Summary

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Hand function is of extreme importance in interacting with our environment. From the moment we rise until the moment we go to bed our hands perform numerous daily activities. Normal daily activities may become challenges if hand function is compromised. For instance, only a cutting wound at the top of the index finger may force you to use the hand differently for a couple of days. What if hand function is compromised since you were born? Will you encounter problems in feeding, bathing, or leisure activities? Will you be able to manage household activities? How will you experience your quality of life? Exactly these questions arise in the minds of parents who are faced with a child with a congenital hand difference (CHD). In this thesis, we try to find answers on these questions. For this purpose, we conducted two cross-sectional studies. This thesis presents these studies on HRQoL and daily functioning in 10-14 year old children with a CHD and in children and adults with Apert Syndrome.

The introductory **Chapter 1** describes congenital hand differences (CHD) and its possible consequences. These consequences are described using the international classification of functioning, disability and health, the child and youth version (ICF-CY). This model shows how the different domains and child's characteristics may interact. Although health-related quality of life (HRQoL) officially stands outside the ICF-CY framework, it has close relations with the domains of activities and participation.

After discussing HRQoL of our patient group, we follow the top-down approach that we practise in our institution in older children with CHD. We start with the "complex comprehensive " level of functioning, social participation, and end with the "basic" level, functions of the hand, i.e. mobility and muscle strength.

In **Chapter 2**, we compared HRQoL, as measured with the PedsQL generic core scales, in children with CHD with healthy peers. Additionally, we examined the associations between HRQoL, severity of the CHD and ease of activity performance at the activity level. Except for a lower score on social functioning in children aged 13-14 years, children with CHD did not score lower than their healthy peers. When looking at predictors of HRQoL, we found that if activities were performed more easily, this lead to higher HRQoL scores. On the other hand, the presence of comorbidity lowered the scores on all HRQoL subdomains, except for school functioning. Additionally, we described the positive effect of more affected digits and the negative effect of age, bilateral involvement and ethnicity on some subdomains. We concluded that we can reassure parents that their child with CHD will probably rate their own HRQoL as high as that of healthy peers.

Knowledge on the child's HRQoL can be of major importance to the child's physician. In some cases, however, it is not possible to obtain the child's opinion. In those cases, a parent's or caregiver's opinion is often taken as a representative substitute. In **Chapter 3**,

we investigated whether results on HRQoL obtained by measuring the parents' opinion are indeed a representative substitute for the child's opinion. We found that, on group level, the children scores did not differ from their parents; both children and their parents scored high on a scale of 0-100: physical health: 89.1 (SD:14.1) versus 88.0 (SD:15.6), psychosocial health: 80.6 (SD:13.4) versus 79.0 (SD:14.5) and total HRQoL: 83.5 (SD:12.3) versus 82.0 (SD:13.6). In contrast, on individual level, scores showed high variation, with children reporting both higher and lower scores than their parent proxy. The limits of agreement are large and on social functioning and emotional functioning even as large as 30 points on the 0 -100 scale. Despite of this variation, we were not able to detect major determinants for agreement; we only found that children with more affected fingers agreed more with their parents on emotional functioning. This was also true for the agreement on social functioning in bilaterally involved children. Therefore, we suggest that care should be taken in choosing the parents' score as a representative substitute for the child's score and they should not be used interchangeably. For clinical use, we advise to make decisions based on one report, when possible the child's self-report.

To get an impression of the functioning of patients with CHD in the presence of a syndrome, we conducted a cross-sectional study in children and adults with Apert Syndrome **(Chapter 4)**. We found that upper-extremity activity scores were comparable with scores of children with cerebral palsy. Since reference values of healthy peers are lacking, comparison with those children was not possible, but children in our sample scored 60% of the maximal scores. For the adults, upper-extremity activity scores are worse than healthy peers or patients with radial dysplasia after centralization of the wrist, but comparable to a large group of patients with injuries or clinical conditions of the upper limb. For lower limb activity, the scores on the LEFS showed large variance within the group. The adults in our sample scored better than patients with hip or knee osteoarthrosis. Social participation in children with Apert Syndrome score was similar to their healthy peers, regardless of the type of hand. Even so, the adult group did not perceive large restrictions in participation. For HRQoL, all children experience more limitations on self-esteem, emotional parental impact, general health and impact on parental time than healthy peers, but family cohesion is higher in all children with Apert Syndrome. Some subgroups score lower on different domains. All adults in our study sample experienced more limitations on physical functioning, but experienced less pain and felt less limited in roles due to physical problems, or due to emotional problems than the Dutch norm group.

Children with CHD may perceive several problems in performing daily activities. While their surrounding world is designed for two-handed use, they sometimes need to perform activities with one hand. **Chapter 5** examines the bimanual performance of children with CHD using the Prosthetic Upper extremity Functional Index (PUFI) questionnaire and

explored relations with hand function. In this chapter, we demonstrated that 96% of all activities could be performed independently and therefore concluded that children with a CHD generally perform their bimanual activities well. We also found that, although hand function of the affected hand is compromised and numerous alternative strategies can be used to perform bimanual tasks, they are mostly performed with active use of the affected hand. Main predictors of bimanual performance were the manual capacity of the non-dominant hand together with muscle strength (e.g. opposition strength of the non-dominant hand and lateral pinch strength) of the dominant hand**.** Therefore, we suggest that surgical interventions at function level with the ultimate goal to improve bimanual performance should specifically aim at enhancing manual capacity and muscle strength.

Additionally, we determined that spread on the PUFI scores is mainly explained by only 6 of the 38 items. Consequently, we propose that the number of items of the presently very extensive PUFI questionnaire potentially could be reduced when evaluating children with CHD.

Chapter 6 outlines the ICF-CY domains of manual capacity to perform daily activities, hand functions (e.g., strength and mobility) and their interrelations. In this chapter, we present the results on manual capacity and hand function of both hands. Since we found that hand dominance strongly influenced manual capacity, we reported outcome measures for both hands separately. We found that manual capacity of the dominant of the CHD children was only mildly limited, while the non-dominant hand score was more strongly limited. Even when the dominant hand and the non-dominant hand were similarly impaired at hand function level, dominant hands scored higher on manual capacity than non-dominant hands. Although we found small effects of separate hand function measures on manual capacity, we did find that the relation between these outcome measures was stronger in non-dominant hands than dominant hands. We speculated that in bilaterally affected children, interventions, both surgical and conservative, should primarily focus at the non-dominant hand since improving functions such as strength and mobility of this hand may lead to a larger gain in this hand's manual capacity in comparison with the effects of the intervention at dominant hand. In addition, we found that severity of the CHD (bilateral involvement, number of affected digits) and hand function had only small effects on manual capacity.

Finally, **Chapter 7** describes the main findings of this thesis and discusses the methodological considerations, both strengths and limitations. In this chapter we also addressed clinical implications and made recommendations for future research.