parents of children with an anorectal malformation or Hirschsprung disease**

A novelty of this thesis is the fact that anxiety, stress and QoL of parents of patients were investigated. This subject is often overlooked. We feel that the QoL of a child is largely influenced by (the QoL of) his or her parents. Literature shows that psychosocial problems of parents can indeed affect the well-being of children. Therefore, we hypothesized that if parents suffer from having a child with an anorectal malformation or Hirschsprung disease, they may not be able to take good care of their child, and that may in turn affect the QoL of the child.

Parental stress has been investigated in other chronic diseases before, but not in parents of children with an anorectal malformation or Hirschsprung disease.

In Chapter 3 and Chapter 4 we investigated anxiety and QoL of parents. Our hypothesis was that parents will experience anxiety after having a child with an anorectal malformation or Hirschsprung disease. We also assumed that anxiety levels would be different between mothers and fathers. And we hypothesized that this anxiety could lead to a decreased QoL. QoL refers to (dis)satisfaction and can vary over time. Therefore we assumed that anxiety levels of parents would change over time.

Mothers of newborn children with an anorectal malformation or Hirschsprung disease showed more anxiety than fathers did. Also psychosocial QoL of mothers of newborns was decreased compared to fathers. This difference between parents was not measured.
anymore after one year. Mothers scored slightly lower, and fathers experienced slightly more anxiety.\textsuperscript{14}

The difference in anxiety between mothers and fathers was also not measured in parents of older children.\textsuperscript{15} When we compared overall QoL of parents of children with an anorectal malformation or Hirschsprung disease with known reference values, no negative influence was seen.\textsuperscript{14}

Interesting about these results is the fact that we now know that we can expect anxiety levels to be comparable between parents after one year and also that we can expect overall QoL of parents not to be impaired. The exact reason why this anxiety reduces over time, has not been identified in these studies. It could be the result of coping strategies or maybe the result of better functioning of the children but we do know that if we encounter parents that still experience psychosocial problems a year after the birth of their child or after the diagnosis was made, they may need psychosocial care or intervention programs. Programs focusing on psychological interventions and counselling showed a positive influence on anxiety, and a better use of problem-focused coping strategies in other patient populations.\textsuperscript{16,17}

Because of the chronic character of these diseases and possible ongoing treatments, hospital visits and functional problems, parents can experience stress. In \textit{Chapter 5} parental stress of parents was measured using the Pediatric Inventory for Parents (PIP). This instrument investigates areas of anxiety and concern experienced by parents caring for an ill child without being limited to a specific illness. It combines disease related measures with generic measures so that these dimensions can be investigated in a quantitative fashion. One of the assets of the PIP is that parents are asked to rate both the frequency (PIP-F) of stressful illness-related events and the difficulty (PIP-D) they experience with these events since this gives a better insight on both coping and stressors.\textsuperscript{18}

Levels of parental stress of parents of children with an anorectal malformation or Hirschsprung disease were comparable to other chronic diseases, such as inflammatory bowel disease or diabetes.\textsuperscript{11,12} In this study we measured that the parental stress does not reduce during the first 3 years after the diagnosis, meaning that psychosocial interventions focussing on stress reduction may be useful for these parents even years after the initial diagnosis was made.

\textbf{Long term outcome and QoL in patients with (near) total colonic aganglionosis}

The second half of this thesis is more focussed on the long term outcome of patients with an anorectal malformation or Hirschsprung disease. We investigated functional problems and QoL in different patient categories: children, adolescents and adults.
In Chapter 6 patients with (near) total colonic aganglionosis (nTCA) were investigated. A consensus on the best surgical technique for this type of Hirschsprung disease is currently not available worldwide. Patients with nTCA are known to suffer from more functional complaints than patients with a classic short segment Hirschsprung.\textsuperscript{19-22} We performed a national study on functional results and QoL of this specific patient group. We found that the functional complaints were much higher, when reported by patients, using questionnaires, than reported in the charts. Earlier research done in this patient population was almost all done by chart review, meaning that problems could have been underestimated.\textsuperscript{20,21,23,24} QoL in patients with Hirschsprung disease has only been described in a limited number of studies, often without the use of standardised and validated questionnaires.\textsuperscript{20,21,23,25-35} In our study children between 4 and 17 years of age had a diminished overall QoL compared to reference values of healthy peers. Patients with nTCA of 18 years and older had a QoL comparable to their healthy peers despite their functional problems.

A superior technique could not be identified in this study because we felt that statistics could not be done in this group, due to the relatively low number of patients per surgical technique, and also because of the different lengths of follow-up.

**Transitional outpatient clinic for patients with an anorectal malformation or Hirschsprung disease**

The care for patients with an anorectal malformation or Hirschsprung disease is performed by the pediatric surgeon. This covers the initial consult, the operation and the routine follow-up. When patients are 18 years or older, we unfortunately noticed that often this follow-up is discontinued. Partially due to the patient, but also because of the fact that many adult-centered-physicians are not familiar with these diagnoses. We think that this problem can be solved by the availability of a transitional outpatient clinic.

Transitional care is defined as a set of actions designed to ensure the coordination and continuity of health care and safe and timely transfers, as patients transfer between different locations or different levels of care within the same location or in a different setting.\textsuperscript{36,37} In the Pediatric Surgical Center of Amsterdam, we started this transitional outpatient clinic in 2012. The design of the clinic and the use of questionnaires were evaluated in Chapter 7, starting in adult patients. Only 17% of the invited patients made an appointment to come to our clinic. Questionnaires on functional problems and QoL were completed in advance. It was difficult to express the usefulness of the questionnaires in hard numbers, however the experience of both caregivers and patients was positive. It does alleviate the sense of taboo around the subject of defecating and makes problems comprehensible. In individual cases it was observed that complaints were not reported during consultation, but became apparent through questionnaire scores. Also in cases in which a good overall QoL was reported by the patient, significant impairment
in specific aspects of QoL was found. More and better “tailor made” personal care could be given to the patient. The overall experience of the patients that visited the clinic was good and they were satisfied by the advice and treatment given to them. Many patients in this study lived with their complaints for years before they visited our clinic. They did not know where or whom to turn to, and felt not understood by adult caregivers, who did not have sufficient knowledge of their pathology. Starting this transitional care at 12-18 years of age in the future will hopefully identify problems earlier and make the transition into adult care easier.

**Predicting sexual problems in patients with an anorectal malformation or Hirschsprung disease**

Sexuality is a topic not often discussed by the pediatric surgeon with his/her patients. However we know from earlier research that adult patients with an anorectal malformation of Hirschsprung disease did miss discussing this with their doctor when they were adolescents. Patients with anorectal malformations can also have associated urogenital disorders making them more prone to the development of sexual problem in later life. Patients with Hirschsprung disease most often do not have associated urogenital disorders, but the problems, such as soiling or the fear of being continent while having sexual intercourse can be very limiting. We have seen in Chapter 8 of this thesis that sexual functioning in men and women in adulthood is impaired, and that psychosocial factors, such as self-competence, relationships with peers and body image during adolescence are correlated with sexual functioning and distress in adulthood. These results suggest that we can identify patients during adolescence, that would be more prone to develop sexual problems in later life. These psychosocial factors can be identified during follow-up in the out-patient clinic. A more standardized care and follow-up schedule is needed. Therefore, we believe that the formation of a multidisciplinary (transitional) team is an essential element in the treatment of our patients.

**Limitations of this thesis**

Performing research on patients with a congenital disorder can be challenging. In the Netherlands approximately 80 patients are born each year with an anorectal malformation or Hirschsprung disease a year. Unfortunately not all pediatric surgical centres in the Netherlands participated in the longitudinal study (KLANKbord-study) and therefore the number of patients that were analysed was relatively low in some studies. Also the fact that the study is longitudinal and patients and parents need to complete questionnaire at different moments in time can be a problem. Drop outs during the study may influence the results.
Future perspectives and clinical implications

Our goal is that the KLANKbord study will continue for many years to come. As mentioned earlier, knowledge and treatment options of patients with chronic diseases have become better during the last few decades, meaning that these patients will more often reach adulthood or even have the same life expectancy as their healthy peers. Therefore, more attention should be paid to their QoL. The KLIK method is a web based method in which QoL of children and parents is assessed, investigated and discussed. We hope that our study can be incorporated in KLIK in the future.

In this thesis, we have concluded that having a child with an anorectal malformation or Hirschsprung disease can have psychological and psychosocial impact on parents. Healthcare providers should focus more on these problems when treating a child with one of these diagnoses. The additive value of psychosocial interventions has not been investigated in this patient group yet. Therefore future research on psychosocial intervention programs is essential.

Furthermore, it is important that every Pediatric Surgical Center that treats patients with an anorectal malformation or Hirschsprung disease has a multidisciplinary (transitional) outpatient clinic. In this team at least a pediatric surgeon, urologist, gynaecologist, stoma and incontinence nurse and a social worker should be incorporated to provide all the care that is needed. For the transitional clinic an adult urologist, adult gynaecologist, adult gastrointestinal surgeon should be added to this team.

We believe that with this thesis important aspects of QoL and functional problems of patients and their parents have been addressed. And we believe that with a more multidisciplinary approach the treatment of patients with an anorectal malformation or Hirschspung disease and their parents will be more complete and lifted to the next level.
Reference List

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