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APLES – The Achondroplasia Personal Life Experience Scale –

Development of a Questionnaire to Assess Quality of Life, Burden of Disease and Functionality of Children with Achondroplasia

Master Thesis

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Abstract

Background: Achondroplasia is the most common form of disproportionate short stature. It is a result of an autosomal dominant mutation in the fibroblast growth factor receptor 3 gene (FGFR3), which causes an abnormality of cartilage and bone formation. Little is known about the health-related quality of life (HrQoL) of young patients with Achondroplasia and only a few HrQoL instruments exist for this patient group. To better understand the consequences and impact of this condition on the life of the affected children and adolescents, the objective of this study was to develop a condition-specific patient-reported outcome measure to assess HrQoL, wellbeing and functioning, considering the patients' and their parents' perspective.

Method: In a first step, focus group data of a previous study with individuals with Achondroplasia was qualitatively analyzed. To identify relevant concepts associated with HrQoL, wellbeing and functioning, statements in the focus group discussions were coded regarding the International Classification of Functioning – Children and Youth (ICF-CY) and used for item generation. In a second step pilot testing and new focus group discussions with a cognitive debriefing were conducted with children and adolescents with Achondroplasia and their parents. Qualitative and quantitative data was used to finally select items for the subsequent field test.

Results: In total, 59 items were generated based on codes of the ICF-CY and included in a pilot test with a cognitive debriefing. Following the results of the pilot test, the field test version includes seven scales with 35 Likert-scaled items assigned to the main ICF-CY components: Body functions (global and specific mental functions), body structures (structures related to movement), activities and participation (community, social and civic life/ interpersonal interactions & relationships/ mobility/ self-care/ domestic life) and environmental factors (attitudes/ support & relationships). The questionnaire is available in self- and parent report in the age groups 8-11 and 12-14 years and for parent-report in the age group 5-7 years.

Conclusion: The APLES questionnaire is based on the international language of the ICF-CY and addresses in sufficient detail the special situation, specific burdens, restrictions and resources of children and adolescents with Achondroplasia, especially with regard to body structures. Applying the APLES questionnaire in research or in medical practice can help to better understand HrQoL, wellbeing and functioning in this patient group. Subsequently, a field and retest is needed to psychometrically test the new instrument.

Keywords: HrQoL, Achondroplasia, ICF-CY, patient-reported outcomes

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List of Abbreviations

ACH	Achondroplasia
APLES	The Achondroplasia Personal Life Experience Scale
BKMF	Bundesverband Kleinwüchsige Menschen und Ihre Familien e.V. (German Federal Association of Short Statured People and their Families)
Ch/Ad/yA	Children/Adolescents/young adults
GHD	Growth Hormone Deficiency
HrQoL	Health Related Quality of Life
FG	Focus Groups
ICF	International Classification of Functioning Disability and Health
ICF-CY	International Classification of Functioning Disability and Health, Children and Youth Version
ISS	Idiopathic Short Stature
PRO	Patient-Reported Outcomes
QoL	Quality of Life
SD	Standard deviation

1. Introduction

Skeletal dysplasia is a the rare diseases with an approximate prevalence of 1:3000 to 1:5000 (Niethard, 2009). Of the about 350 existing forms of skeletal dysplasia, Achondroplasia is the most common nonlethal form (Krakow & Rimoin, 2010). This disease is characterized by disproportionate short stature with short arms and legs, resulting from an autosomal-dominant mutation in the fibroblast growth factor receptor 3 gene (FGFR3), which causes an abnormality of cartilage and bone formation (Baujat & Legeai-Mallet *et al.*, 2008).

In addition to the significant longitudinal growth reduction, individuals with Achondroplasia experience various orthopedic and neurological complications like a narrowing of the foramen magnum and/or the spinal canal, which can cause severe complications. Moreover, short statured people often face challenges in daily life and may experience stigmatization or social exclusion (Jiang & Rasmussen *et al.*, 1999). Treatment options are very limited, since only a few specialists or specialized clinics are available due to the rarity of the disease (European Organization for Rare Diseases, 2005).

Furthermore, little is known about the impact of Achondroplasia on the life of the patients and on the quality of life of patients living with this condition (Gollust & Thompson *et al.*, 2003). The impact of chronic diseases on wellbeing and functioning of the patient is described by the concept of health-related quality of life (HrQoL). It reflects the subjective perception of health and contains components of physical, emotional, mental and social areas of life (Bullinger, 2002). Assessment of HrQoL can help to evaluate treatment effects or to increase the understanding of the burden of a disease on wellbeing from the patients' perspective. Especially in pediatrics this health indicator is relatively new (Matza & Patrick *et al.*, 2013). Therefore instruments are increasingly developed to assess HrQoL in various pediatric conditions. Apart from generic instruments to assess HrQoL in children and adolescents, condition specific instruments are needed to better understand the consequences and burden of a specific disorder, such as Achondroplasia (Brütt & Sandberg *et al.*, 2009).

To assess HrQoL data of individuals, patient-reported outcome instruments are used. These instruments specifically assess wellbeing and functioning of individuals with regard to their condition from their perspective, without interpretation of someone else. Patient-reported outcome instruments are essential for clinical practice, health-care policy and research (Patrick & Burke *et al.*, 2007).

So far only few studies focus on quality of life of short statured children and adolescents and only a small number of instruments is available to measure HrQoL in this patient group, focusing mainly on idiopathic short stature (ISS) or growth hormone deficiency (GHD) (Bullinger & Koltowska-Haggstrom *et al.*, 2009). However, the existing instruments to measure quality of life in children and adolescents with short stature do not capture specific disease and functioning burdens, such as disproportionality, that are associated with Achondroplasia. According to Rohenkohl & Sommer *et al.* (2015) children and adolescents with Achondroplasia are especially impaired in physical and social life and report a lower HrQoL in these domains, in comparison to short statured people with a normal proportioned body. Quitmann & Witt *et al.* (2014) showed, that a variety of social, physical and emotional problems have an impact on the HrQoL of young people with Achondroplasia. Moreover, affected persons report disease specific limitation and the need for assistance in daily life.

The socio-emotional and medical consequences of Achondroplasia are broad. Especially the disproportionate body of young patients with Achondroplasia seems to have an impact on their body functions, to cause physical problems as well as restrictions and experiences of stigmatization in daily life. However, regarding Achondroplasia no disease specific quality of life questionnaire for children and adolescents with Achondroplasia exists, that covers not only quality of life, but also further aspects of health, body functionality and disability that are relevant issues for the disease Achondroplasia. To identify these concepts in patients with Achondroplasia, a condition specific patient-reported outcome instrument is needed.

Therefore, the aim of the study is to develop a condition specific patient-reported outcome instrument that captures information about disease, functioning, psychosocial wellbeing and quality of life of young patients with Achondroplasia, considering the patients' and their parents' perspective (The Achondroplasia Personal Life Experience Scale (APLES)).

In the development process of a patient-reported outcome instrument the use of focus groups is essential to understand patients' needs and concerns of their health, because many aspects of health conditions are only known by the patients themselves (Schmidt & Thyen *et al.*, 2008). In this study, previous focus group data was used to generate items for the pilot version of the APLES questionnaire. Therefore, the first empirical part of this study consists of an in-depth content analysis of focus group data of a previous study with children, adolescents and young adults with Achondroplasia as well as their parents, regarding the children and youth version of the International Classification of Functioning, Disability and Health (ICF-CY). Subsequently the pilot version was tested in a pilot test within new focus group discussions with children and adolescents with Achondroplasia and their parents. Hence, the second empirical part of the study consists of a quantitative analysis of the pilot test and a qualitative analysis of the new focus group discussions.

By these two in depth qualitative content analysis from both focus groups discussions and the results of the pilot test, a better understanding of the impact of Achondroplasia on the patients' HrQoL, physical functioning and wellbeing is expected. The new instrument would clarify special limitations and domains of interests that are associated with the disease Achondroplasia. Moreover, as a patient-reported outcome instrument it would identify needs directly from the patients' perspective and help to optimize health care in this patient group within the framework of the ICF-CY.

2. Theoretical Background

The following paragraphs provide detailed information about short stature and particularly about Achondroplasia. Furthermore, the concept of health related quality of life, patient-reported outcome instruments and the International Classification of Functioning, Disability and Health are described.

2.1 Short Stature

Statistically about 3% of the children in a population are of small stature (Hoepffner & Pfäffle *et al.*, 2011). Short stature is defined as a body height of 2.0 or more standard deviations (*SD*) below the population mean with regard to age and gender, (Wit & Clayton *et al.*, 2008) or if the body height falls below the third percentile (Koletzko, 2013). It is a symptom of a broad variability of pathologic conditions and disorders. Especially in childhood growth is an indicator for the overall health of a child and abnormal growth can be a signal for a present disease (Grimberg & Kutikov *et al.*, 2005). Besides genetic factors, ethnic origin and environmental factors like socioeconomic conditions influence height as well. Therefore, the growth reference data of the populations norm should originate from a country's population itself (Wit *et al.*, 2008).

Growth disorders can be divided into primary and secondary causes. Primary growth disorders are caused by dysfunctions in the skeletal system, affecting the body growth, whereas the bone maturation is usually normal. The most common form of primary growth disorders is familial short stature, which is genetically determined. In this case the body height is abnormal compared with the average population, but normal within the family (Ranke, 2007). Besides familial short stature, skeletal dysplasia is also classified in primary growth disorders. It is characterized by a disproportionate short stature, with abnormal body proportions between the trunk and extremities. Examples are Achondroplasia, Hypochondroplasia and Osteogenesis Imperfect. Furthermore, chromosomal defects (e.g. Ulrich-Turner-Syndrome, Trisomy 21), intrauterine growth radiation, malfunctions in the bone metabolism and syndromological growth disorders rank among primary growth disorders (Koletzko, 2013; Ranke, 2007).

In comparison with primary growth disorders, secondary growth disorders are always associated with a retardation of the maturation and dysfunctions of the endocrine system, which is among other things responsible for the body growth. Hence, the growth process is delayed and the beginning of puberty retarded. However, the possibility to reach a normal height in adulthood is given, if treatment starts early in childhood and the reasons for the growth disorder are known. These can be very broad, since the actual reason for secondary growth disorders is not associated with the skeletal system. Common reasons for secondary growth disorders are constitutional developmental delay, nutritive disorders (e.g. malnutrition), several organic diseases and metabolic or hormonal disorders (Koletzko, 2013).

Within endocrine causes for short stature, growth hormone deficiency (GHD) is the most common one. This disease is caused by a complete or partial absence of growth hormone secretion in the hypophysis (Binder & Woelfe, 2010). However, among all causes for short stature, GHD represents a relatively rare cause for short stature. According to laboratory tests, most short statured children are growth hormone sufficient (Lindsay & Feldkamp *et al.*, 1994).

A remaining group where no medical reason for short stature can be diagnosed is classified as idiopathic short stature (ISS). In this heterogeneous group of short statured patients no laboratory abnormalities, no evidence of a systemic disease, no hypothyroidism, no malnutrition or GHD can be found (Wit *et al.*, 2008).

2.1.1 Psychosocial Aspects of Short Stature

Short statured people face challenges in daily life and their environment caused by height related physical limitations. In research it is often discussed, that short stature correlates with social stigmatization and social isolation which can cause behavioral and emotional problems and affect self-perception and social integration (Bullinger & Quitmann *et al.*, 2013; Dogba & Rauch *et al.*, 2014; Gollust *et al.*, 2003; Voss & Mulligan, 2000). Consequently, short statured people are at a higher risk for psychosocial stress and show tendencies towards psychosocial problems like depression and a low self-esteem (Abe & Okumura *et al.*, 2009). However, this is always associated with the ability and effectiveness of coping with psychosocial stresses and experiences in daily life related to short stature. Especially in childhood, psychosocial effects of short stature are often related to the child’s experiences and barriers in everyday life as well as to a reduced autonomy. Moreover, self-perception and social integration are affected by negative experiences with peers, being bullied in school, low self-confidence or stigmatization related to the short stature (Bullinger *et al.*, 2009; Sandberg & Voss, 2002; Voss & Mulligan, 2000).

Although short statured children have cognitive functions within the typical range, results of a review by Wheeler & Bresnahan *et al.* (2004) revealed that short stature in children is often related to a decrease in intelligence and academic achievement as well as to a significant reduction and delay in visual motor skills. However, it is not clear if a low performance in education and intelligence directly refer to short stature or if the consequences of short stature, namely psychosocial impairments, are the reason for it (Wheeler *et al.*, 2004). When considering results of other studies, no association between functional impairment and short stature has been found, (Chaplin & Dahlgren *et al.*, 2006; Kranzler & Rosenbloom *et al.*, 2000) and the physical appearance of a short statured body not generally predicts the children’s psychological adaption or psychological status (Bullinger, 2011; Sandberg & Voss, 2002). Hence it is uncertain why some short statured children have no psychological impairments and a normal psychological development, while others do not (Erling, 2004). Besides, no association between short stature and a weaker psychosocial functioning has been found in young adults with short stature. Height did not affect areas like education and employment or interpersonal relationships in this patient group (Ulph & Betts *et al.*, 2004).

2.2 Achondroplasia

Skeletal dysplasia is classified as a primary growth disorder. These form a heterogeneous group of genetically caused diseases based on disorders in the bone development. Of about 350 existing forms of skeletal dysplasia, Achondroplasia is the most common form of genetically determined short stature (Krakow & Rimoin, 2010; Warman & Cormier-Daire *et al.*, 2011). With an incidence of 1:20.000 live births it is a rare disease, affecting 250.000 people worldwide. In Germany about 40 to 45 children are born with Achondroplasia each year (Mohnike & Klingebiel *et al.*, 2013).

In more than 95% of the patients the disease is caused by a new mutation in the genetic disposition of the fibroblast growth factor receptor 3 gene (FGFR3). The mutation is characterized as autosomal-dominant. Hence, the patient has a 50% risk to transmit the genetic defect to the next generation. A mutation in the FGFR3 gene can also lead to nonviable forms and Hypochondroplasia, however, Achondroplasia is the most common nonlethal disease within mutations of the FGFR3 gene (Horton & Hall *et al.*, 2007; Mohnike *et al.*, 2013).

The mutation affects the growing skeleton and causes an abnormality of cartilage and bone formation, resulting in a phenotype which is characterized by a disproportionate short stature (Horton *et al.*, 2007). Due to the dysfunction in the cartilaginous growth plate the long bones of the patients stay short and induce the disproportionate body with shorter arms and legs, while the trunk has a relatively normal size (Niethard, 2009).

Besides the disproportionate short stature, the phenotype of Achondroplasia is associated with an enlarged head, a depressed nasal bridge and a prominent forehead. Affected persons show a relatively long torso with an extreme lumbar hyperlordosis and a prominent buttock. Further characteristics are short hands and fingers, with an often enlarged space between the third and fourth finger, which is designated as a ‘trident hand’. An enhanced growth of the fibula in contrast to the tibia often results in a genu varum. The final adulthood height for men with Achondroplasia is 125 cm and for women 120 cm (Baujat *et al.*, 2008; Krakow & Rimoin, 2010; Richette & Bardin *et al.*, 2008).

Medical consequences of Achondroplasia are broad, but are mostly consequences of the abnormal bone growth. Affected persons often suffer from joint pain and joint limitations due to skeletal abnormalities like a genu varum or the excessive lumbar hyperlordosis. A serious complication in childhood is a cervical cord compression because of a stenosis of the foramen magnum. This complication manifests itself in neurological dysfunctions like motor delay, sleep apnea, hydrocephalus, disorders of respiration, vomiting or in the worst case it can lead to a sudden infant death (Baujat *et al.*, 2008; Richette *et al.*, 2008). In adulthood the narrowed spinal canal in combination with the lumbar hyperlordosis can cause paresthesia in the lower extremities (Krakow & Rimoin, 2010). Another common complication in children with Achondroplasia is caused by the too short Eustachian Tubes in the middle ear. This complication often results in ear infections, otitis media or deafness. Furthermore, hypotonia can cause a thoracolumbar kyphosis. In more than 10% of the patients it results in a fixed thoracolumbar kyphosis deformity (Richette *et al.*, 2008). Moreover, Achondroplasia is often related to obesity. Hence the morbidity increase, particularly joint problems (Baujat *et al.*, 2008). Figure 1 summarizes the described medical problems and their consequences in children and adults with Achondroplasia.

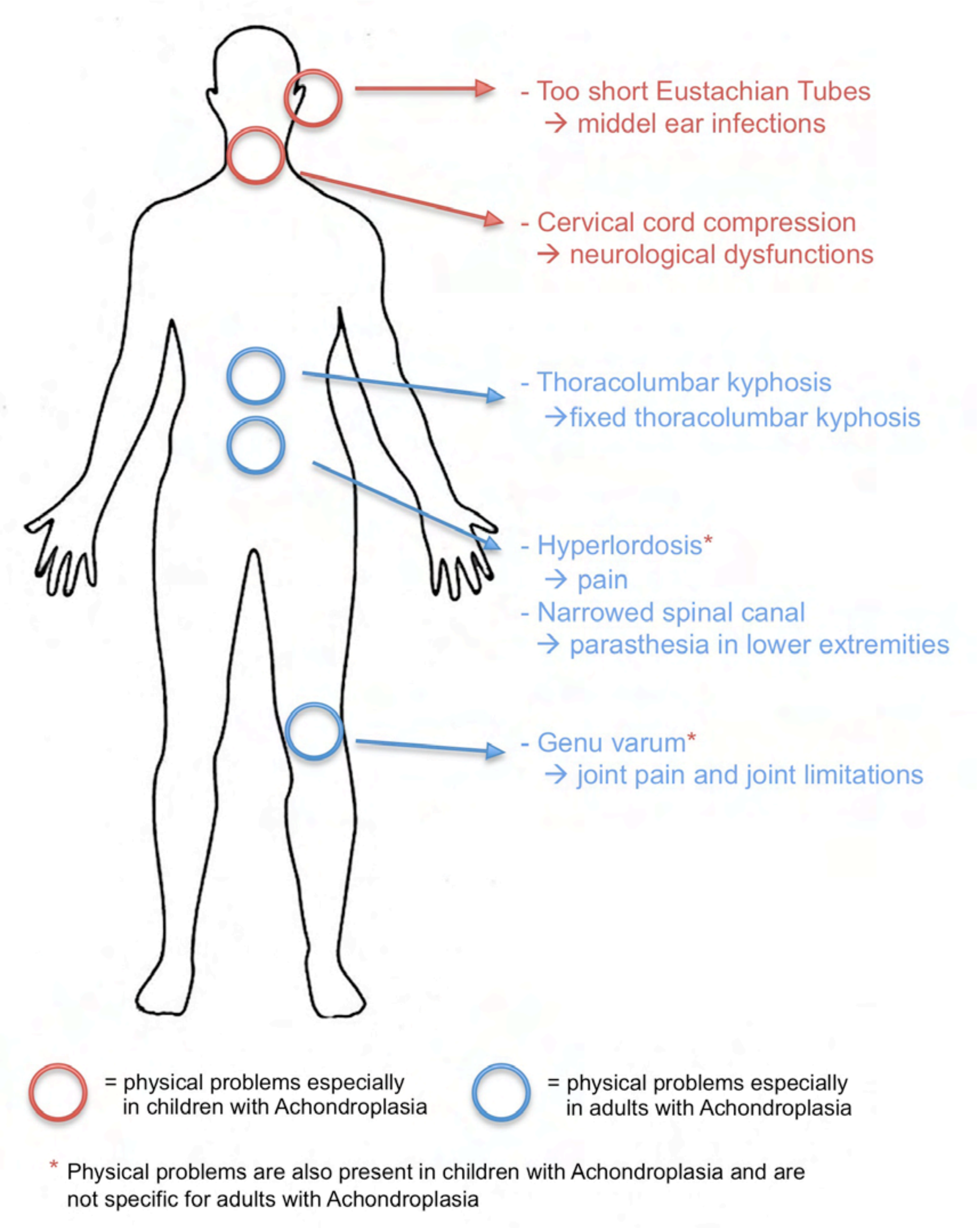

Fig. 1 Physical medical problems of adults (in blue) and children (in red) with Achondroplasia and the related consequences

Due to the rareness of the disease, it is very difficult for the families and the patients to find adequate medical treatment or specialists. To assist primary care physicians, specific guidelines were developed that should help to control and support the development of the child. These guidelines include explicit growth curves for Achondroplasia for height, head and chest circumference. Additionally, specific testing for known complications at different ages is relevant to provide the best care for the patients (Horton *et al.*, 2007). However, therapy is still difficult because often only symptomatic therapy can be provided, which consists of orthopedic and physiotherapeutical methods to reduce pain and skeletal dysfunctions. Often a decompression surgery is necessary to reduce the foramen magnum compression or an osteotomy to correct the leg axis in case of a genu varum (Richette *et al.*, 2008). The common problem of otitis media is treated with ventilation tubes to prevent deafness (Horton *et al.*, 2007).

Therapies to increase short stature are often discussed, since growth hormone therapy is not recommended for Achondroplasia. One possibility is the surgical limb lengthening by breaking bones in the lower extremity, and to stretch them with the help of an external fixator during the process of healing. However, this technique is very painful for the patient and has a lot of disadvantages like repeated surgeries, wound infections, reduced quality of life and other complications that come along with a surgery. With this treatment, the final height can be increased by about 15 to 30 cm. Nevertheless, the aspect of disproportionality is not solved with this surgery because the upper extremity is still too short (Baujat *et al.*, 2008; Horton *et al.*, 2007).

New therapeutic approaches refer directly to the signals and effects of the FGFR3 gene. Promising study results demonstrate an effect of a natural human peptide (C-type natriuretic peptide), which is a positive regulator of bone growth and inhibits the overactive FGFR3 pathway. Studies with mouse models showed a positive effect on longitudinal growth as well as the ability of this peptide to correct the skull and narrowed spinal canal (Mohnike *et al.*, 2013).

2.2.1 Psychosocial Aspects of Achondroplasia

Most individuals with Achondroplasia have cognitive functions within the normal range. Unless no serious neurological consequence is present, affected persons can live a normal and independent life within the normal life span (Baujat *et al.*, 2008; Richette *et al.*, 2008). However, within the first seven years of life, children with Achondroplasia show delayed motor milestones compared to population norms. Due to musculoskeletal impairments that are associated with the disease, they have a greater need for assistance in daily life, mainly in self-care and mobility including access to toilets, cabinets or wash bins. The strongly reduced longitudinal growth complicates the everyday life of the children. Especially their too short upper limbs restrict children under the age of seven years to perform personal hygiene in the intergluteal region during bathing or toileting or to reach their head to brush their hair. By the age of seven, children are mostly able to perform personal hygiene themselves with assistive devices like stools (Ireland & Mcgill *et* *al.*, 2011). Consequently, the children experience a reduced autonomy in their lives, particularly in early childhood.

Furthermore, social stigmatization and an increased experience of bullying or teasing induce a higher risk for psychosocial stresses in children with Achondroplasia. Previous studies document a lower self-esteem in individuals with Achondroplasia and an increase in depression in adults (Gollust *et al.*, 2003; Hunter, 1998). However, psychosocial problems are always related to coping resources like self-efficacy and do not necessarily have to occur in this patient group. In a sample of a study by Nishimura and Hanaki (2014) psychosocial maladjustment cannot be proven, although children with Achondroplasia experience increased psychological and physiological stressors related to short stature. The results emphasize that height is not necessarily an indicator for psychosocial impairments and highlight the importance of assisting children in developing coping strategies from an early age on. However, not only the patients face disease related challenges, often the whole family experience various psychosocial challenges in their lives and an increased time exposure to assist the child (Quitmann *et al.*, 2014).

Next to the variety of medical and psychosocial aspects, the topic quality of life plays an important role as well in rare diseases, such as Achondroplasia. However, so far health related quality of life in children and adolescents with skeletal dysplasia in general or Achondroplasia in particular has been rarely investigated (Thompson & Shakespear *et al.*, 2008).

2.3 The Concept of Health Related Quality of Life

Quality of life (QoL) is a broad construct and describes the overall wellbeing of a person including physical, psychosocial, emotional and family dimensions. Health is an important element and condition of QoL and the absence of health can have a negative impact on someone’s quality of life. However, the general construct QoL is not only affected by the health status, other valued aspects like political freedom, economical concerns and a safe environment influence QoL as well (Radoschweski, 2000).

Over the last years, QoL has become a relevant endpoint in medicine and health science. In this setting, QoL is associated with the patients’ subjective perception of their health condition, which is described as health related quality of life (HrQoL). Nowadays HrQoL has become an important outcome indicator in medicine, because not only objective medical aspects like a change in symptoms or life-time extension are relevant to evaluate the health status or medical treatments, also the patients’ own view on their health condition is taken into account (Bullinger, 2002; Cohen & Biesecker, 2010; The European

QoLISSY Group, 2013). Hence, HrQoL can be seen as an important element of the broader construct QoL (Bullinger, 2002; Bullinger *et al.*, 2013).

According to the World Health Organization (WHO), HrQoL is defined as “an individual’s perception of his/her 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. It is a broad-ranging concept affected in a complex way by the person’s physical health, psychological state, level of independence, social relationships, personal beliefs and their relationship to salient features of their environment” (WHO, 1997, p. 1).

This definition emphasizes that HrQoL is an indicator for the subjective perception of health in terms of wellbeing and functionality from the patients’ point of view. As a multidimensional construct it covers physical, social, emotional, mental and behavioral aspects of wellbeing from the patients’ as well as from an observers’ perspective (Bullinger, 2002; Bullinger & Schmidt *et al.*, 2007).

2.3.1 Assessment of Health Related Quality of Life

The development and psychometric testing of measurements to assess HrQoL is essential in order to understand the effects of a health condition from the patient’s perspective. This research started in the 1980s and has strongly increased over the past years. However, it was not before the year 1990 that measures were included in studies to assess HrQoL (The European QoLISSY Group, 2013). Nowadays HrQoL is increasingly assessed in clinical trials to evaluate the subjective health of a patient and to evaluate treatment effects.

According to Brütt *et al.* (2009) HrQoL can be assessed in four different levels as shown in figure 2. Generic instruments are applicable independently of the context of the health condition or treatment situation. They provide the opportunity to compare the health status across populations or between different diseases. However, generic scales are often not sensitive enough to detect small changes in HrQoL (Cohen & Biesecker, 2010). A common example for a generic tool is the 36-item Short Form (SF-36) of the Medical Outcome Study which surveys the HrQoL of a patient in eight different dimensions (McHorney & Ware *et al.*, 1993). The second level includes chronic generic measures. These measures focus on the experience of having a chronic health condition, independent of which. Hence they are not detailed enough to assess specific characteristics of a condition. On the third level, condition-specific instruments are adapted to the disease and the associated problems. They have the strength to identify the impact of a specific disease on HrQoL. However, a comparison between different illnesses is not possible when using a disease specific instrument (Cohen & Biesecker, 2010). The last level of HrQoL assessment is the treatment-specific level. In this level the assessment directly refers to the patients' perspective regarding the type of care received.

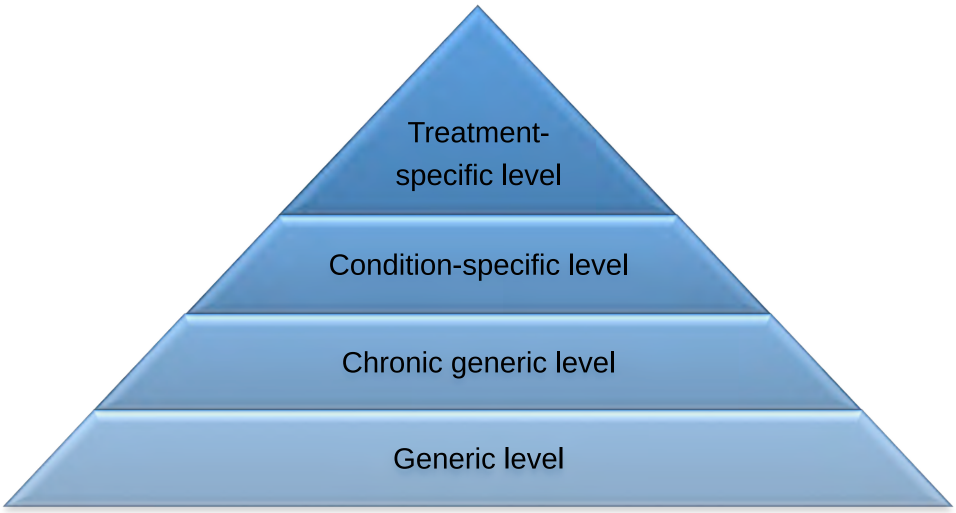

Fig. 2 Levels of HrQoL assessment (Brütt et al., 2009, p. 66)

2.3.2 Health Related Quality of Life Research in Children and Adolescents

While HrQoL research is common in adults and progressed steadily over the last years, this field of research is still relatively new in pediatrics and is still in the developmental stage (Bullinger *et al.*, 2007). However, HrQoL research is just as important in pediatrics as it is in adult health care. Using measures to assess HrQoL in pediatric healthcare would support patient-physician communication and improve the satisfaction of the child and parent. The instruments can serve as a screening tool and would help to identify unknown morbidities and physical or psychosocial health problems from the patient and parent perspective. Furthermore, it would assist clinical decision making and provide information about the patient's perspective on health and treatment, which is essential for a successful therapy (Varani & Burwinkle *et al.*, 2005). Hence, the assessment of HrQoL can be used to evaluate the success of a therapy and to optimize health care. Especially in chronic diseases and mental health problems, information about the subjective perception of health is essential to counteract restrictions in wellbeing and functionality prematurely (Ravens-Sieberer & Klasen *et al.*, 2013). However, assessing HrQoL in young populations is very challenging because several aspects like age, reporting method and the cognitive ability and emotional awareness of the child need to be taken into account (Ravens-Sieberer & Ellert *et al.*, 2007).

A HrQoL instrument for use in pediatric practice needs to be available for different age groups and validated in each group, because the domains which define QoL, vary according to age. Information about HrQoL can be obtained from the children themselves in a self-assessment report or from observing persons, mostly the parents, in a parent-report version (Cohen & Biesecker, 2010; The European QoLISSY Group, 2013). Normally children from the age of eight years and onwards are able to judge their QoL in a self-report. However, if the child's cognitive ability is impaired or if the child is too young, a parent can serve as a proxy (Riley, 2004).

In 1994, the World Health Organization, Division of Mental Health, released guidelines for measurements of quality of life in children. According to them an instrument that assesses HrQoL in children should contain the following characteristics (WHO - Division of Mental Health, 1994):

- child centred
- employ subjective self-report wherever possible
- age related or developmentally appropriate
- cross culturally comparable
- generic core and specific modules
- put an emphasis on health enhancing aspects of quality of life rather than merely negative aspects

Besides these characteristics, special aspects in the life of children and adolescents like school, sports, family and friends need to be considered in a pediatric HrQoL instrument (Ravens-Sieberer *et al.*, 2013).

Within HrQoL research in young populations, mostly generic measures have been developed; only recently disease-specific measures have been published. Thus such measures are lacking for many young and small populations, among them Achondroplasia. Furthermore, only a few instruments can be used in an international study context, because often the instruments are not developed under a cross-cultural approach and do not take cultural differences into account (The European QoLISSY Group, 2013).

2.3.3 Health Related Quality of Life Research in Achondroplasia

HrQoL research in short statured people focus especially on Growth Hormone Deficiency (GHD) or Idiopathic Short Stature (ISS) (Bullinger *et al.*, 2009). Since only a few methodological inadequate instruments exist to measure HrQoL in short statured people, the aim of the European QoLISSY (Quality of Life in Short Stature Youth) project was to develop and psychometrically test an instrument that evaluates the impact of short stature on HrQoL in children and adolescents who are diagnosed with ISS or GHD and to assess the effect of interventions from both the patient and parent perspectives in different European countries. The QoLISSY questionnaire consists of 22 items which are assigned to the core HrQoL dimensions: Physical, social and emotional as well as 28 additional items which are assigned to coping, beliefs and treatment (Bullinger *et al.*, 2013; Quitmann & Rohenkohl *et al.*, 2013; The European QoLISSY Group, 2013).

Compared to HrQoL research in children and adolescents who are diagnosed with endocrine short stature or ISS, limited research is available that assesses the impact of Achondroplasia on the life of the patient. Clinical characteristics like the significant reduction in height and the disproportionate body are likely to have a negative impact on the QoL of people living with Achondroplasia. Especially in childhood and adolescence the body-image highly correlates with HrQoL (Haraldstad & Christophersen *et al.*, 2011). However, the diagnosis Achondroplasia should not be directly associated with a lower HrQoL because a strong predictor that influences HrQoL is the level of individual self-esteem and the severity level of Achondroplasia (Gollust *et al.*, 2003).

So far, only little research has been done to assess HrQoL of individuals with Achondroplasia. The few available studies in this field focus especially on HrQoL assessments of adults with Achondroplasia using different QoL instruments. In a study by Mahomed & Spellmann *et al.* (1998), the generic quality of life instrument SF-36 was used to assess HrQoL in this patient group (McHorney *et al.*, 1993). Results showed a reduction in HrQoL of adults with Achondroplasia with increasing age, compared to population norms. Another study by Apajasalo & Sintonen *et al.* (1998) used the generic HrQoL instrument 15D and 16D (Sintonen, 2001). The results indicate as well, that HrQoL was lower in adults and adolescents with Achondroplasia, compared to the normal sized control group. Gollust *et al.* (2003) came up with the same results, using the generic Ferrans and Powers Quality of Life Index, which assesses quality of life in four different dimensions, namely health/functioning, psychological/spiritual, socioeconomic and family, based on the level of satisfaction (Kimura & Vitor da Silva, 2009). Moreover, they showed that the level of self-esteem and social challenges are a strong predictor for HrQoL in individuals with Achondroplasia (Gollust *et al.*, 2003).

HrQoL of children and adolescents with Achondroplasia has even more rarely been studied (Thompson *et al.*, 2008). The main reason for this is a lack of disease-specific instruments, to assess HrQoL of children and adolescents with Achondroplasia adequately. Therefore the QoLISSY questionnaire, which is already validated in several European countries for children and adolescents with short stature diagnosed with ISS and GHD, was further examined in conditions of skeletal growth disorders. The study by Rohenkohl & Bullinger *et al.* (2014) aimed to test the psychometric performance of the German QoLISSY questionnaire in patients with Achondroplasia. An additional goal of this study was to understand the HrQoL of children, adolescents and young adults with Achondroplasia, using generic and disease-specific instruments. The sample included 89 children, adolescents and young adults (8-28 years) and 63 parents of participating children (8-17 years). Quantitative measures used in the study to assess HrQoL included the generic KIDSCREEN questionnaire (The Kidscreen Group, 2006), the chronic-generic DISABKIDS questionnaire (The Disabkids Group Europe, 2006) and the disease specific QoLISSY questionnaire (The European QoLISSY Group, 2013). Available representative population data of the KIDSCREEN questionnaire was used for a comparison between patient and population norm. The results document that children and adolescents with Achondroplasia evaluate their physical HrQoL poorly and the emotional HrQoL more positive, compared to population norms. The results show as well, that parents of children with Achondroplasia evaluate the HrQoL of their children lower than population norms. Furthermore, the results prove a satisfactory psychometric performance of the instrument which suggests that the QoLISSY questionnaire is a reliable instrument that can be used to assess the subjective wellbeing of patients with skeletal dysplasia. It appears to capture concerns of the patient population in terms of symptoms, treatment effects and specific challenges due to the condition and can be used as a screening instrument to assess the subjective wellbeing of the patients in clinical practice (Rohenkohl *et al.*, 2014).

However, it might be problematic to use HrQoL instruments which are originally designed for patients with proportionate short stature for this patient group, because people with Achondroplasia experience other psychosocial and physical difficulties due to their disproportionate body, than individuals with proportionate short stature (Gollust *et al.*, 2003).

Results of a recent study indicate that especially the disproportion of the body of young patients has an impact on their body functions, physical problems as well as experiences and restrictions in daily life. Children and adolescents with Achondroplasia are especially impaired in physical and social life and report a lower quality of life in these domains in comparison to short statured people with a normally proportioned body (e.g. diagnosed with ISS or GHD) (Rohenkohl *et al.*, 2015). According to results of a study by Quitmann *et al.* (2014) especially adolescents (13-18 years) and young adults (19-28 years) show coping strategies with regard to the disease and rate their HrQoL higher than children (8-12 years) with Achondroplasia. Still, the affected persons report disease specific limitations in social, physical and emotional life domains and the need for assistance in daily life, which has a negative impact on their HrQoL.

Hence, although the QoLISSY questionnaire is a validated tool to assess HrQoL of young patients with Achondroplasia, it does not cover the unique aspect of disproportionality and the related experiences, burdens and restrictions in the daily life of this patient group. Up to now, no existing questionnaire measures these particular topics and no condition-specific questionnaire exists that covers not only HrQoL but also further aspects of health, body functionality, disability and disproportionality that are relevant issues for the disease Achondroplasia. A condition-specific patient-reported outcome instrument for children and adolescents that assesses these aspects is needed in order to fill the research gap and to optimize health care in this patient group. In the development process of such an instrument it is obligatory to respect guidelines that have been developed for patient-reported outcome (PRO) instruments. These guidelines are presented in the following paragraph.

2.4 Guidelines for the Development of Patient-Reported Outcome Instruments

A patient-reported outcome (PRO) is a report about the health condition which comes directly from the patients themselves. Hence, patient-reported outcome instruments are used to collect data to assess wellbeing and functioning of individuals with regard to their disease, condition or treatment from their perspective, without interpretation of someone else (Patrick *et al.*, 2007). In medical practice and clinical trials such measures are increasingly considered and often included to evaluate the effects of a disease or treatment on the patient‘s health condition from their perspective, since many effects of a disease or treatment are only known by the patients themselves. This gives the patients a voice in health care and directly includes their perspectives in medical practice regarding health care decision-making. In general, a PRO instrument can be used to assess simple symptoms like pain intensity, but also complex aspects like HrQoL are often assessed with such an instrument (Frost & Reeve *et al.*, 2007; Rothman & Beltran *et al.*, 2007).

The increased use of PRO in medical practice requires valid and reliable PRO instruments (Frost *et al.*, 2007). Therefore the U.S. Food and Drug Administration (FDA) released guidelines for the development and evaluation of PRO instruments in 2006 (FDA, 2006). A revised version was published in 2009 (FDA, 2009). Central in this approach is the 'bottom-up' (patient-derived) nature of questionnaire construction. With this approach it is essential to include the patients' perspective in the development and testing of the instrument because a PRO instrument should reflect issues that are relevant to the target population and condition, and capture their experiences and burden of disease. Figure 3 describes the development process of a PRO instrument regarding the recommendations of the FDA.

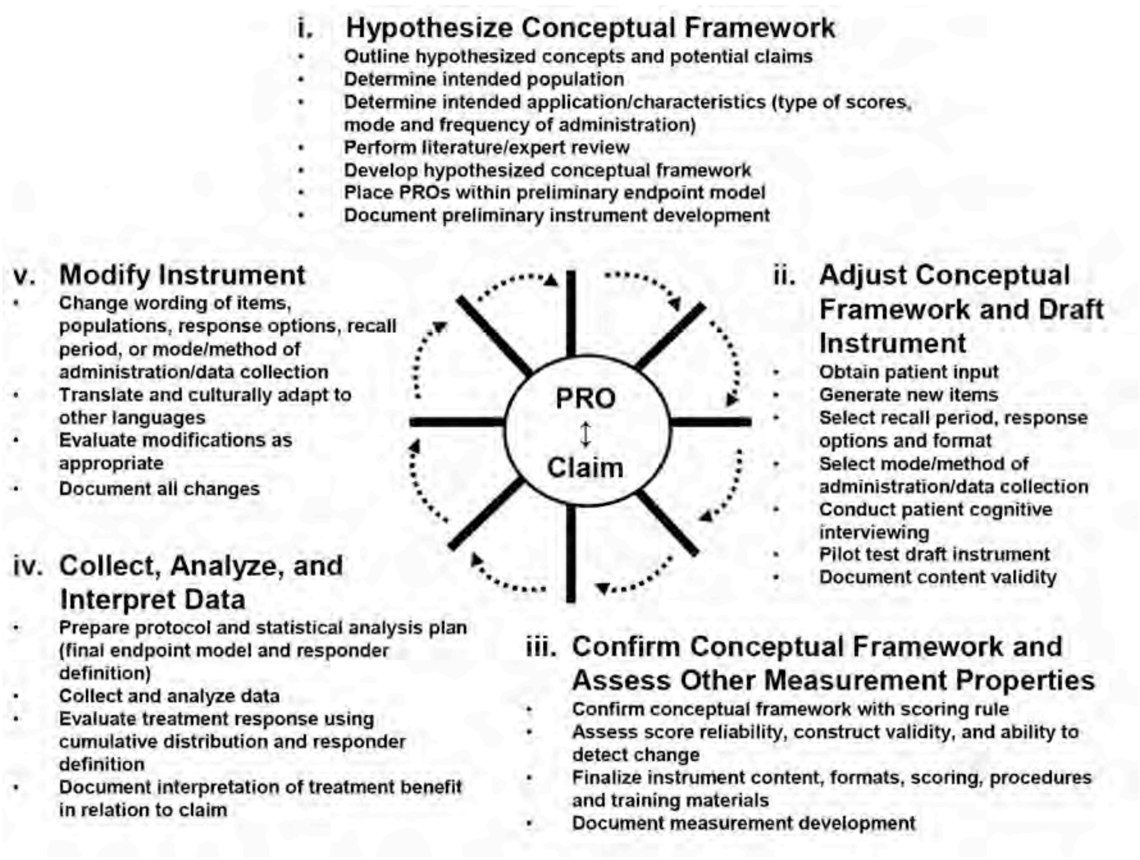

Fig. 3 Development of a PRO instrument (FDA, 2009, p. 7)

To ensure that a PRO instrument is a useful tool to assess PRO data, important psychometric characteristics need to be respected. These include content and construct validity, reliability and ability to detect change (FDA, 2009; Frost *et al.*, 2007). Content validity is defined as the extent to which the instrument measures the concerns and interests specific of the target population. Construct validity proves if the instrument really measures the characteristics it should and if it captures relevant issues in the target population. Reliability refers to test-retest reliability to ensure reproducibility as well as to internal consistence reliability tests, to test to what extent the items are related to each other and measure the same dimension (e.g. Cronbach´s alpha). Ability to detect change demonstrates if the instrument is sensitive enough to measure changes with effective treatment. It is usually assessed by effect size statistics (FDA, 2009; Frost *et al.*, 2007).

Also in pediatrics, it is essential to understand the impact of a disease and treatment effects on the child's health condition and HrQoL from his or her perspective. The use of PRO instruments is necessary to thoroughly understand the health status in this population group and to assess psychosocial outcomes beyond clinical effects (Matza & Swensen *et al.*, 2004). Since the guidelines towards PRO instrument development by the FDA focus primary on adults the task force of the International Society for Patient-Reported Outcomes (ISPOR) released recommendations for PRO instrument development in pediatric populations (Matza *et al.*, 2013). The most important issues that arise in the development of a pediatric PRO tool are, to consider an appropriate age at which children can report PRO data, and whether parents or children should be used for reports.

In children from the age of 8 years and upward child-reports can be assessed. If a child is not able to fill out a PRO instrument because it is too young or cognitively impaired, an informant-report measure can be used. However, the children's and parents' view about the health status can differ. Therefore, child-reported measures should be filled out whenever possible and supplemented by parent-reports because their perspective is important as well, since they take care of the children and make decisions regarding their health care (Matza *et al.*, 2013; The European QoLISSY Group, 2013).

Furthermore, the environment of the children needs to be considered, because the disease of a child can affect the family, peers and school. Therefore, PRO instruments designed to assess HrQoL in children need to respect these child-specific conditions (Matza *et al.*, 2004). Moreover, age-related vocabulary, child appropriate response scales and an adequate length and format of the instrument need to be considered (Matza *et al.*, 2013).

2.4.1 The Use of Focus Groups in the Development Process of a PRO Instrument

To ensure content validity in a PRO instrument, the use of focus groups (FG) is a key element in the developing process. FG discussions are a qualitative research method, aiming to provide the researcher insight in the life of the people. In the discussion direct information of the participants' perspective on their experiences, perceptions, feelings and attitudes on a specific topic can be obtained, especially when little is known about it (Heary & Hennessy, 2002; Jayasekara, 2012). Ideally FG should consist of 6 to 12 participants and the discussion should be directed by a trained moderator (Vaughn & Schumm *et al.*, 1996). In medicine FG can be used to understand patients‘ experiences of disease and treatment and their impact on HrQoL as well as their health care needs (Heary & Hennessy, 2002; Schmidt *et al.*, 2008).

Also in pediatric populations, FG proved to be a useful tool to gather qualitative data on the children‘s view on specific aspects. In general, children from the age of eight years are able to participate in FG discussions. When conducting FG discussions with children, developmental differences should be considered and controlled by conducting the FG in an age span of two years. By doing this, it is unlikely that the differences in age endanger the group dynamic and influence the results of the discussion. Moreover, the composition of the group needs to be considered. Especially when the topic is very sensitive, it is recommended to conduct single-sex groups (Greene & Hogan, 2005; Heary & Hennessy, 2002).

The data of the FG discussions forms the foundation of a PRO instrument and is used to generate items according to the population group (Rothman & Burke *et al.*, 2009). Using FG in the process of item generation, guarantees that the items directly refer to the target population and reflect its members‘ concepts of interests (FDA, 2009). This research method has already been applied successfully in pediatric research projects to assess patients' needs and to gather in-depth knowledge about the patients' condition and psychosocial functioning (e.g. QoLISSY study, KIDSCREEN study, DISBAKIDS study) (The Disabkids Group Europe, 2006; The European QoLISSY Group, 2013; The Kidscreen Group, 2006). Children and adolescents proved to be experts of their own condition and should be included in establishing PRO instruments (Matza *et al.*, 2013).

In the development process of a PRO tool, FG discussions are also used to refine and confirm a preliminary version of the new tool. For this, a cognitive debriefing should be conducted. This method is used to asses understanding, applicability and content of items from the respondent and allows a direct feedback on these aspects (Irwin & Varni *et al.*, 2009).

Especially when the instrument should be applied to a young population this process is essential to clarify thoroughly understanding of the target population. Often the process of a cognitive debriefing is combined with a quantitative pilot test of the draft version. With the data of the cognitive debriefing and pilot test possible modifications in the instrument can be undertaken, before the new instrument is used in a field- and re-test to psychometrically test it. Furthermore, FG discussions are used to figure out if any aspects are missing and not addressed in the PRO instrument, which would be relevant for the target population and the related issues. (Rothman *et al.*, 2009; The European QoLISSY Group, 2013).

In this study, the focus group methodology was used as well, to generate items for the new instrument. For this, previous focus group data was analyzed regarding the children and youth version of the International Classification of Functioning, Disability and Health. Therefore, the following paragraph concentrates on this classification concept.

2.5 International Classification of Functioning, Disability and Health

The International Classification of Functioning, Disability and Health (ICF) is developed by the World Health Organization (WHO) and aims at providing an international standardized framework to define health, health outcomes and their determinants. Furthermore, it provides a standardized language for describing the health status of an individual and thus helps to decrease difficulties in communication between different participants in the health care system. It is an organized coding scheme to assess health information and gives the opportunity to compare health care data across countries. Moreover it can be used as a research tool to assess needs, HrQoL and environmental factors of a population group, like it is done in this study (WHO, 2001). Besides this, the ICF provides a universal framework to compare the content of a HrQoL measure to assess which ICF components are covered by the items of a specific instrument. The comparison of HrQoL measures based on the ICF enables researchers or clinicians to select the most suitable instrument for HrQoL assessment or for clinical trials (Cieza & Stucki, 2005).

Originally, the health status of an individual was classified with the ICD-10 (International Classification of Diseases, Tenth Revision). This classification focuses on the diagnosis of diseases. The ICF supplements this classification, since it focuses on functioning and disability of a health condition and describes the bio-psycho-social aspects of a disease, considering environmental and personal factors. Hence, both classifications complement each other and provide a complete picture regarding the health of individuals or populations (DIMDI, 2014).

The ICF describes different components of health and not primarily consequences of a disease. Hence it can be referred to all individuals, regardless of the degree of disability to describe health and health-related conditions.

The ICF consists of two main parts including each two components (WHO, 2001):

Part 1 - Functioning and Disability:
- Body functions (b) and structures (s)
- Activities and participation (d)

Part 2 - Contextual Factors:
- Environmental factors (e)
- Personal factors (e)

In the ICF classification, the components are marked with the letters b, s, d, e. Each component is divided into several categories and these are again further divided into different subcategories. Thus, each chapter consists of individual two-, three- or four level categories to classify the health status. By selecting an appropriate code that reflects an individual's health or health-related state, the health status of an individual can be described. This code is supplemented by numeric codes, which define the degree of functioning or disability. Hence, the ICF provides a multidimensional construct that includes an interaction between the health status and environmental as well as personal factors. Figure 4 illustrates these complex interactions (WHO, 2001).

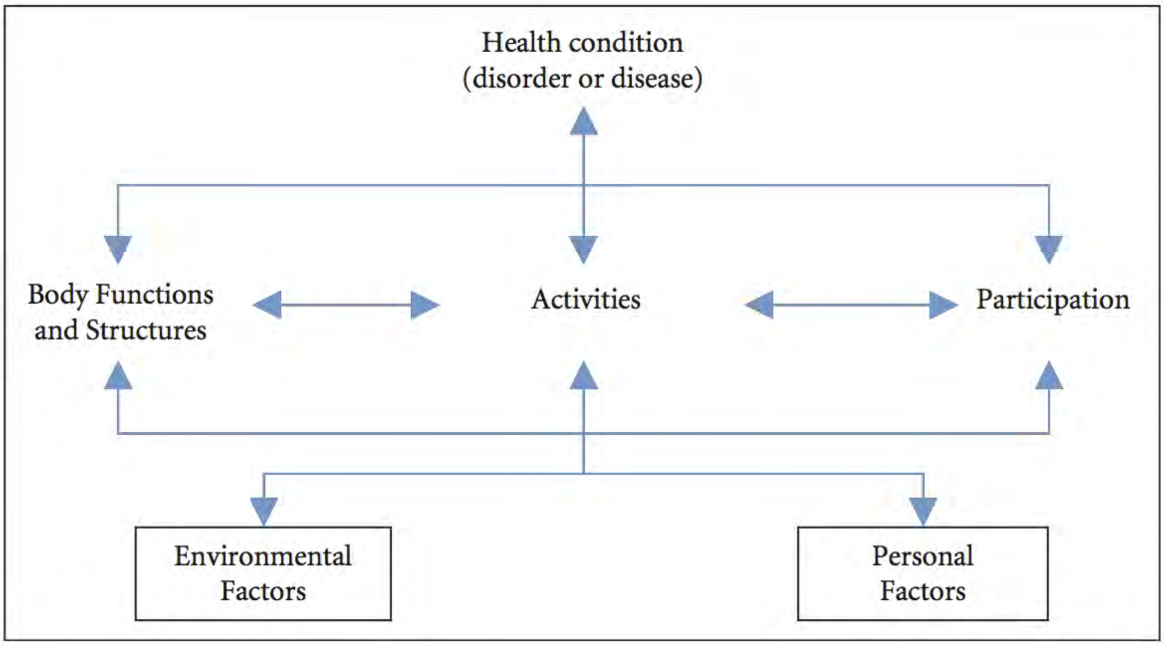

Fig. 4. Interactions between the components of ICF (WHO, 2001, p. 18)

In 2007 the WHO published a version of the ICF specifically for children and adolescents, named the International Classification of Functioning, Disability and Health, Children & Youth Version (ICF-CY) (WHO, 2007). This version derived from the original ICF classification and aims to consider specific environmental and functioning aspects, which are related to childhood and adolescent, because especially in this period of life, young people experience various changes in their psychological, physical and social development. The framework of this version is identical to the original classification. However, the domains and categories focus more on special features which are characteristic for this population group and include developmental changes of different age groups. It is essential to consider these specific characteristics because quality, intensity and effects of disability and functioning in children and adolescents differ from adults. The ICF-CY can be applied to all individuals from the moment of birth up to the age of 18 years (WHO, 2007). Since this study focuses on children and adolescents, the ICF-CY version is used for analysis of the focus group discussion.

2.6 Objective of the Study

According to the current literature, no existing PRO instrument covers all relevant domains that are important for Achondroplasia or takes into account the aspect of disproportionality that influences the functionality of these patients.

Therefore, the aim of this study is to develop a condition specific PRO instrument according to the methodology outlined by the U.S. Food and Drug Administration that captures information about burden of disease, functioning, psychosocial wellbeing and HrQoL of young patients with Achondroplasia. The main objective is to develop a pilot version of the new instrument within the framework of the ICF-CY, using qualitative data of previous FG discussions of children and adolescents with Achondroplasia and their parents.

Furthermore, the pilot version is tested in a pilot test and cognitively debriefed within new FG discussions in this patient group, conducted by trained psychologists and research scientists.

Hence, two different in-depth qualitative content analyses of FG discussions with children and adolescents with Achondroplasia and their parents are part of this study as well as a quantitative analysis of the pilot test. A subsequent field- and re-test to test the psychometric properties and reliability of the questionnaire is not part of this study.

In particular the results of the study will focus on the following research questions:

- What are the QoL/ICF-CY related domains of interest in Achondroplasia from the children’s/parents’ perspective?
- What is the impact of Achondroplasia on the child’s wellbeing and functionality from the children’s and parents’ perspective?

The study will lead to the first condition-specific instrument for children and adolescents with Achondroplasia named APLES (The Achondroplasia Personal Life Experience Scale), which can be used in a subsequent field- and re-test. It aims to clarify special limitations within the framework of the ICF-CY that are associated with the disease and contribute to improved understanding of condition- related consequences from the children’s and parents’ perspectives. Moreover, as a patient-reported outcome instrument it would identify needs from the patients’ perspective and help to optimize health care in this patient group.

3. Method

In this non-interventional study, qualitative and quantitative methods were used for data collection and analysis. The development of the APLES questionnaire was carried out in three phases, according to the methodology outlined by the U.S. Food and Drug Administration on PRO instrument development (FDA, 2009).

1. Development of a pilot version of the APLES questionnaire, using previous FG data.
2. Pilot testing and cognitive debriefing of the pilot version within new FG discussions.
3. Field- and re-test to psychometrically test the new instrument.

Phase one includes the development of a condition-specific PRO instrument, using previous FG data to assess wellbeing and functioning in consideration of the disproportionality aspect of patients with Achondroplasia. In phase two, new FG discussions and a cognitive debriefing were conducted to test the pilot version of the new instrument and to identify further relevant topics for individuals with Achondroplasia. In a subsequent third phase, a field- and re-test will be conducted to psychometrically test the new measure. However, phase three is not part of this study. Figure 5 describes the detailed development process of the APLES questionnaire of phases one and two for the current study.

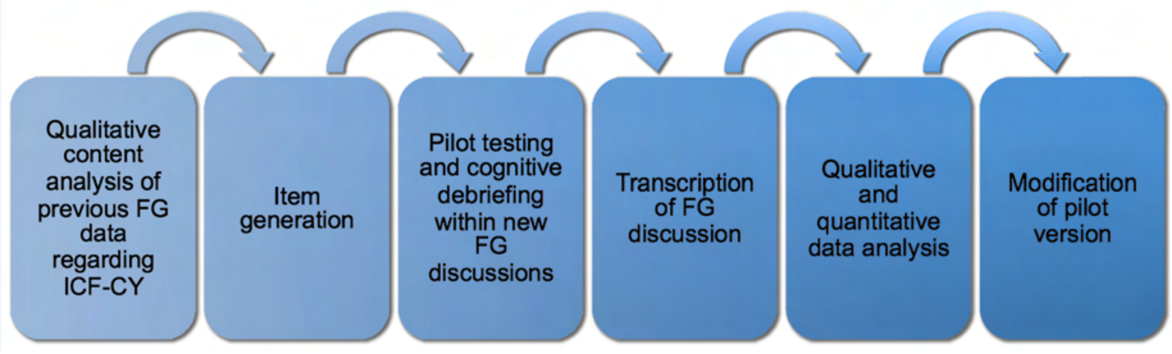

Fig. 5. Phases one and two of the questionnaire development

3.1 Instrument Development Phase I

In phase one the pilot version of the APLES questionnaire was developed. For this, FG data of a completed study funded by the German Federal Ministry of Education and Research (BMBF) was used for analysis and item generation (VeLeFaAch, 2011-2013).

Aim of the VeLeFaAch study was to assess HrQoL and psychosocial stresses and resources of children, adolescents and young adults with Achondroplasia as well as their parents' perception of their children's respective stresses and resources, using validated questionnaires and FG discussions. Afterwards, qualitative and quantitative data was used to design consulting modules for the affected persons and their families and to test these in a six-month psychosocial intervention, aiming to improve the HrQoL of these patients (Quitmann *et al.*, 2014; Rohenkohl *et al.*, 2015).

In the study eleven FG discussions had been conducted with 34 children, adolescents and young adults with Achondroplasia (8-12 years, 13-18 years, 19-28 years) and 21 parents of children/adolescents with Achondroplasia aged 8 to 18 years, who discussed major aspects of quality of life in their condition. Analyzing these FG for the current study was important to determine patients' needs and concerns regarding their health condition from their own experiences and to ensure that the items of the APLES questionnaire directly refer to the target population. Hence, the transcripts of the FG discussions with children, adolescents and young adults with Achondroplasia and their parents form the foundation for the new APLES questionnaire.

3.1.1 Qualitative Content Analysis Procedure

In a first step of the analysis, these transcripts were qualitatively analyzed. The main function of the qualitative analysis was to code the texts based on a category system that allowed allocating individual text elements to a specific category. The ICF-CY was chosen as a category system, since this classification covers all aspects of health and includes the main domains body functions, body structures and activity and participation as well as environmental factors (WHO, 2007).

In a deductive fashion, two independent raters coded verbatim statements of the affected persons and their parents regarding the codes of the ICF-CY, using the qualitative analysis program MaxQDA (VERBI-Software MaxQDA 10). By using this systematic procedure, relevant statements in the FG material were identified according to the classification outlined by the ICF-CY. Hence, the resulting category system was based on the ICF-CY, including the four main dimensions: Body functions and structures, activities and participation and environmental and personal factors, in which the statements were classified. These main categories were further divided into sub-codes according to the ICF-CY. If necessary, all four levels of the ICF-CY were used to classify the statements. During the analysis, both raters had to agree on only one code to classify a statement.

If two or more codes were possible to classify the statement, the most suitable needed to be chosen upon consultation with both raters. All FG transcripts were analyzed using the same category system. After both raters had analyzed the transcripts independently, both category systems were combined for further analysis and to assess the inter-rater agreement between both raters.

3.1.2 Item Generation

In a second step of the analysis, the category system was sorted by the number of statements per code, since codes that include many statements are likely to reflect a high relevance of the topic for the affected persons and their parents. All codes that include at least 20 statements on the third-level classification of the ICF-CY were included in the resulting ranking sequence for further item generation.

Afterwards, relevant statements of each code in the ranking sequence were summarized to determine which core statements reflect the concept. Subsequently, these statements were analyzed and items were generated out of the coded statements. Furthermore, items that address the same issue were categorized in one scale. After a final decision about the response scale and item wording, a self-report pen and paper questionnaire for children from 8 to 14 years with Achondroplasia and a parent report version for children with Achondroplasia aged 5 to 14 years with a five-point Likert scale was available for pilot testing.

3.2 Instrument Development Phase II

In the second phase the pilot version was tested within new FG discussions and a cognitive debriefing. Furthermore, issues that had not been addressed in the pilot version, but were relevant for the affected persons and their parents, were identified in the FG discussions.

3.2.1 Sample Description of the Pilot Test and Focus Groups

Upon having obtained ethics approval, patients for the FG discussions were recruited from the German Federal Association of Short Statured People and their Families (Bundesverband Kleinwüchsige Menschen und ihre Familen e.V - BKMF). Inclusion criteria were children/adolescents with diagnosed Achondroplasia aged 5 to 14 years and one parent per child who was willing to participate. Excluded were patients who were diagnosed with other forms of short stature, who had an insufficient knowledge of the German language to participate in FG discussions or to fill out the questionnaire and patients who were mentally ill or suffered from other major diseases. If the patients met the inclusion criteria and gave their consent they were included in the study.

The sample size for the FG discussions and for the pilot test was planned to consist of 12 children and adolescents with Achondroplasia and one parent of each child, as well as six parents of younger children aged 5 to 7 years, considering the age (5-7 years, 8-11 years, 12-14 years) and gender. A total of five FG discussions should be conducted in the required age groups. These included three discussions with parents of children aged 5 to 7 years, 8 to 11 years and 12 to 14 years and one discussion each with children aged 8 to 11 years and 12 to 14 years. A target number of six people was aimed at for each FG. The FG discussions were conducted within the national annual meeting of the German Federal Association of Short Statured People and their Families on May 15th 2015 in Hohenroda, Germany.

Table 1. Intended sample size for FG discussions, cognitive debriefing and pilot test in phase II

Age groups	Parents	Children/Adolescents	Total
5-7 years[1]	6	0	**6**
8-11 years	6	6	**12**
12-14 years	6	6	**12**
Total	**18**	**12**	**30**

[1] only parents were invited to participate in this age group

3.2.2 Focus Group Discussion

The FG discussions were used to gather new qualitative as well as quantitative data. A trained interviewer and an assistant moderated the FG discussions. Both needed to be familiar with group discussions as well as in work with children. The overall time for the group discussions was estimated to be about two hours. For each discussion two voice recorders were used to audiotape the session for subsequent transcription. To ensure that each session was carried out similarly, the moderator conducted the sessions based on a previously developed guideline.

The FG were organized in three sessions: (1) introduction, (2) questionnaire completion and cognitive debriefing, and (3) concept elicitation.

1. Introduction

In the introduction session, the moderators introduced themselves and explained the organization and purpose of the meeting. Afterwards the participants were asked to briefly introduce themselves.

2. Questionnaire completion and cognitive debriefing

In the second part, the participants filled out the new APLES questionnaire, as a pilot test. After filling out a scale, each question in the respective scale was cognitively debriefed. During the debriefing, the participants were specifically asked to evaluate the items in terms of clarity, sensitivity, importance, and relevance for their personal situation to gather more detailed opinions from the respondents. Furthermore, the moderator asked questions concerning the general assessment of the questionnaire (e.g. “What do you think of the questionnaire you have just completed? Why?”) and concerning the alignment of the questionnaire to the condition (e.g. “Do you think the questions/items/statements of the questionnaire reflect the feelings or the problems you have (your child has) in daily life?”, “Do you think the questions/items/statements of the questionnaire are related with Achondroplasia?”, or “Do you think that the questionnaire really reflect what you are (your child is) experiencing, on a daily basis?”).

3. Concept elicitation

During the last session, the participants were asked one after another if any aspects were missing in the questionnaire that were relevant for their condition. By this, detailed information about life experiences, emotions, needs as well as specific aspects concerning quality of life got clear, and functional effects of Achondroplasia were openly discussed.

The pilot test and cognitive debriefing data was completed using clinical and socio-demographic data of the patients and their parents, which was assessed by a trained physician on the same day. Clinical data included: age, sex, weight, height, arm range, seat height, presence of severe chronic illness, mother’s and father’s height.

3.2.3 Transcription

All interviews were recorded and subsequently literally transcribed, using the transcription software f4 (Version v4.2 – audiotranskription.de). Incomprehensible words were marked with three question marks (???) and clear and longer pauses were marked with suspension points (…). Short pauses, intonation, laughs, sighs or other linguistic characteristics were not specifically marked. Furthermore, fillers such as ‘ah’, ‘hm’ or ‘erm’ were not transcribed. Each statement was annotated with a timestamp and the speakers were marked with an ID to reproduce who has said what and when and to ensure data privacy. All transcripts were filed into the respective parent or child groups as well as the respective age groups. Irrelevant text passages that did not refer to the topic were removed.

3.2.4 Data Analysis

First, the sample of the FG discussions and the pilot test was analyzed. The participants were described in terms of sample size (n), age (mean, standard deviation), gender distribution and height (mean, standard deviation). Then qualitative data of the FG discussions as well as quantitative data of the pilot test were analyzed.

3.2.4.1 Qualitative Data Analysis

After having transcribed the FG discussions, each transcript was screened to abstract relevant verbatim statements. The analysis consisted of a descriptive analysis of the cognitive debriefing and concept elicitation of children and parent FG discussions in the different age groups. With regard to the cognitive debriefing, it was assessed whether the items are understandable and applicable to the target population. Furthermore, suggestions for improvement of items or rewording were abstracted. Regarding the concept elicitation, further concepts and concerns that were important to the participants, but not addressed in the pilot version, were analyzed and described.

3.2.4.2 Quantitative Data Analysis

A data mask for the pilot test was prepared with the statistical analysis program SPSS (IBM SPSS Statistics V 18) and subsequently completed with the pilot test data to perform statistical analysis.

In the analysis, operating characteristics of the items such as mean (*M*) and standard deviation (*SD*) were analyzed on a scale level, for both children and parents. All scores were transformed from raw scores to 0 to 100 scores, with higher values representing higher functioning and a higher HrQoL. Criterion validity of the APLES pilot version was tested via correlation analysis. Each item was analyzed in correlation to the Total Score, height and proportionality of child and parent data, using Pearson Correlation Coefficient (*r*). For the analysis, the variable proportionality was calculated using the following formula: *seat height in cm/ (height in cm –seat height in cm)*. All statistical analyses were performed using SPSS (IBM SPSS Statistics V 18).

3.2.5 Modification of Pilot Version

The results of the quantitative and qualitative analysis of the pilot version were used to further guide the selection of the items and to modify the pilot version. The final decision which items were included in the field test version of the questionnaire was based on the quantitative results of the correlation analysis and on qualitative results of the FG discussions with the cognitive debriefing. All items that had a correlation value of $r \geq 0.4$, and were considered as appropriate and relevant for Achondroplasia by the participants on the FG discussions, were included in the refined version of the questionnaire. Furthermore, results of the cognitive debriefing were used to improve wording of the items to ensure comprehensibility of the target population. The refined questionnaire will be used in a subsequent field and re-test, but this is not part of the current study.

4. Results

The following paragraphs show the results of the first and second phase of the instrument development. First, qualitative results of the FG analysis are described and the pilot version of the APLES questionnaire is presented. Then, quantitative results of the pilot test and qualitative results of the new FG discussions are described within the second phase.

4.1 Results of Phase I

4.1.1 Sample Description

The sample of the FG discussions used for the first analysis included in total 34 children, adolescents and young adults in the age groups 8-12 years, 13-18 years and 19-28 years. The participants were distributed very unevenly over the groups with regard to sex and age groups. Altogether there were more male than female participants and most participants were in the age group 19-28 years (see table 2).

Besides children, adolescents and young adults, the sample included 21 parents of children (8-12 years) and adolescents (8-18 years) with Achondroplasia. Predominantly mothers of the affected children and adolescents were represented in the FG (18 mothers, 3 fathers). In the age group 8-12 years, five parents of girls and four parents of boys participated. In the age group 13-18 years, three parents of girls and nine parents of boys participated in the groups (see table 2).

Table 2. Sample characteristics of children, adolescents and young adults with Achondroplasia and their parents in the FG discussions in phase I

Age group	**Ch/Ad/yA**		**Parents**		**Total Ch/Ad/yA**	**Total parents**
	female	male	female	male		
8-12 years	1	4	5	4	**5**	**9**
13-18 years	2	12	3	9	**14**	**12**
19-28 years	11	4	0	0	**15**	**0**
Total	**14**	**20**	**8**	**13**	**34**	**21**

The sample was divided in eleven FG discussions with four to six participants per group. These included four parent FG discussions with two discussions with parents of children aged 8-12 years and two discussions with parents of children aged 13-18 years; one FG with children aged 8-12 years and three FG with adolescents aged 13-18 years as well as three FG with young adults aged 19-28 years.

4.1.2 Category System According to the ICF-CY

In the first step of the instrument development, these FG transcripts were analyzed to generate items for the pilot version. Statements in the FG analysis were coded based on a category system which included codes of the ICF-CY that were voiced by children, adolescents and young adults with Achondroplasia and their parents in the FG discussions. The resulting category system consisted of 125 codes in total in the four main components body functions, body structures, activities and participation and environmental factors as outlined by the ICF-CY. Depending on the issues addressed in the FG discussions, these four main categories were further divided into sub-codes.

The component "body functions" describes the physiological functioning of body systems. The category system of this dimension includes 20 detailed sub-codes of the ICF-CY that are allocated to the four main categories "mental functions", "sensory functions and pain", "functions of the digestive metabolic and endocrine system" and "functions of the cardiovascular, hematological, immunological and respiratory systems" based on the first-level of the ICF-CY classification (see table 3).

Table 3. Category system in the component "body functions"

	One-level classification	**Two-level classification/Detailed classification**
Body Functions	Mental functions	b 1801 Body image
		b 1800 Experience of self
		b Confidence
		b Optimism
		b 1348 Sleep functions, other specified
		b 1260 extraversion
		b 1263 Psychic stability
	Sensory functions and pain	b 2801 Pain in body part
		b 2702 Sensitivity to pressure
		b 265 Touch function
		b 2308 Hearing functions, other specified
		b 28015 Pain in lower limb
		b 28013 Pain in back
		b 28016 Pain in joints
		b 280 Sensation of pain
	Functions of the digestive metabolic and endocrine system	b 530 Weight maintenance functions
		b 560 Growth maintenance functions
		b 515 Digestive functions
	Functions of the cardiovascular, hematological, immunological and respiratory systems	b 440 Respiration functions
		b 4401 Respiratory rhythm

The component “body structures” classifies anatomical parts of the body like organs or extremities and their related structures. The main categories that were voiced by the affected people and their parents include the categories “structures of the nervous system”, “skin and related structures”, “structures related to movement”, “structures related to digestive, metabolic and endocrine systems”, “structures involved in voice and speech” and “the eye, ear and related structures”, which are divided in 27 detailed codes.

Table 4. Category system in the component “body structures”

	One-level classification	**Two-level classification/Detailed classification**
Body Structures	Structures of the nervous system	s 1200 Structure of spinal cord
		s 110 Structure of brain
	Skin and related structures	s 8109 Structures of areas of skin, unspecified
	Structures related to movement	s 760 Structure of trunk
		s 710 Structure of head and neck region
		s 7302 Structure of hand
		s 7201 Bones of face
		s 75011 Knee joint
		s 75019 Structure of lower leg, unspecified
		s 71008 Bones of cranium, other specified
		s 7108 Structure of head and neck region, other specified
		s 7508 Structure of lower extremity, other specified
		s 740 Structure of pelvic region
		s 73020 Bones of hand
		s 7600 Structure of vertebral column
		s 7500 Structure of thigh
		s 7701 Joints
		s 730 Structure of upper extremity
		s 7702 Muscles
		s 75021 Ankle joint and joints of foot an toes
		s 75029 Structures of ankle and foot, unspecified
	Structures related to the digestive, metabolic and endocrine systems	s 530 Structure of stomach
	Structures involved in voice and speech	s 3301 Oral pharynx
	The eye, ear and related structures	s 2509 Structure of the middle ear, unspecified
		s 2508 Structure of the middle ear, other specified
		s 299 Eye, ear and related structures, unspecified
		s 260 Structure of inner ear

52 codes of the category system are allocated to the component “activities and participation”. This dimension focuses on activities, which is the performance of a task or action and on participation, which is described as the inclusion in life situations. In the category system of this dimension the main categories “community, social and civic life”, “major life areas”, “interpersonal interactions and relationships”, “domestic life”, “self-care”, “mobility” and “general tasks and demands” were voiced by the affected people and their parents (see table 5).

Table 5. Category system in the component “activities and participation”

	One-level classification	**Two-level classification/Detailed classification**
Activities and Participation	Community, social and civic life	d 9209 Recreation and leisure, unspecified
		d 9103 Informal community life
		d 9208 Recreation and leisure, other specified
		d 9201 Sports
		d 9202 Arts and culture
		d 9200 Play
		d Informal associations
	Major life areas	d 820 School education
		d 825 Vocational training
		d 8502 Full-time employment
		d 830 Higher education
	Interpersonal interactions and relationships	d 7601 Child-parent relationship
		d 7402 Relating with equals
		d 710 Basic interpersonal interactions
		d 7602 Sibling relationships
		d 7500 Informal relationship with friends
		d 7509 Informal social relationships, unspecified
		d 7700 Romantic relationships
		d 730 Relating with strangers
		d 7504 Informal relationships with peers
	Domestic life	d 6309 Preparing meals, unspecified
		d 699 Domestic life, unspecified
		d 630 preparing meals
		d 699 Domestic life, unspecified
		d 6609 Assisting others, unspecified
		d 6200 Shopping
		d 6406 Helping to do housework
	Self-care	d 5404 Choosing appropriate clothing
		d 530 Toileting
		d 53011 Carrying out defecation appropriately
		d 5200 Caring for skin
		d 5701 Managing diet and fitness
	Mobility	d 4700 Using human-powered vehicles
		d 499 Mobility, unspecified
		d 498 Mobility, other specified
		d 450 Walking
		d 4509 Walking, unspecified
		d 480 Riding animals for transportation
		d 4702 Using public motorized transportation
		d 4452 Reaching
		d 4501 Walking long distances
		d 4750 driving human-powered transportation
		d 4751 Driving motorized vehicles
		d 4554 Swimming
		d 4551 Climbing
		d 4300 Lifting
		d 4105 Bending
		d 4552 Running
		d 4103 Lifting and carrying objects
	General tasks and demands	d 230 Carrying out daily routine
		d 2409 Handling stress and other psychological demands, unspec.
		d 2301 Managing daily routine

The last component "environmental factors" describes the environment in which people live their lives according to the physical, social and attitudinal environment. In this component, the category system consists of 26 codes assigned to the main categories "support and relationship", "services, systems and policies", "products and technology" and "attitude". These were further sub-divided as shown in table 6.

Table 6. Category system in the dimension "environmental factors"

	One-level classification	Two-level classification/Detailed classification
Environmental Factors	Support and relationships	e 320 Friends
		e 310 Immediate family
		e 340 Personal care providers and personal assistants
		e Other professionals
		e 345 Strangers
		e 399 Support and relationships, unspecified
		e 325 Acquaintances, peers, colleagues, neighbors and community
		e 315 Extended family
		e 330 People in position of authority
		e 350 Domesticated animals
	Services, systems and policies	e 5800 Health services
		e 5751 General social support systems
	Products and technology	e 130 Products and technology for education
		e 1501 Design, construction and building products and technology for gaining access to facilities inside building for public use
		e 1201 Assistive products and technology for personal indoor and outdoor mobility transportation
		e 1351 Assistive products and technology for employment
		e 1551 Design, construction and building products and technology for gaining access to facilities in buildings for private use
		e 1151 Assistive products and technology for personal use in daily living
		e 1150 General products and technology for personal use in daily living
	Attitude	e 415 Individual attitudes of extended family members
		e 450 Individual attitudes of health professionals
		e 410 Individual attitudes of immediate family members
		e 445 Individual attitudes of strangers
		e 425 Individual attitudes of acquaintances, peers, colleagues, neighbors and community
		e 420 Individual attitude of friends
		e 498 Attitudes, other specified

4.1.3 Item Development

The ICF-CY based category system as described above reflects the content of the FG discussions. It was subsequently used to allocate individual verbatim statements in the 11 transcripts of children, adolescents and young adults with Achondroplasia and parents of children and adolescents with Achondroplasia to a specific code. This process led to a rich material of statements classified in the components and sub-codes, as outlined in the category system.

In the analysis of the FG discussions, both raters identified a total of 1950 statements. Of these, 1200 statements were allocated to children, adolescents and young adults with Achondroplasia and 750 statements were allocated to parents of children and adolescents with Achondroplasia. Inter-rater agreement was satisfactory with about 80% agreement of coded statements by both raters.

The results of the FG analysis led a preliminary conceptual model of the APLES questionnaire, which consisted of statements that were allocated to a specific ICF-CY code of the category system. This model was further used to guide the process of item generation.

For the process of item generation, the statement list needed to be reduced to identify the most relevant topics and contents that were voiced by children, adolescents and young adults with Achondroplasia as well as their parents. Individual codes that were assigned to many statements are likely to reflect a higher relevance for the affected people and their parents. Therefore, the codes of the category system were organized in a ranking order, which included all codes with 20 or more assigned statements in the third-level classification of the ICF-CY. Thus, a total of 30 codes with 20 or more statements were included in this ranking. Of these codes, statements were most frequently allocated to the component “environmental factors” with a total of 723 codings (43,9% of 1647 codings). There were 467 codings (28,4%) in the component “activities and participation”, 280 codings (17,0%) in the component “body functions” and 177 codings (10,7%) in the component “body structures” (see table 7).

On a detailed code-level, the majority of the statements were allocated to the ICF-CY code e 410 “individual attitudes of family members” with 145 codings, followed by 142 statements assigned to the code b 180 “experience of self and time functions” which includes statements concerning the body image and experience of oneself. Statements regarding the “individual attitude of strangers” (e 445) were also frequently identified in the FG analysis. In this context, this code classifies statements, which refer to the behavior of strangers like starring or laughing at the affected people. Furthermore, the affected people and their parents often mentioned topics concerning “handling stress and other psychological demands” (d 420) (see table 7).

Table 7. Ranking list of statements assigned to codes of the ICF-CY

Domains	One-level classification	Two-level classification	Third-level classification	Codings
Environmental Factors	Attitudes		e 410 Individual attitudes of immediate family members	145
Body Functions	Mental Functions	Specific mental functions	b 180 Experience of self and time functions	142
Environmental Factors	Attitudes		e 445 Individual attitudes of strangers	136
Activities and Participation	General tasks and demands		d 240 Handling stress and other psychological demands	130
Environmental Factors	Products and technology		e 115 Products and technology for personal use in daily living	95
Environmental Factors	Support and relationships		e 310 Immediate family	88
Body Structures	Structures related to movement		s 750 Structure of lower extremity	79
Body Functions	Sensory functions and pain	Pain	b 280 Sensation of pain	77
Activities and Participation	Community, social and civic life		d 920 Recreation and leisure	71
Environmental Factors	Services, systems and policies		e 580 Health services, systems and policies	65
Activities and Participation	Interpersonal interactions and relationships	Particular interpersonal relationships	d 760 Family relationships	60
Environmental Factors	Attitudes		e 420 Individual attitudes of friends	51
Body Structures	Structures related to movement		s 710 Structure of head and neck region	44
Activities and Participation	Mobility	Carrying, moving and handling objects	d 445 Hand and arm use	42
Body Functions	Mental Functions	Global mental functions	b 126 Temperament and personality functions	41
Environmental Factors	Products and technology		e 155 Design, construction and building products and technology of buildings for private use	34
Body Structures	The eye, ear and related structures		s 299 Eye, ear and related structures, unspecified	31
Environmental Factors	Attitudes		e 425 Individual attitudes of acquaintances, peers colleagues, neighbours and community members	30
Environmental Factors	Attitudes		e 450 Individual attitudes of health professionals	30
Activities and Participation	Interpersonal interactions and relationships	Particular interpersonal relationships	d 730 Relating with strangers	29
Environmental Factors	Support and relationships		e 320 Friends	26
Activities and Participation	Mobility	Walking and moving	d 455 Moving around	24
Activities and Participation	Interpersonal interactions and relationships	General interpersonal interactions	d 710 Basic interpersonal interactions	24
Body Structures	Structures related to movement		s 760 Structure of trunk	23
Activities and Participation	Mobility	Walking and moving	d 450 Walking	23
Activities and Participation	Self-care		d 540 Dressing	23
Activities and Participation	Interpersonal interactions and relationships	Particular interpersonal relationships	d 770 Intimate relationships	21
Body Functions	Functions of the digestive, metabolic and endocrine systems	Functions related to metabolism and the endocrine system	b 560 Growth maintenance functions	20
Activities and Participation	Domestic life	Acquisition of necessities	d 620 Acquisition of goods and services	20
Environmental Factors	Products and technology		e 120 Products and technology for personal indoor and outdoor mobility and transportation	20

To further guide the reduction of the statement list, repetitious or unclear statements assigned to the codes of the ranking list were removed. Hence, 30 codes with each up to 6 example statements remained in the ranking order. By this, statements that reflect the most frequently addressed ideas and mentioned concepts of the allocated ICF-CY codes were identified and the content used for the subsequent item generation. Subsequently, items were developed and formulated based on these statements. Thus, each item is directly linked to one of the 30 included ICF-CY codes of the ranking list.

4.1.3.1 Items of the Component “Body Functions”

In the component “body functions” nine items were formulated based on statements that were allocated to a particular ICF-CY code of the category system in this component. Table 8 shows the ICF-CY code in this component and the corresponding items in the child version.

In total seven items were based on the ICF-CY category “mental functions”. Of these four items were generated out of statements with the ICF-CY code “experience of self and time functions”. Statements with this code refer to the child’s attitude and experience of him or herself and how they feel and differ from other children. In general children/adolescents and young adults with Achondroplasia state to be satisfied with their appearance, although they often attract attention due to their short statured body. Furthermore, some state to have problems concerning their weight and wish to be taller. The generated items based on this code reflect these statements. Moreover, three items were generated out of statements with the code “temperament and personality functions” of this category. Statements allocated to this code reflect that affected persons feel good and self-confident in their situation. Furthermore, most of them state to be independent in daily life.

One item was generated out of the code “sensation of pain”. It summarizes statements that refer to any kind of pain. Affected persons state to have more pain than others and often suffer from headache, back pain, earache and joint pain.

No item was formulated for the ICF-CY code b 560 “growth maintenance functions” because statements according to this code refer mostly to the wish of being taller. This issue is also addressed in the code b 180 “experience of self and time functions” with the item “I like my body” and therefore was not addressed again.

Table 8. Items based on the ICF-CY component "body functions"

	ICF-CY Code	Item
Body Functions	b 180 Experience of self and time functions	I find myself to be too fat
		I like my body
		I always draw attention from others because of my body
		I think I am just like others and just look somewhat different
	b 280 Sensation of pain	I am often in pain
	b 126 Temperament and personality functions	I feel okay the way I am
		I dare to ask strangers for help
		Attracting attention by strangers because of my short stature hurts me
		I am independent in daily life
	b 560 Growth maintenance functions	-

4.1.3.2 Items of the Component "Body Structures"

In total five items are based on the component "body structures" (see table 9). Of these, four items refer to the ICF-CY category "structures related to movement". In detail, one item was generated out of statements with the code "structure of lower extremity". Statements within this code address different issues of the lower extremity that people with Achondroplasia experience, like bandy legs, pain or surgeries. Another item was formulated of statements with the ICF-CY code "structure of trunk". Statements within this code address various complaints related to the trunk like a narrowed spinal canal or a hyperlordosis and the related consequences. The remaining two items of this category were formulated of statements with the code "structure of head and neck region". Statements within this code refer to the large head and other facial characteristics like a high forehead or the narrowed midface that are mentioned by children, adolescents and young adults in the FG discussions.

One item was generated of the code "eye, ear and related structures, unspecified". This item summarizes frequently mentioned ear issues like otitis media.

Table 9. Items based on the ICF-CY component "body structures"

	ICF-CY Code	Item
Body Structures	s 750 Structure of lower extremity	The shape and length of my legs causes problems for me
	s 710 Structure of head and neck region	I find my head to be too big
		I like my face
	s 299 Eye, ear and related structures, unspecified	I had/have physical problems with my ears
	s 760 Structure of trunk	I experience physical complaints (e.g. my legs falling asleep)

4.1.3.3 Items of the Component “Activities and Participation”

In total 18 items were formulated based on the component “activities and participation” (see table 10). Seven items of this component refer to the ICF-CY category “interpersonal interactions and relationships”. Of these, two items were generated out of statements with the ICF-CY code “family relationships”. This code includes statements that address issues within a family. Often parents of children and adolescents with Achondroplasia state to treat them specially or pay them much more attention than their other children. Another three items were generated out of statements with the ICF-CY code “relating with strangers”. Children, adolescents and young adults are often confronted with reactions of strangers on their external appearance. According to the statements they have different ways of dealing with strangers. Some children, adolescents and young adults state to be confident and talk to strangers, if they stare at them, others prefer to ignore them. Furthermore, one item refers to the ICF-CY code “basic interpersonal interactions”. It summarizes different statements of the affected persons concerning human interaction. They appeal to more respect and tolerance of the society towards people with Achondroplasia. The remaining item that refers to the category “interpersonal interactions and relationships” is based on the ICF-CY code “intimate relationships”. Statements within this code refer mostly to male adolescents and young adults who state to have problems finding a girlfriend.

Four items refer to the ICF-CY category “mobility”. In detail, two items of this category are based on the ICF-CY code “hand and arm use”. This code includes all statements of children, adolescents and young adults with Achondroplasia that refer to the problem of not being able to reach out to everything because of too short arms. Another item was generated of statements with the ICF-CY code “moving around”. The item reflects statements of children, adolescents and young adults with Achondroplasia concerning running and moving. Most of them state to be not able to run fast and are not able to keep up with their friends. Furthermore, one item was generated out of the ICF-CY code “walking”. It summarizes statements that refer to any kind of problems with walking. Affected persons state to be not able to walk for a long time or long distances due to pain.

Another item was formulated based on statements with the ICF-CY code “handling stress and other psychological demands”. These statements reflect how the affected persons feel with regard to strangers or how they cope with negative experience with strangers. Most of the coping strategies identified in the FG analysis included listening to music or talking to immediate family members when having a bad experience with strangers.

Furthermore, four items are based on the ICF-CY code “recreation and leisure“ in the category “community social and civic life”. They reflect the most important topics that were addressed in the statements within this code. These include being able to do various kinds of sports like horse riding or swimming but also to experience restrictions in leisure time due to short stature like not being allowed to ride on a roller coaster.

Moreover, one item was generated out of the statements with the ICF-CY code “dressing”. Children, adolescents and young adults with Achondroplasia state to have problems to find age-appropriate clothing and shoes due to their short statured body. Furthermore, the large head circumference complicates putting on a shirt or wearing caps or hats. The formulated item based on this ICF-CY code reflects these issues.

The ICF-CY code “acquisition of goods and services” contains statements of children, adolescents and young adults with Achondroplasia who state to have problems when shopping groceries or other goods because they are not able to reach everything in the store and are dependent on other people. One item that reflects these problems was generated out of the statements.

Table 10. Items based on the ICF-CY component “activities and participation”

Activities and Participation	**ICF-CY Code**	**Item**
	d 240 Handling stress and other psychological demands	Reactions of strangers bother me
	d 920 Recreation and leisure	I am able to do the sports that I want to
		In my free time I can do what I like
		I spend too much time at the doctors/therapists
		I need to go to the doctor/therapist quite often
	d 760 Family relationships	My family shows consideration for me
		I am treated pretty normal by my family
	d 445 Hand and arm use	I am able to reach out to everything
		The shape and length of my arms causes problems for me
	d 730 Relating with strangers	I am willing to answer questions of strangers about my short stature
		If strangers stare at me I talk to them
		I am open minded towards strangers
	d 455 Moving around	I am not able to run fast
	d 710 Basic interpersonal interactions	A lot of people are not familiar with short stature
	d 450 Walking	It is hard for me to move long distances
	d 540 Dressing	I find clothes I like
	d 770 Intimate relationships	I have problems to find a friend/boyfriend/girlfriend
	d 620 Acquisition of goods and services	Shopping is difficult for me (e.g. because of not being able to reach things)

4.1.3.4 Items of the Component “Environmental Factors”

In total 27 items were formulated based on the ICF-CY codes in the component “environmental factors” (see table 11). In detail, 14 items refer to the ICF-CY category “attitudes”. Of these, two items refer to the ICF-CY code “individual attitudes of immediate family members”. The statements within this code refer to the attitude of the family towards the child, adolescent or young adult with Achondroplasia. Some parents state to treat their child differently from others due to their lack of independence. Still, short stature is not seen as an important topic for the family because they got used to the special situation when having a short statured child. Another four items are based on the ICF-CY code “individual attitude of strangers”. Statements within this code refer to the experience with strangers. It is frequently mentioned that strangers stare or laugh at children, adolescents and young adults with Achondroplasia and often treat them not age appropriately, since they estimate their age far younger. Besides, the statements reveal that some strangers are interested in the condition and speak to the affected persons concerning their short stature; others do not dare to talk to them. Furthermore, two items are based on the ICF-CY code “individual attitudes of friends”. This code refers to statements that reflect how friends of children, adolescents and young adults with Achondroplasia think about their condition and particularly their short statured body. The FG analysis revealed that friends of the affected persons treat them in a normal way and take their condition and external appearance for granted. Another six items within the category “attitudes” were generated out of statements with the ICF-CY code “individual attitudes of acquaintances, peers colleagues, neighbors and community members”. The statements within this code refer primarily to the attitudes of peers in school and kindergarten. Some children, adolescents and young adults with Achondroplasia describe negative experiences with peers like being teased or laughed at and getting special treatment by teachers. Others describe positive experiences like being supported by peers and teachers.

Eight further items refer to the ICF-CY category “support and relationships”. In detail, three items were formulated of statements with the ICF-CY code “immediate family“. These statements describe in which way parents of children and adolescents with Achondroplasia support their children. They need to show consideration for their children in daily life and need to support them physically as well as psychologically. Hence, they tend to act overprotective. Furthermore, five items of this category are based on the ICF-CY code “friends”. This code refers to statements that reflect how friends of children, adolescents and young adults with Achondroplasia support them. They provide emotional support but also physical support if needed. Moreover, they show consideration for them during activities and protect them if necessary.

Further three items refer to the ICF-CY category “products and technology”. Of these, one item was generated of statements with the ICF-CY code “products and technology for personal use in daily living”. These statements summarize objects or tools that are useful for the affected persons in daily life. The most frequently addressed tool is a stool. Children, adolescents and young adults state to use stools very often to reach out to things. Most of them have several stools in the household at different places. Another item was generated out of the ICF-CY code “products and technology for personal indoor and outdoor mobility and transportation”. Statements with this code summarize different transportation options of the affected people. For an independent mobility a lot of them use scooters or specially designed bicycles that are adapted to short statured people to move around. A further item was generated out of the ICF-CY code “design, construction and building products and technology of buildings for private use”. Statements with this code reflect modifications in the home of the affected people. Common modifications that are described in the FG discussions include a lowering of the washbasin or light switch and a reconstruction of the kitchen.

An additional item was generated out of the ICF-CY code “health services, systems and policies”. It summarizes statements that refer to any kind of medical services. Affected persons state to see different health professionals like physiotherapists, speech therapists or osteopaths. Furthermore statements with this code reflect that surgeries are a common medical procedure for the affected persons. Most of them already had various kinds of surgeries.

Table 11. Items based on the ICF-CY component “environmental factors”

	ICF-CY Code	Item
Environmental Factors	e 410 Individual attitudes of immediate family members	My short stature is a big deal to my family
		I am not treated differently from others by my family
	e 445 Individual attitudes of strangers	I can accomplish more than others might think
		I am treated by strangers like other kids my age
		Strangers often make fun of me or laugh at me behind my back
		Strangers often stare at me
	e 115 Products and technology for personal use in daily living	With the help of my tools I am able to master daily life by myself (e.g. stool)
	e 310 Immediate family	Because of my short stature my family is less confident in me
		Members of my family help me in daily life
		I am allowed less than other children my age
	e 580 Health services, systems and policies	I need to see many different doctors/therapists
		I have already had many surgeries
	e 420 Individual attitudes of friends	My friends think my height and appearance is pretty normal
		My friends think my short stature is pretty normal
	e 155 Design, construction and building products and technology of buildings for private use	At home a lot of things are adapted to my size (reconstruction, furniture)
	e 425 Individual attitudes of acquaintances, peers colleagues, neighbours and community members	Because of my height I am teased or laughed at by others in school/kindergarten
		Other kids at school/kindergarten help me
		I am treated considerately at school/kindergarten
		I am treated differently in school/kindergarten than other kids
		My short stature is a big topic in school/kindergarten
		I am supported by my teachers
	e 320 Friends	I get no special treatment by my friends
		My friends are considerate of me
		My friends help me
		My friends stand up for me
		My friends protect me
	e 120 Products and technology for personal indoor and outdoor mobility and transportation	With the help of assistive tools I am able to move around

A detailed overview of all original example statements of each ICF-CY code in the ranking order is shown in annex 1 (in German).

4.1.4 Pilot Version of the APLES Questionnaire

To carry out the pilot test items that derived from the qualitative FG analysis of the previous study were arranged in a preliminary questionnaire format for children (see annex 2 a) and parent report (see annex 2 b). The pilot version consisted of a total of 59 items, that were assigned to 9 scales:

- Self-perception
- Family
- Friends
- Recreation
- Kindergarten/School
- Medical
- Strangers
- Aids
- Physical

The first scale “self-perception” includes in total seven items. The items address how the child perceives his or her own body and assess the child’s general beliefs about his or her short stature.

On the scale “family” seven items address how the family is coping with the child’s condition in everyday life and how the child is treated by his or her family.

The scale “friends” includes eight items. The items address how short stature influences relationships with friends and what the child’s friends think about short stature.

On the scale “recreation”, four items address how short stature might interfere with the child’s leisure activities.

The scale “kindergarten/school” includes six items. The items refer to the way short stature influences the child’s social life at school/kindergarten. Furthermore it addresses feelings of social integration and acceptance by others.

On the scale “medical”, four items refer to medical and therapeutical aspects of the disease.

The scale “strangers” includes ten items. The items address how strangers deal with and react towards the child and how the child is coping with negative experiences with strangers due to their reaction on his or her short stature.

On the scale “aids”, three items refer to tools or reconstructions, which should help to facilitate the daily life of the children.

The last scale “physical” includes ten items. The items refer to the physical limitations that the child can experience in everyday life due to his or her short stature.

As a response scale a 5-point Likert scale was chosen, which includes the response options “I don´t agree at all”, “I agree somewhat”, “I agree”, “I agree a lot” and “I totally agree”.

The pilot questionnaire is constructed as a self-reported pen and paper questionnaire for children from 8 to 14 years with Achondroplasia. A parent report was derived from the child version. This version includes the same items but from the perspective of the parents. It is available for parents of children with Achondroplasia aged 5 to 14 years.

Subsequently, this questionnaire was used in a pilot test. Moreover, it was cognitively debriefed to evaluate understanding and applicability to the target population within new FG discussions. Results of this second phase of the instrument development are presented in the following paragraph.

4.2 Results of Phase II

4.2.1 Sample Description of the Focus Groups and Pilot Test

Participants for the pilot test and FG discussions were recruited from the BKMF. All patients and their parents who were willing to participate in the study and met the inclusion criteria were included in the study. Since the pilot test was conducted within the FG discussions, the identical sample was used to gather quantitative data of the pilot test and qualitative data of the FG discussions.

Overall, the actual sample consisted of 27 participants across the different age groups. The intended sample size for children and adolescents was not achieved, but the intended sample size for parents was achieved. However, the distribution of parents was very uneven with regard to the different age groups of the children or adolescents (see table 12).

Table 12. Actual sample size of children and adolescents with Achondroplasia and their parents for the FG discussions and pilot test with difference to intended sample size

Age groups	Parents	Children/Adolescents	Total
5-7 years[1]	12 (+6)	0 (0)	**12 (+6)**
8-11 years	5 (-1)	4 (-2)	**9 (-3)**
12-14 years	3 (-3)	3 (-3)	**6 (-6)**
Total	**20 (+2)**	**7 (-5)**	**27 (-3)**

[1] only parents were invited to participate in this age group

The analyzed sample included in total seven children and adolescents with Achondroplasia in the age groups 8-11 years and 12-14 years. The participants were distributed very unevenly with regard to sex. In the age group 8-11 one girl and three boys participated and in the age group 12-14 years no male patients were represented. The mean age in the age group 8-11 years was 10.2 years and in the age group 12-14 years 13.3 years. With regard to height, children in the age group 8-11 years had a mean height of 106.10 cm (*SD*: 5.10) and adolescents in the age group 12-14 years had a mean height of 118.66 cm (*SD*: 8.39) (see table 13).

Next to children and adolescents, the sample included 20 parents of children and adolescents with Achondroplasia. Participating parents were in all cases father or mother of the participating children and adolescents, except for one additional parent who participated in the FG discussions in the age group 8-11 years. The parents were distributed very unevenly across the different age groups. Most parents participated in the age group of children with Achondroplasia aged 5-7 years. In this age group, five parents of boys and seven parents of girls participated. In the age group 8-11 years four parents of boys and one parent of a girl participated in the FG discussions, and in the age group 12-14 years only parents of girls participated. The mean height of children aged 5-7 years was 87.07 cm (*SD*: 7.3) (see table 13).

In total six FG discussions were conducted, including four parent FG discussions in each age group. In the age group 5-7 years two FG discussions were conducted due to the high number of participating parents in this age group. Furthermore, two FG discussions with children and adolescents were conducted, one each in the age groups 8-11 years and 12-14 years. Parent FG discussion lasted on an average about 2–2.5 hours; children and adolescent FG discussions were shorter with an average time of 1.5-2 hours.

Table 13. Sociodemographic and clinical characteristics of children and adolescents in the FG discussions and pilot test

Age group		**Mean Age (*SD*)**	**Mean height in cm (SD)**	**Gender**	
				Male	**Female**
5-7 years	Parents	5.59 (*SD*: 1.01)	87.07 (*SD*:7.32)	5	7
8-11 years	Children	10.17 (*SD*: 1.07)	106.10 (*SD:*5.10)	3	1
	Parents	9.68 (*SD*: 1.42)	-	4	1
12-14 years	Children	13.3 (*SD*: 0.72)	118.66 (*SD:*8.39)	0	3
	Parents	13.3 (*SD*: 0.72)	-	0	3

4.2.2 Qualitative Focus Groups Results

In the qualitative part of the second phase new FG discussions were conducted. After a brief introduction and explanation of the purpose of the groups, the participants completed the pilot version of the APLES questionnaire. Subsequently, the cognitive debriefing procedure and concept elicitation followed to gain further insight into the participants' overall impression of the questionnaire.

The cognitive debriefing was used to gather a detailed opinion from the respondents regarding the general impression of the questionnaire as well as understanding, clarity and relevance of the items. The relevance refers to the extend the participants thought the questionnaire is reflecting the burden of Achondroplasia and the clarity describes the extend to which the questionnaire appeared to be clear to the group. Concept elicitation was used to identify if any aspects, which the respondents considered important, were missing in the questionnaire.

In general, the qualitative FG analysis showed that the APLES questionnaire was considered as very appropriate, comprehensive and clear for the participants. Consensus was reached in the groups that the questionnaire is very specific for Achondroplasia. However, the analysis also revealed different opinions about the items of each scale, or missing aspects as shown in the following paragraphs.

1. Items of the scale “Self-perception”

Items of the scale “self-perception” were highly valued by parents of children aged 8 to 14 years, since they rated this scale as specific with regard to Achondroplasia. However, some parents of children aged 5 to 7 years mentioned to have difficulties to answer questions concerning their child´s emotions because they do not know the thoughts, emotions and feelings of their child or have not talked about it before. These comments referred to the item “My child likes his/her body” and “My child finds him/herself to be too fat”. Besides, parents of children aged 5 to 7 years expressed to miss detailed questions that assess how the child is coping with everyday life and how independent the child really is. These detailed questions should address various aspects of daily life, like if the child is able to use the toilet independently or is able to perform personal hygiene independently. Thus, the question “My child masters daily life all by him-/herself” was rated as too general to assess these detailed important aspects of everyday life. Furthermore, some parents of children aged 12 to 14 years criticized the scale “self-perception” as too superficial. They would include further questions that address self-confidence and emotions of their children in more detail. In addition, most parents disagreed on the item “My child thinks (s)he is just like others and just look somewhat different”, because they respect their child the way it is and do not and do not want that their child feels different in daily life.

In general children aged 8 to 14 years rated the scale “self-perception” as relevant for their condition. However, adolescents in the age group 12 to 14 years appraised the questions “I find myself to be too fat” and “I like my body” as too direct. Furthermore, they suggests to include a differentiation of the item “I like my body” to be able to state in particular what they like or dislike about their body. In contrast to this, children in the age group 8 to 11 years highly valued the item “I find myself to be too fat”.

2. Items of the scale “Family”

The scale “family” appeared to be relevant for the affected children and their parents with regard to Achondroplasia. However, especially parents disagreed on the items “My child is allowed less than other children that age”, “My child is not treated differently from others in our family” and “My family shows consideration for me”, because these items represent their children as very negative and special, compared to other children or family members. Furthermore, parents of children aged 12 to 14 years voiced that it would be important to differentiate if the term “family” includes only parents and siblings or also other family members like grandparents, because views and attitudes can differ within the family. In addition, parents of children aged 5 to 7 years expressed to miss questions concerning the social environment of the child, e.g. whether the family lives in a rural, suburban or urban area and whether the child has siblings. According to the parents, these environmental factors influence the emotions and actions of the child and thus would be important to consider in the questionnaire.

Children mostly agreed on the items of the scale “family”. However, some children aged 8 to 11 years had difficulties to answer the question “My short stature is a big deal to my family” because they were not able to judge this at this age.

3. Items of the scale “Friends”

Items of the scale “friends” were clear and relevant for the affected children and adolescents and their parents. However, the item “I have problems to find a friend/boyfriend/girlfriend” appeared to be irrelevant for parents of children at this age. Furthermore, some parents of children aged 5 to 7 years voiced to have difficulties to answer some questions of the scale “friends” because they rated their child´s friendships rather superficial at this young age. In addition, the youngest children in the age group 8 to 11 years had difficulties to understand the question “I get no special treatment by my friends”, because they were not familiar with the term “special treatment”.

4. Items of the scale “Recreation”

In general, the affected children and adolescents and their parents rated this scale as very appropriate for their condition and clearly understood the items. However, parents of children aged 5 to 7 years appraised this scale as too general and superficial. They would include further topics, in addition, to the aspect of sports e.g. music. Also parents of children aged 8 to 11 years missed detailed questions regarding leisure activities, school trips, holiday activities and school sports.

5. Items of the scale “”Kindergarten/School”

Overall, this scale was rated as important because it addresses an essential and characterizing setting in the life of young people. However, some parents of children aged 8 to 11 years stated to have difficulties to answer questions of situations in which they are not present or do not know the emotions and feelings of their child e.g. if the child is teased or laughed at, if the child is treated differently in school/kindergarten or if their child’s condition is a big topic school. Moreover, especially parents of younger children aged 5 to 7 years voiced that the items that address these topics were difficult to answer because the child is still too young to perceive strong differences between them and other peers, within the kindergarten/school setting.

6. Items of the scale “Medical”

The participants in the FG discussions of this study rated most items of the scale “medical” as not appropriate for Achondroplasia. Surgeries or frequent doctors appointments were not relevant at this age for the participants in the FG. Furthermore, parents stated difficulties when answering questions that include a subjective perception like “many surgeries” or “quite often”. To be able to evaluate the answer to these questions adequately, they would need further concrete information or a reference value.

7. Items of the scale “Strangers”

This scale was rated as very important for the affected people and their parents. Due to their disproportionate short stature, children and adolescents with Achondroplasia have an affect on their social environment, especially on people that are not familiar with their appearance. However, children and adolescents mentioned that this scale is very detailed and can be shortened to reduce the respondent’s burden.

8. Items of the scale “Aids”

In the FG discussions of this study, items of the scale “aids” were rated as irrelevant and difficult to understand for the affected children and adolescents and their parents.

9. Items of the scale “Physical”

The affected children and adolescents and their parents appraised the scale “physical” as very specific for Achondroplasia, saying it covers what they really experience in daily life. However, the questions “I am often in pain”, “I have/had physical problems with my ears” and “I am not able to reach out to everything” appeared not relevant for children and adolescents with Achondroplasia in the FG discussions of this study. Furthermore, especially parents of younger children mentioned to have problems to answer the questions “My child finds his/her head to be too fat” and “My child likes his/her face”, because they have not talked with their children about these topics yet.

4.2.3 Quantitative Results of the Pilot Test

The pilot test was conducted within the FG discussions. The sample consisted of the same 27 participants as in the FG discussions, including 20 parents and 7 children and adolescents across different ages and gender (see table 12 and 13).

4.2.3.1 Scale Characteristics

The scores achieved on the sub scale of the APLES module and the total score of the APLES module represent an empirical assessment of the child's and adolescent's functioning from their own point of view and from their parents' point of view. To analyze the scores, all scores were transformed from raw scores to 0 to 100 scores with higher values representing higher functioning and higher HrQoL.

As it can be seen in the overall data for children, the scale "strangers" identifies the lowest score with a mean of 42.50 (*SD*: 4.33). Also parents rated their children's HrQoL and functioning in this domain lowest with a mean of 43.68 (*SD*: 4.22). Furthermore, the mean scores of children and parents on the scale "physical" were rather low as well (see table 14).

Compared to this, children scored the domain "medical" highest with a mean of 75.89 (*SD*: 35.24). However, the substantial standard deviation of this scale also indicates the variance across the scale. In the parents' group, the scale "kindergarten/school" was scored highest with a mean of 62.45 (*SD*: 13.62) (see table 14).

Overall parents valued their children's HrQoL and functioning lower than the children did, except for the dimensions "aids" and "strangers". The mean total score for children is 58.79 (*SD*: 8.70), which is slightly higher than the parent's total score with a mean of 53.21 (*SD*: 10.00).

Furthermore, the analysis revealed substantial standard deviations in the child's data set on the scales "friends" (*SD*: 24.71), "recreation" (*SD*: 27.00) and "medical" (*SD*: 35.24). In the parental data set standard deviations were also high on the scales "recreation" (*SD*: 21.97) and "medical" (*SD*: 31.73) as well as on the scale "aids" (*SD*: 21.75). These values represent a high variance in the responses across the scale.

Table 14. Descriptive statistics on scale level for children and adolescents

Domain		**Children**			**Parents**		
	***N* Items**	***N***	**Mean**	***SD***	***N***	**Mean**	***SD***
Self-perception	7	7	57.14	9.88	20	57.23	14.70
Family	7	7	64.37	8.62	20	60.47	9.67
Friends	8	7	67.85	24.71	20	58.05	17.32
Recreation	4	7	56.25	27.00	20	47.18	21.97
Kindergarten/school	6	7	61.30	12.66	20	62.45	13.62
Medical	4	7	75.89	35.24	20	44.37	31.73
Strangers	10	7	42.50	4.33	20	43.68	4.22
Aids	3	7	55.95	20.24	20	61.45	21.75
Physical	10	7	47.85	14.39	20	43.98	9.39
Total Score*	**59**	**7**	**58.79**	**8.70**	**20**	**53.21**	**10.00**

* Sum of all scales

4.2.3.2 Item Correlation

To analyze to what extent each item is associated with Achondroplasia and to select items for the field version, statistical correlation analysis was performed using Pearson's Correlation Coefficient (*r*). Therefore, each item was analyzed in correlation to the variables total score, proportionality and height. Table 15 shows each item of the children's version that was included in the field version and correlates with the children's Total Score (Ctot100), children's height (Cheight) and children's proportionality (Cprop) and each item of the parents' version that correlates with the parents' Total Score (Ptot100), their child's height (Pheight) or their child's proportionality (Pprop). Items that correlated with at least one of these six variables and had a correlation value of $r \geq 0.4$ were considered as relevant for Achondroplasia and thus included in the analysis table. In the analysis table, most items correlate with the children's Total Score (Ctot100) and least items correlate with the parents' Total Score (Ptot100). Overall, most of the items correlate with the children's data than with the parents' Total Score, their child's height or their child's proportionality. However, some correlations in the analysis were not significant ($p \geq 0.05$). Significant values are marked in bold print (see table 15). A detailed overview of the complete analysis with all items is shown in annex 4 and 5.

Table 15. Item correlations (Pearson Correlation Coefficient *r*) with the variables Total Score, height and proportionality with children and parent data

Items	Ctot100	Ptot100	Cheight	Pheight	Cprop	Pprop
My child thinks (s)he is okay the way (s)he is.		*r*=0.42 (p=0.06)				*r*= 0.45 (p=0.10)
My child always draws attention from others because of his/her short stature. .	*r*= 0.74 (p=0.05)				***r*= -0.95** **(p≤0.05)***	
My child can accomplish more than others might think.	*r*= -0.59 (p=0.15)				*r*= 0.47 (p=0.41)	
My child finds him-/herself to be too heavy.	*r*= 0.58 (p=0.16)			***r*= 0.61** **(p≤0.01)****	*r*= -0.43 (P=0.46)	
My child masters daily life all by him-/herself.	*r*= 0.60 (p=0.15)			***r*= 0.77** **(p≤0.01)****	***r*= -0.94** **(p≤0.05)***	***r*= -0.76** **(p≤0.01)****
My child likes his/her body.			***r*= -0.81** **(p≤0.05)***			
My child´s short stature is a big deal for my family.	*r*= -0.73 (p=0.78)				*r*= 0.48 (p=0.51)	*r*= -0.42 (p=0.11)
My child receives help by members of our family in daily life.	*r*= -0.69 (p=0.08)			*r*= -0.48 (p=0.05)	***r*= 0.91** **(p≤0.05)***	***r*= -0.61** **(p≤0.05)***
Because of my child's short stature we have less confidence in him/her.	*r*= 0.50 (p=0.23)			***r*= -0.84** **(p≤0.01)****		***r*= -0.61** **(p≤0.05)***
We treat our child pretty normal.	*r*= 0.50 (p=0.24)		*r*= -0.54 (p=0.26)	*r*= 0.48 (p=0.05)		
My child's friends think his/her short stature is nothing special.	*r*= 0.55 (p=0.19)					
My child is not treated differently from others by his/her friends.	***r*= 0.76** **(p≤0.05)***				*r*= -0.56 (p=0.31)	
My child´s friends are considerate of him/her.	***r*= 0.79** **(p≤0.05)***				*r*= -0.49 (p=0.40)	
My child's friends help him/her.	*r*= 0.72 (p=0.06)				*r*= -0.49 (p=0.40)	
My child's friends stand up for him him/her.	***r*= 0.84** **(p≤0.05)***	***r*= 0.46** **(p≤0.05)***	*r*= 0.51 (p=0.29)		***r*= -0.88** **(p≤0.05)***	
My child's friends protect him/her.	*r*= 0.74 (p=0.05)	*r*= 0.41 (p=0.07)			*r*= -0.41 (p=0.49)	*r*= 0.44 (p=0.10)
My child's friends think that his/her height and appearance is pretty normal.	*r*= 0.42 (p=0.33)		*r*= 0.54 (p=0.26)		*r*= -0.46 (p=0.43)	
My child is able to do the sports, that (s)he wants to.	*r*= 0.71 (p=0.06)				*r*= -0.44 (p=0.45)	
In his/her leisure time my child can do, what (s)he likes.	*r*= 0.66 (p=0.10)				*r*= -0.44 (p=0.45)	*r*= 0.42 (p=0.11)
My child finds clothes, (s)he likes.	***r*= 0.93** **(p≤0.01)****				***r*= -0.98** **(p≤0.01)****	*r*= -0.40 (p=0.13)
Shopping is difficult for my child.			*r*= -0.61 (p=0.19)			
Other kids at school/kindergarten help my child.	*r*= 0.56 (p=0.23)					
My child is treated considerately at school/kindergarten.	*r*= 0.50 (p=0.24)			*r*= 0.41 (p=0.10)		
My child is supported by his/her teachers.			*r*= 0.61 (p=0.19)	*r*= 0.45 (p=0.08)	*r*= -0.58 (p=0.30)	
My child spends too much time at the doctors/therapist.			*r*= 0.66 (p=0.14)	***r*= -0.54** **(p≤0.05)***	*r*= -0.66 (p=0.22)	*r*= 0.43 (p=0.10)

(Table 15 cont.)

Items	**Ctot100**	**Ptot100**	**Cheight**	**Pheight**	**Cprop**	**Pprop**
My child is treated by strangers like others his/her age.	*r*= -0.49 (p= 0.26)		*r*= -0.54 (p=0.26)		***r*= -0.91 (p≤0.05)***	
Strangers often make fun of my child or laugh at him/her behind his/her back.	*r*= 0.54 (p=0.20)				*r*= -0.85 (p=0.06)	*r*= -0.40 (p= 0.13)
My child dares to ask strangers for help.	*r*= -0.75 (p= 0.05)		*r*= -0.44 (p=0.37)		*r*= 0.77 (p=0.12)	
It hurts my child, that (s)he attracts attention by strangers because of his/her height.			*r*= 0.66 (p=0.15)			
The shape and length of my child´s legs causes problems for him/her.	*r*= 0.59 (p=0.15)				*r*= -0.47 (p=0.41)	
It is hard for my child to move long distances.			*r*= -0.56 (p=0.24)			
The shape and length of my child´s arms causes problems for him/her.			*r*= -0.69 (p=0.12)		*r*= -0.41 (p=0.48)	
My child experiences problems with his/her body (e.g. due to bad blood circulation in extremities).	*r*= -0.63 (p=0.12)	***r*= 0.58 (p≤0.01)****			*r*= 0.85 (p=0.06)	
My child finds his/her head to be too big.	*r*= -0.55 (p=0.19)				*r*= 0.43 (p=0.46)	
My child likes his/her face.	*r*= 0.40 (p= 0.36)		*r*= -0.75 (p=0.08)		*r*= -0.44 (p=0.45)	*r*= 0.43 (p=0.10)

* The correlation if significant at a level of 0.05 (2-tailed)
** The correlation if significant at a level of 0.01 (2-tailed)

4.2.4 Item Selection for the Field Test

Quantitative data of the pilot test as well as qualitative data of the FG discussions were reviewed to finally select items for the field test version of the questionnaire. The final decision for retaining or removing items for the field test version was based on the quantitative results of the correlation between each item and the variables Total Score, height or proportionality and on the qualitative results of the FG discussions with the cognitive debriefing. Hence, all items that performed best with regard to quantitative and qualitative results were included in the field test version. Therefore, items that showed a moderate to high correlation with the parents' and children's total score, height or proportionality (*r*≥ 0.4) and were highly valued by the participants in the FG discussions regarding understandability, clarity and relevance were selected for the field test version.

In total 35 items out of 59 items fulfilled this criterion and were included in the field test version of the questionnaire, even though not all correlations between the items and any external criteria were significant[1] (see table 15). But to guarantee an adequate number of items for the field test and to ensure that the remaining scales in the field test version are represented with at least three items, it was necessary to include also items that showed a non-significant correlation with the external variables. Furthermore, these items showed good qualitative results since they were appraised as relevant and clear for the participants in the FG discussions and thus selected for the field test version.

Items that had a correlation value of $r \leq 0.4$ or were irrelevant and difficult to understand for the affected people and their parents were not included in the field test version. These items were considered as not appropriate for the questionnaire and not particularly suitable and relevant for Achondroplasia.

Finally, with regard to the quantitative results of the correlation analysis and the qualitative results of the FG discussions with the cognitive debriefing, the final version of the APLES questionnaire for the field test consists in total of 35 items. Hence, 24 items of the pilot version were rejected. Table 16 shows how many items were rejected in each scale of the pilot version and thus how many items were included in each scale of the field version.

Table 16. Comparison of items in the pilot and field test versions

Scale	**Number of items in pilot version**	**Rejected items (n)**	**Number of items in field version**
Self-perception	7	1	6
Family	7	3	4
Friends	8	1	7
Free time	4	-	5*
Kindergarten/School	6	3	3
Medical	4	3	**
Strangers	10	6	4
Aids	3	3	-
Physical	10	4	6
Total	**59**	**24**	**35**

*Received one item from the scale “medical”

**The remaining item in the scale “medical was moved to the scale “free time”

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[1] This was intended by the working group “Quality of Life” of the Institute for Medical Psychology at the University Hospital Hamburg-Eppendorf who is the responsible leader in the development of the APLES questionnaire in cooperation with the BKMF and the University Hospital Magdeburg.

By rejecting all three items in the scale “aids” and nearly all in the scale “medical” these two scales were completely removed in the field version. The remaining item “My child spends too much time at the doctor/therapist” in the scale “medical” was moved to the scale “recreation”. Consequently, the field version consists of 35 items assigned to seven of the originally nine scales.

According to the results of the cognitive debriefing, some items were reworded to ensure better comprehensibility. For example the item “My child is independent in daily life” was rephrased into “My child masters daily life all by him-/herself” or the item “My child always draws attention to itself because of his/her body” was rephrased into “My child always draws attention from others because of his/her short stature”.

Following the qualitative and quantitative results of the first and second phase of the questionnaire development, the final refined field test version is available in self-report for children and adolescents with Achondroplasia aged 8-11 and 12-14 years (see annex 3 a) as well as for parent report of children with Achondroplasia aged 5-7 years, 8-11 years and 12-14 years (see annex 3 b). These versions will be used for a subsequent field test to psychometrically test the questionnaire and in a following re-test to test the reliability of the instrument.

All items of the field test version can be traced back to the original ICF-CY code they emerged from, since they were all developed based on statements of children, adolescents and young adults with Achondroplasia and their parents within the framework of the ICF-CY. In total five of six items of the APLES scale “self-perception” emerged from the ICF component “body functions” and one item from the ICF component “environmental factors”. On the scale “family” three of four items can be traced back to the component “environmental factors” and one item to the component “activities and participation”. On the scale “friends” and “school/kindergarten” all items were generated of the component “environmental factors” and on the scale “recreation” all items emerged from the component “activity and participation”. On the scale “strangers” two items were generated of the ICF component “environmental factors” and two of the component “body functions”. Four items on the scale “physical” emerged from the ICF component “body structures” and two items emerged from the component “activities and participation” (see table 17).

Table 17. Items of the field test version with the corresponding ICF component

	Item	ICF Component	ICF Code
Self-perception			
1.	I feel okay the way I am.	Body Functions	b126
2.	I always draw attention from others because of my short stature.	Body Functions	b180
3.	I can accomplish more than others might think.	Environmental Factors	e445
4.	I find myself to be too heavy.	Body Functions	b180
5.	I master daily life all by myself.	Body Functions	b120
6.	I like my body.	Body Functions	b180
Family			
7.	My short stature is a big issue in my family.	Environmental Factors	e410
8.	Members of my family help me in daily life.	Environmental Factors	e310
9.	Because of my short stature my family is less confident in me.	Environmental Factors	e310
10.	I am treated pretty normally by my family.	Activities&Participation	d760
Friends			
11.	My friends think my short stature is nothing special.	Environmental Factors	e420
12.	I am not treated differently from others by my friends.	Environmental Factors	e320
13.	My friends are considerate of me.	Environmental Factors	e320
14.	My friends help me.	Environmental Factors	e320
15.	My friends stand up for me.	Environmental Factors	e320
16.	My friends protect me.	Environmental Factors	e320
17.	My friends think that my height and appearance is pretty normal.	Environmental Factors	e420
Recreation			
18.	I am able to do the sports that I want do.	Activities&Participation	d920
19.	In my free time I can do the activities I like.	Activities&Participation	d920
20.	I find clothes that I like.	Activities&Participation	d540
21.	Shopping is difficult for me (e.g. because of not being able to reach things).	Activities&Participation	d620
22.	I spend too much time at the doctor/therapist.	Activities&Participation	d920
Kindergarten/School			
23.	Other kids at school/kindergarten help me.	Environmental Factors	e425
24.	I am treated considerately at school/kindergarten.	Environmental Factors	e425
25.	I am supported by my teachers.	Environmental Factors	e425
Strangers			
26.	I am treated by strangers like other kids my age.	Environmental Factors	e445
27.	Strangers often make fun of me or laugh at me.	Environmental Factors	e445
28	I dare to ask strangers for help.	Body Functions	b126
29.	Attracting attention by strangers because of my height is annoying to me.	Body Functions	b126
Physical			
30.	The shape and length of my legs cause problems for me.	Body Structures	s750
31.	It is hard for me to move long distances.	Activities&Participation	d450
32.	The shape and length of my arms causes problems for me.	Activities&Participation	d445
33.	I experience physical complaints (e.g. my legs falling asleep).	Body Structures	s760
34.	I find my head to be too big.	Body Structures	s710
35.	I like my face.	Body Structures	s710

5. Discussion

This study describes the development of a disease-specific instrument to assess HrQoL, burden of disease and functionality of children and adolescents with Achondroplasia, using qualitative and quantitative methods. The final version of the Achondroplasia Personal Life Experience Scale (APLES) consists of 35 Likert-scaled items assigned to the ICF-CY dimensions: Body functions (global and specific mental functions), body structures (structures related to movement), activities and participation (community, social and civic life/ interpersonal interactions & relationships/ mobility/ self-care/ domestic life) and environmental factors (attitudes/ support & relationships).

The methodological development of the APLES questionnaire was based on guidelines of the U.S. Food and Drug Administration on patient-reported outcome instruments. This approach requires the use of FG discussions to identify relevant concepts associated with HrQoL to ensure that the instrument is applicable to the target population and addresses their topics of interests (Patrick *et al.*, 2007). Therefore, the development process of the APLES questionnaire was guided by the use of FG data of a previous study. Relevant statements associated with HrQoL, burden of disease and functionality of children and adolescents with Achondroplasia were categorized. Afterwards, the statements were used to identify relevant concepts for item generation. This approach was already applied successfully in other projects aiming to develop a HrQoL instrument. For example the European QoLISSY project (The European QoLISSY Group, 2013) and the DISABKIDS project (The Disabkids Group Europe, 2006) used FG discussions to identify patients' needs and interests and used this data material to generate items.

5.1 Result Discussion Phase I

For the current study, the ICF-CY proved to be a useful tool to classify statements in the FG discussions of children, adolescents and young adults with Achondroplasia and their parents to identify aspects of HrQoL, burden of disease and functioning. A total of 125 codes of the ICF-CY across the four main components body functions, body structures, activities and participation, and environmental factors were identified and thus used to classify the statements. These codes reflect the content of the FG discussions and revealed the ICF-CY related domains of interest of children and adolescents with Achondroplasia as well as their parents from their own perspective. Moreover, special impairments and resources of children and adolescents with Achondroplasia assigned to the components of the ICF-CY were identified as shown in the following paragraphs.

Body Structures:

The mutation in the genetic disposition of people with Achondroplasia results in several changes of body structures. Especially body structures that refer to the ICF-CY category “structures related to movement” are affected by these changes and induce medical consequences. Within this category the majority of statements referred to the ICF-CY code s750 (structure of lower extremity), indicating that especially the lower limbs account for physical problems in Achondroplasia. Besides, ICF-CY codes that referred to structures of the trunk (s760), structures of the head and neck region (s710), structures of the upper extremity (s730) and eye and ear related structures (s299) were also often identified in the FG analysis. These ICF-CY codes are the areas where most physical problems may arise in relation to Achondroplasia and emphasize the effect of Achondroplasia across the lifespan of the affected individuals. Hence, physical problems often affect the whole body of individuals with Achondroplasia and influence their HrQoL and functioning negatively. This is confirmed by Ireland & Pacey *et al.* (2014), who described that a complexity of medical issues is associated with the disproportionate short stature in Achondroplasia, including cardiorespiratory, neurological and musculoskeletal impairments as well as nose, ear and throat complications and impairments.

Body Functions:

Most frequently addressed areas within the component “body functions” referred to the ICF-CY categories “mental functions” and “sensory functions and pain” representing the impact of the altered body structures on body functions in people with Achondroplasia. The majority of identified statements of this component was classified with the code b180 (experience of self and time functions) of the category “mental functions”. This emphasizes that mental functions and especially body image and experience of self are of high interest for the affected persons and have a high influence on their HrQoL, burden of disease and functioning. This is in line with the results of a study by Haraldstad *et al.* (2011) who revealed that the perceived body image is a strong predictor of HrQoL in children and adolescents. In addition, the ICF-CY code b280 (pain in body part) was also often addressed. Children, adolescents and young adults with Achondroplasia mentioned experiencing pain in various body parts, but mainly in lower limbs, back and joints. This is in concordance with other studies which described that individuals with Achondroplasia often suffer from pain due to the skeletal abnormalities (Baujat *et al.*, 2008; Richette *et al.*, 2008). Furthermore, it is noticeable that the affected people did not address neuromusculoskeletal and movement-related functions within the component “body functions” in the FG discussions. This may indicate that mental functions are more relevant for the affected persons than physical aspects.

Activities and Participation:

Most frequently addressed areas within the component “activities and participation” were “community, social and civic life”, “self-care”, “mobility”, “domestic life”, “general tasks and demands” and “interpersonal interactions and relationships” representing the direct impact of Achondroplasia on the life of the patients. Limitations in these areas are mainly associated with changes in body structures and functions of people with Achondroplasia. Especially the disproportionate short statured body has a direct impact on major areas of activities and participation.

In detail, limitations refer to the ICF-CY code d920 (recreation and leisure), which was often addressed in the FG discussions. Children and adolescents with Achondroplasia are often restricted in their leisure activities or sports and cannot do the same things as their friends or peers do, because of their condition. Besides, restrictions in the area of self-care were often mentioned with regard to the ICF-CY-codes d540 (dressing) and d530 (toileting). Also Ireland *et al.* (2011) described that clinicians and parents often report limitations in children with Achondroplasia, especially concerning self-care (e.g. d530 toileting, d540 dressing) and mobility (e.g. d450 walking, d455 moving around). In the analyzed FG discussions of this study, the ICF-CY category “mobility” was also frequently addressed and associated with restrictions and limitations in using public motorized transportation, walking, running, climbing or swimming due to the special condition of people with Achondroplasia. Furthermore, limitations in areas of domestic life were mainly reported with regard to the ICF-CY codes d620 (shopping), d630 (preparing meals) and d640 (helping to do housework). In addition, the ICF-CY category “interpersonal interactions and relationships” was often mentioned in the FG discussions. Especially family relationships were highly valued, which suggests that the family has a positive influence on HrQoL, wellbeing and functioning of children and adolescents with Achondroplasia.

Within the component “activities and participation”, the ICF-CY code d240 (handling stress and other psychological demands) was addressed most often in the FG discussions, which can be explained by a stronger emotional impact of a chronic condition on children. Especially in Achondroplasia, altered body structures and the related limitations in the areas of activities and participation result in greater psychological demands for children and adolescents with Achondroplasia due to their special condition. To deal with the increased psychological demand, the affected persons developed various coping strategies. This is in line with the result of Quitmann *et al.* (2014) who also described that many individuals with Achondroplasia show coping strategies to better deal with their condition.

Environmental Factors:

The change in body structures in people with Achondroplasia also affects the social and physical environment of the affected people and their parents. Within the component “environmental factors”, the ICF-CY categories “support and relationships” and “attitudes” were frequently addressed in the FG discussions. In detail, most statements were assigned to the code e410 (individual attitude of immediate family members). Children and adolescents with Achondroplasia valued especially the support and attitude of immediate family members and friends highly positive. Hence, this aspect is very important for the affected persons and has a positive influence on their HrQoL, wellbeing and functioning if they are treated in a normal and adequate way by their parents and are accepted by them the way they are. In contrast to this, negative influences on HrQoL, wellbeing and functioning were associated with experiences with strangers or peers because they often tease or stare at the affected people. Also Gollust *et al.* (2003) described that children with Achondroplasia are exposed to a higher risk to experience bullying or teasing in daily life.

Furthermore, the code e580 (Health services) was often addressed in the FG analysis. This code is associated with an increased medical treatment that children and adolescents with Achondroplasia need to deal with, since they need to undergo surgery more often than normal statured people, to prevent or to treat medical complications. This confirms with Baujat *et al.* (2008) who described that individuals with Achondroplasia suffer from complex medical issues that need to be prevented and treated with optimal medical care in a multidisciplinary team. Moreover, the category “products and technology” was frequently mentioned, which emphasizes the dependency on assistive products for people with Achondroplasia. They often need products that should help to facilitate their daily life. Therefore, the ICF-CY codes e115 (general/assistive products and technology for personal use in daily living), e1201 (assistive products and technology for personal indoor and outdoor mobility transportation) and e1351 (assistive products and technology for employment) were detected in the FG discussion. Moreover, reconstructions in the house are often necessary to facilitate daily life for the affected people and to enable them to participate in domestic life.

The FG analysis based on the ICF-CY also illustrates that the components “environmental factors” and “activities and participation” were addressed most often, which emphasizes the relevance of these topics on Achondroplasia. However, statements that were assigned to the code b180 (experience of self and time functions) of the component “body functions” and s750 (structures of lower extremity) of the component “body structures” were also frequently identified in the analysis, which indicates that these topics account for a high relevance for the affected persons and their parents with regard to their HrQoL, burden of disease and functioning as well.

Besides the reflection of ICF-CY related domains of interest in Achondroplasia from the children’s and parents’ perspective, the results of the FG analysis served as a broad construct for the APLES questionnaire and were essential for item generation. Items of the pilot version of the APLES questionnaire were developed based on 30 ICF-CY codes of the category system and the associated statements that represent the interests of the affected children and adolescents as well as their parents’ perception on their children’s HrQoL, burden of disease and functioning. In detail, the items emerged from eleven codes of the ICF component “activities and participation”, from eleven codes of the component “environmental factors”, from four codes of the ICF component “body functions” and from four codes of the component “body structures”.

Most items of the pilot version are based on the component “environmental factors”. In total 27 items emerged from this component assigned to the ICF-CY categories “attitudes” of immediate family members, strangers, friends and peers and colleagues (e410, e445, e420. e425, e450), “support and relationships” of immediate family and friends (e310, e320), “products and technology” (e115, e155, e120) and “services, systems and policies” (e580).

In the component “activities and participation”, 18 items of the APLES questionnaire are assigned to the ICF-CY categories “general tasks and demands” (d240), “community, social and civic life” (d920), “interpersonal interactions and relationships” (d760, d730, d770), “mobility” (d445, d455, d450), “self-care” (d540) and “domestic life” (d620). These ICF-CY categories reflect major areas of life that are considerably affected by Achondroplasia and the associated disproportionate short stature.

Nine items assigned to the ICF-CY categories “mental functions” (b180, b126) and “sensory functions and pain” (b280) are based on the component “body functions”.

Least items of the APLES questionnaire are based on the component “body structures”. In total five items emerged from this component assigned to ICF-CY categories “structures related to movement” (s750, s710, s760) and “the eye and ear related structures” (s299). These ICF-CY codes reflect the medical impairments and complications that are special for this condition and thus relevant when assessing HrQoL, burden of disease and functioning in this patient group.

Altogether, 59 items assigned to nine scales were used for pilot testing. Integrating the ICF-CY in the development process of the APLES questionnaire assured that each item is directly linked to a specific code of the ICF-CY. This provides information for clinicians or researchers which areas of the ICF-CY are represented in the APLES questionnaire and transfers the content into the internationally used classification of the ICF. Across all scales of the APLES questionnaire items are mostly represented by the ICF-CY components “environmental factors” and “activities and participation”. Hence, the ICF-CY component “body functions” and “body structures” were less represented. This emphasizes that HrQoL, functioning and wellbeing is also associated with psychosocial concepts apart from physical aspects and underlines the importance of a broad perspective on health.

5.2 Result Discussion Phase II

The results of the cognitive debriefing proved that the identified ICF-CY related domains of interests of the first FG analysis and the generated items correspond with the interests of the participants of the new FG discussions and thus provide evidence for the selected items of the pilot version. Overall the APLES questionnaire was valued positive by the affected children and adolescents and their parents as well as by parents of younger children aged 5 to 7 years. However, the qualitative results showed as well, that especially the scale “family”, “kindergarten/school”, “medical”, “strangers” and “aids” needed to be revised. Consequently the scales “medical” and “aids” were completely removed in the field test version and the remaining scales were reduced regarding the number of items. In addition, the results of the cognitive debriefing showed that sometimes formulation of the items caused misunderstandings and led to some difficulties during questionnaire completion. Furthermore, especially parents mentioned problems to answer questions concerning their child’s emotions because they cannot directly observe the emotions of their child. The results of the concept elicitation revealed some additional aspects that were important for parents of children with Achondroplasia. From the point of view of the parents coping, leisure time activities like music, arts and handicrafts as well as the environment in which the child lives have an impact on their children’s HrQoL, wellbeing and functioning and should be considered in a condition specific questionnaire for Achondroplasia. However, from the parents’ point of view these topics are not addressed in sufficient detail in the pilot version of the questionnaire. This underlines, that recreation and leisure are important topics for individuals with Achondroplasia because they are often restricted in these areas due to changes in body structures that are associated with the disproportionate short stature. The knowledge of challenges in the area of recreation and leisure activities supports the need for counseling and to introduce children and adolescents with advices how to organize their leisure activities best and what leisure activities are appropriate for Achondroplasia. Consequently, detailed questions concerning leisure time should be included in a condition specific questionnaire in order to identify adequately the needs and resources of individuals with Achondroplasia in the area of activities and participation.

Furthermore, the quantitative results of the pilot test provide preliminary evidence of the impact of Achondroplasia on the child's HrQoL, wellbeing and functioning from the children's and parents' point of view. Children and parents rated especially the scale "strangers" of the APLES questionnaire lowest. This indicates low functioning and HrQoL in terms of feelings of acceptance by strangers and limitations in social integration. Besides, the scale "physical" was rated low as well by children and parents, which suggests that children and adolescents with Achondroplasia experience low physical functioning and physical limitations in daily life, which have a negative impact on the child's wellbeing. This is in line with the results of Rohenkohl *et al.* (2015), who demonstrated that children and adolescents with Achondroplasia are especially impaired in the domains of physical and social life. Positively valued by children and their parents were the scales "friends" and "family". This indicates that support and relationships from friends and family members are very relevant for individuals with Achondroplasia in daily life. Hence, a stable family environment and true friendships have a highly positive influence on the child's HrQoL, functioning and wellbeing. Furthermore, the results showed that parents perceive their children's HrQoL, functioning and wellbeing on the scale "kindergarten/school" as the highest ranking. Also children themselves rated this scale rather high. To comprehend this score it would be useful to know the type of school the children visit and the support they receive at school because the disproportionate short statured body leads to physical mobility challenges and the need for special equipment at school. Unfortunately, it was not assessed if the children visit a special-needs school where they are often more supported by teachers or assisted by using special equipment, or a regular school where special equipment to facilitate school life for individuals with Achondroplasia is often not available. According to Haga & Kosaki *et al.* (2013), remodeling of the toilet, sink and chair at school is necessary to support children and adolescents with Achondroplasia in school. Furthermore, they often need assistance during swimming lessons and are restricted in physical education lessons. Consequently, the type of school and environment as well as the support the child receives in school have an influence on the child's experiences within the kindergarten/school setting. Hence, the scores of the scale "kindergarten/school" of the current study reflect that children and adolescents experience their school environment as acceptable and are supported in school. However, still the scores are not very high which suggests for improvements within the educational setting. Ireland *et al.* (2014) recommended to include physiotherapists or occupational therapists to assist the school staff in an adequate modification of facilities (e.g. remodeling of toilet, washbasin, chair), problem solving and equipment prescription.

Furthermore, noticeable differences between children's and parents' scores occur in the scales "friends", "recreation" and "medical". In all three scales children rated their HrQoL, functioning and wellbeing higher than their parents. Hence, children value the support of friends higher than their parents and highly appreciate their friends. The results of a study by Quitmann *et al.* (2014) also highlight the positive importance of friends with regard to HrQoL of children and adolescents with Achondroplasia. On the scale "recreation", parents perceive that their child is a lot more restricted in their recreational activities like sports and shopping than their children themselves. Regarding medical aspects, children do not perceive their medical condition as very serious, since they rated the scale "medical" highest. In contrast to this, parents rated this scale very low. The high discrepancy between the children's and parents' perception of medical aspects appears to be justified by the fact that parents focus more on medical problems and perceive doctors' appointments, hospital stays and surgeries often as more intense than their children.

With regard to the Total Score, the children's total score is higher with a mean of 58.79 (*SD*: 8.70) than the total score of parents with a mean of 53.21 (*SD*: 10.00). This difference suggests that children with Achondroplasia perceive their HrQoL, functioning and wellbeing in general slightly higher than their parents do. This is in concordance with the results of a study by Erling & Wiklund *et al.* (1994), who demonstrated that parents of prepubertal children with short stature evaluate their child's wellbeing lower than the children themselves. Qualitative focus group results of a study by Quitmann *et al.* (2014) also revealed strong differences between the perception of children with Achondroplasia on their HrQoL and the perception of parents on their children's HrQoL.

Besides, the correlation analysis revealed additional evidence of the impact of Achondroplasia on patients' lives. A reduced height correlates highly negative with the acceptance of the own body (Item: I like my body; r=-0.81, p≤0.05). Hence, children and adolescents are less likely to accept their body if their height is strongly reduced. Moreover, children's height correlates highly negative with the parents' confidence in their child (Item: Because of my child's short stature we dare him/her less; r=-0.84, p≤0.01), which suggests that a reduced height in their children can induce parents to overprotect their child, since they dare him/her less. Sandberg and Voss (2002) also described that parents tend to overprotect short statured children more frequently than normal statured children. Furthermore, finding clothes the child likes correlates highly negative with the children's proportionality (Item: I find clothes I like; r=-0.98, p≤0.01). Hence, for children and adolescents with Achondroplasia it is very difficult to find clothes they like if their body is very disproportionate. This item also highly correlates with the children's total score (r=0.93, p≤0.01), which suggests that finding appropriate clothes is important for young people with Achondroplasia with regard to their HrQoL and wellbeing. Moreover, parents and children data show a high negative correlation between the proportionality of the body and the independence in daily life (Item: My child is independent in daily life; parents r=-0.76, p≤0.01; children r=-0.94, p≤0.05). Hence, children and adolescents with Achondroplasia are less independent in daily life. This is supported by Ireland *et al.* (2011), who noted that especially the too short upper limbs hamper younger children to perform personal hygiene themselves. Furthermore, the strongly reduced longitudinal growth complicates everyday life of the affected people, mainly in the area of self-care. Regarding the correlations between the items and the total score, the item "My friends stand up for me" highly correlates with the children's total score (r= 0.84, p≤0.05). This emphasizes again the importance of the support of friends for the affected children and adolescents and suggests that support by friends has a high positive impact on HrQoL, wellbeing and functioning in children and adolescents with Achondroplasia. In contrast to this, the item "My child experiences problems with his/her body (e.g. due to bad blood circulation in extremities" shows a high correlation (r= 0.58, p≤0.01) with the parents' total score. This suggests that parents rate the influence of physical aspects on their children's HrQoL higher than social aspects like friends.

The preliminary quantitative results of the pilot test and qualitative results of the second focus group discussions with a cognitive debriefing induced to revise the item list with regard to number of items and formulation of the items. Based on qualitative and quantitative results, the item list was strongly reduced. Hence, items that were not suitable and relevant for Achondroplasia and would influence the criterion validity of the questionnaire negatively were removed. Furthermore, it was necessary to reduce the item list to lower the burden for the affected people during questionnaire completing, to counteract respondent fatigue and to ease evaluation time in order to make the questionnaire more applicable in medical practice. Consequently, the field test version consists of seven scales with 35 items instead of nine scales with 59 items used for pilot testing. In total 16 items in the field test version were generated of ICF-CY codes that are counted among the component “environmental factors”, eight items are based on the component “activities and participation”, seven items are based on “body functions” and four items are based on the component “body structures”. The knowledge of the ICF-CY components and categories that are represented in the APLES questionnaire is very useful for clinicians and researchers and offers the opportunity to make it comparable to other HrQoL measures.

5.3 Comparison of the APLES Questionnaire to Other HrQoL Measures

In clinical practice or in research it is essential to select the most suitable HrQoL instrument for a specific goal. Therefore it is relevant to examine the content of different HrQoL measures in a standardized manner (Riva & Bullinger *et al.*, 2010). The concept of HrQoL and the ICF are often used simultaneously in research or clinical practice because they represent two different perspectives to assess functioning and health. Hence, the ICF provides a universal standardized framework to compare the content of different HrQoL measures. By linking items of a HrQoL instrument to a specific ICF category it is possible to assess and compare the representation of the four main ICF components body structures, body functions, activities and participation and environmental factors and to identify the focus of an instrument on the different components of the ICF (Cieza & Stucki, 2005). Since the APLES questionnaire was developed within the framework of the ICF-CY and is based on the ICF-CY an additional linking of the items to the categories of the ICF is not necessary anymore. This makes the APLES unique and differentiates it from other HrQoL measures.

Already various studies compared the content of a HrQoL measure to the ICF. In a study by Sommer & Bullinger *et al.* (2014), items of the short stature specific QoLISSY questionnaire were linked to the ICF-CY. The results of the study demonstrate that items of the QoLISSY questionnaire are mainly represented by the component “environmental factors” and “activities and participation”, followed by the component “body functions”. This is in concordance with the ICF-CY categories represented by the items of the APLES questionnaire. A closer look at the detailed ICF-CY categories represented in the QoLISSY and in the APLES questionnaire revealed that both questionnaires address predominantly similar ICF-CY categories.

However, the most striking difference between both questionnaires refers to the component “body structures”. According to Sommer *et al.* (2014), no item of the QoLISSY questionnaire is based on this component, whereas this component is represented in four items of the APLES field version with the ICF-CY category “structures related to movements”. In detail the items are based on the ICF-CY codes s710 (structure head and neck region), s750 (structure of the lower extremity) and s760 (structure of trunk), which are the most affected body structures in Achondroplasia. These ICF-CY codes reflect multiple impairments and changes of body structures that are special in individuals with Achondroplasia, compared to individuals with short stature that is caused by dysfunctions in the endocrine system (e.g. ISS or GHD). According to Baujat *et al.* (2008) and Ireland *et al.* (2014), changes in structures of the lower extremity in Achondroplasia refer to a genu varum, which is often associated with leg pain. Changes in structures of the head and neck region refer to an enlarged head with typical facial characteristics in Achondroplasia like a depressed nasal bridge and a prominent forehead. Furthermore, changes in body structures of the trunk are associated with an excessive lumbar hyperlordosis or a narrowed spinal canal, which can cause back pain and neurological complications in the lower limbs. Moreover, individuals with Achondroplasia often suffer from a thoracolumbar kyphosis, which is a major musculoskeletal impairment related to the body structures of the trunk. In addition to the lumbar hyperlordosis this may result in a narrowing of the spinal canal and thus can contribute to spinal damage.

The significant difference between both questionnaires emphasizes the need for the condition-specific APLES questionnaire. This questionnaire specifically addresses the ICF-CY component “body structures”, which covers the characteristic medical complications that people with Achondroplasia experience in their lives. Furthermore, it underlines that body structures are a relevant topic for individuals with Achondroplasia because this component addresses the unique aspect of disproportionality that is associated with Achondroplasia. Therefore, this concept needs to be considered when assessing HrQoL, burden of disease and functioning in this patient group. Consequently, it is important to mention that although the QoLISSY questionnaire is a validated tool to assess HrQoL in Achondroplasia (Rohenkohl *et al.*, 2014), it is only applicable to children and adolescents with Achondroplasia to some extent, since it covers not all domains relevant for Achondroplasia as shown in this study. This confirms that it is problematic to use HrQoL measures that were originally designed for patients with proportionate short stature (e.g. QoLISSY questionnaire) for patients with a disproportionate body like in Achondroplasia or in other forms of skeletal dysplasia (Gollust *et al.*, 2003).

Besides the QoLISSY questionnaire, other HrQoL instruments do not cover the component “body structures” as well. In a study by Cieza and Stucki (2005) six different HrQoL measures (Sf-36, NHP, Ql-I, WHOQOL-BREF, WHODASII and EQ-5D) were linked to the ICF. The results showed that most items were linked to categories of the component “activities and participation” followed by the component “body functions” and “environmental factors”. No items referred to the component “body structures”. Petersson & Simeonsson *et al.* (2013) also examined the content of six different HrQoL instruments specific for children (ChQ-CF, DCGM-37, EQ-5D-Y, KIDSCREEN-52, Kid-KINDL and PedsQL) to the ICF-CY. The linking also revealed that no concept of the questionnaires was linked to the component “body structures”. Furthermore, the results showed that the ICF category “mental functions” of the component “body functions” was referenced most frequently in all reviewed HrQoL measures for children. Especially the code b180 (temperament and personality functions) and b126 (experience of self and time functions) were represented most in the reviewed HrQoL measures. This concurs with the results of the current study, since the component “body functions” is also only represented by the category “mental functions” (b126, b180). Hence, body functions were only associated with mental functions and not with physical body functions like neuromusculoskeletal and movement related functions.

In a study by Karsuska & Riva *et al.* (2012) the hemophilia-specific HrQoL questionnaire for children and adolescents (Haemo-QoL) was also linked to the ICF-CY. The results showed, that the ICF-CY code e410 (individual attitude of immediate family members) was represented as the most important of the component “environmental factors”. This agrees with the results of the FG analysis of the current study, because the code e410 was addressed most frequently in the discussions. The agreement suggests that especially in chronic health conditions, like Achondroplasia, the attitude of the family plays an important role for the affected people with regard to their HrQoL.

The results of studies that linked the ICF to other HrQoL measures emphasize that HrQoL measures are in general mainly represented by the ICF components “activities and participation”, “body functions” and “environmental factors”. Within the APLES questionnaire all four components of the ICF are represented which distinguish this questionnaire from other HrQoL measures. Furthermore, it demonstrates the importance of body structures and indicates that besides “body functions”, “activities and participation” and “environmental factors” the ICF component “body structures” has an important impact on HrQoL, wellbeing and functioning for people with Achondroplasia. Consequently the APLES questionnaire provides an overall picture of health, since all four components of the ICF are demonstrated. However, following the results of the FG analysis, which was used for item generation, the focus of this questionnaire lies more on psychosocial aspects as described by the components “activities and participation” and “environmental factors” and less on physical aspects that are represented by the component “body structures”. Also the domain “body functions” is less represented in the APLES questionnaire and refers only to mental functions and not to physical functions.

This study provides a preliminary version of the first condition specific instrument to assess HrQoL, burden of disease and functioning of children and adolescents with Achondroplasia. However, some methodological limitations in the development need to be considered.

5.4 Methodological Limitations

First of all it needs to be considered that this study is part of a larger project. Therefore, the methodological process of the questionnaire development was predetermined and thus partially limited. This was especially the case during the decision-making of which items were to be included in the refined field test version. The choice which items were included in the field test version was based on quantitative data of the correlation analysis and on qualitative data of the FG discussions with the cognitive debriefing. Each item was analyzed as to whether it correlates with the selected external criteria proportionality, height and Total Score. These selected criteria are relevant variables for the symptoms of Achondroplasia. Hence, the higher the correlation between the items and any external variable was, the more relevant was the item for the condition and thus the questionnaire. Therefore, a limit value of $r \geq 0.4$ was chosen to select the items that showed a moderate to high correlation with the external variables. The items that fulfilled this criterion and were positively valued in the FG discussions were selected for the field test version. Hence, also some items that showed a non-significant correlation with the external variables were included in the field test version. This was necessary to ensure that each scale is represented in the field test version, including at least three items. Moreover, it is expected that after analysis of the field test even more items will be excluded. Therefore, the field test version should consist of an appropriate number of items to provide the opportunity to further reject items. However, the small sample size of the pilot test needs to be considered, which can have an influence on the significance of the correlations. Therefore, the field test version will be tested in a subsequent field and re-test in a larger sample and will hopefully show reliable psychometric properties. Furthermore, using this mixed method approach to select items for the field test version also required to exclude items that showed acceptable quantitative results. However, based on the qualitative focus group results of this study, these items needed to be excluded in the field test version, in order to include the opinions and views of the target population in the development process of the instrument. Still, the FG discussions were conducted with a small and very selective sample, which limits the results of the cognitive debriefing and the correlation analysis. However, not all suggestions of the participants in the FG discussions were included to revise the pilot version. Otherwise, it would have been necessary to include further items that address recreation activities, independence in daily life and the environment the child lives in, in more detail.

Another limitation refers to the sample used in the first as well as in the second phase of the instrument development. In the first phase, FG data of a previous study was used for analysis and item generation. Hence, the items are based on the content of these specific FG. During all analyzed FG discussions the moderator asked structured questions about the life of the patients. Consequently, the answers of the participants in the FG discussions were based on these questions and leave little space for further topics concerning the life of the affected people and their parents. Hence, the structured questions might have influenced the content of the APLES questionnaire. Furthermore, the sample in the FG discussions consisted of children, adolescents and young adults with Achondroplasia across the age groups 8-12 years, 13-18 years and 19-28 years. Within the sample, the age group 8-12 years was represented with less participants compared to the other age groups. Hence, the statements identified in the FG analysis referred predominately to affected persons aged 13-28 years. This age group does not correspond with the age groups 5-7 years, 8-11 years and 12-14 years the APLES questionnaire is designed for. However, the pilot test and cognitive debriefing revealed support of the conceptual framework, which suggests that the age differences between both samples were less relevant then assumed. Nevertheless, it is important to consider that all patients in the sample of the previous FG discussions were recruited from a self-help organization (German Federal Association for Short Statured People and their Families). Hence, the sample is very selective and represents a homogeneous group within all patients with Achondroplasia. Patients and families who joined a self-help group already deal actively with their disease and receive support from other members or professionals. Therefore, this sample is not representative for the overall target population.

Also the sample used for the pilot test and new FG discussions was recruited over a self-help group. Hence, the results of the pilot test and FG discussions are not representative for the target population either. Moreover, the sample size was very small; especially children and adolescents in the age groups 8-11 and 12-14 years were underrepresented in the sample. This allows only a cautious interpretation of the quantitative results and no generalizations of the results. Besides, the small sample size does not allow analyzing gender or age differences in this study. However, children and adolescents undergo rapid development and are continually changing, which also influences how they experience their condition (Grootenhuis & Koopman *et al.*, 2007). The beginning of puberty in the age group 12-14 years is often associated with other experiences and problems than the ones children in the age group 8-11 years experience in their life. Therefore, it would be important to differentiate the results of age and gender groups to identify problems, needs and resources in the particular age group and to really understand gender specific experiences and problems associated with Achondroplasia of children and adolescents. Also the parental data includes data of parents with children aged 5-14 years of both genders. Using this broad age group inhibits to draw reliable conclusions because parents of younger children experience problems of their child related to Achondroplasia differently than parents of adolescents. This is due to the fact that younger children are more dependent on their parents or on other people who support them in daily life (Ireland *et al.*, 2011). Hence, it needs to be considered that data of the children includes the age groups 8-11 years and 12-14 years across both genders and parental data includes data of parents with children aged 5-7 years, 8-11 years and 12-14 years across both genders when interpreting the results of the pilot test.

Furthermore, only one version of the questionnaire for children and adolescents with Achondroplasia was developed. Hence, the questionnaire does not consider limitations of Achondroplasia that are age specific. Especially younger children up to 7 years require greater assistance with regard to self-care and mobility in daily life, due to their too short upper limbs that restrict them to perform hygiene and dressing alone (Ireland *et al.*, 2011). Furthermore, regarding the results of the cognitive debriefing some parents mentioned that a few topics that are addressed with the items of the APLES questionnaire are yet not relevant for younger children (e.g. shopping, finding clothes the child likes). Consequently, to assess HrQoL, wellbeing and functioning of children and adolescents with Achondroplasia adequately, these age specific aspects need to be considered and assessed in more detail in each age group. Therefore, it would be useful to develop different versions of the questionnaire that consider age specific aspects in a following study. However, this would also complicate the evaluation process of the questionnaire.

Overall, the described results of the current study’s pilot test are limited in their generalizability due to the sample characteristics. In order to identify age and gender-specific aspects in the perception of HrQoL, wellbeing and functioning of children/adolescents with Achondroplasia and parents of children/adolescents with Achondroplasia and to draw reliable conclusions the questionnaire should be applied in a larger and more heterogeneous sample. Besides, data was assessed in a cross-sectional design, which limits the possibility to draw conclusions over different time periods and only assesses the current state of HrQoL, wellbeing and functioning of the affected people. In addition to this, the length of the pilot questionnaire (59 items) might have increased the burden for the respondents, especially for younger children, which could have falsified the results of the pilot test. Furthermore, the current study provides no information on the reliability and validity of the APLES questionnaire, which should be considered when interpreting the results. Therefore, a follow-up study with a field and retest is necessary to test the psychometric properties of the instrument. The items in the questionnaire should be tested in terms of distribution characteristics (mean, *SD*, skewness and kurtosis), score distribution and reliability of the scores (internal consistency, retest, intraclass correlation coefficients). Criterion validity should be tested with correlations to other HrQoL measures specific for chronic conditions in children. These analyses are relevant to prove the APLES instrument as a valid and reliable instrument to assess HrQoL, burden of disease and functioning of children and adolescents with Achondroplasia.

6. Conclusion and Outlook

Assessment of HrQoL from the patient perspective gained increasing recognition. Especially the impact of chronic conditions is increasingly studied to better understand the burden of specific disorders. Apart from generic instruments to assess HrQoL condition-specific instruments are needed to assess HrQoL adequately. So far no instrument is available to assess HrQoL, functioning and wellbeing of children and adolescents with Achondroplasia. Although the short stature specific QoLISSY questionnaire is a validated instrument to assess HrQoL of young patients with Achondroplasia it does not take sufficient account of the special situation, specific burdens, restrictions and resources of children and adolescents with Achondroplasia, especially with regard to body structures. To close this gap, this study aimed to develop a new instrument for this patient group. The new instrument “APLES - The Achondroplasia Personal Life Experience Scale“ assesses HrQoL, burden of disease and functionality of children and adolescents with Achondroplasia, considering the patient and parent perspective.

The questionnaire was developed based on statements of previous FG material, which were classified in the ICF-CY. Using FG data helped to gain further insight in patients’ needs and interests. In general, environmental factors like support and relationships with friends and family and assistive products were associated with resources for the affected children and adolescents, whereas changes in body structures were often associated with psychological stressors and pain and led to restrictions in major areas of “activities and participation” including mobility, community, social and civic life, self-care, domestic life and interpersonal interactions and relationships. Each item of the APLES questionnaire is based on a specific code of the ICF-CY. Thus, when using the questionnaire, all results or rather answers can be traced back to the category system outlined by the ICF-CY. By this, it is possible to translate the answers into the international language of the ICF to make them internationally comparable and comprehensible. Furthermore, the APLES questionnaire contains all components of the ICF-CY, namely body functions, body structures, activities and participation and environmental factors. Hence, when applying the questionnaire it provides an overall picture of the patient’s condition.

The questionnaire is designed as a patient-reported outcome instrument and thus has the advantage to identify patient outcomes directly from the patients’ perspective without interpretation of someone else. This gives children and parents a voice in health care and describes the impact of Achondroplasia on the child`s wellbeing from the patients’ perspective. Furthermore, the questionnaire contains the recommendations of the WHO for measurements of quality of life in children (WHO - Division of Mental Health, 1994). It is child centered and offers the opportunity for self-report of children and adolescents with Achondroplasia in two different age groups. Especially the environment of the children and adolescents is considered in the questionnaire, since kindergarten/school, sport, friends and family aspects are addressed. In addition, the questionnaire emphasizes positive aspects on health like the support of friends rather than negative aspects. Moreover, the questionnaire is based on the international language of the ICF-CY, which offers the opportunity to apply the questionnaire in other countries and cultures and to examine the cross-cultural performance. Finally, identified ICF-CY related domains of interests in the FG discussions can serve as a first basis towards the approach to develop an ICF core set, since so far no ICF core set for Achondroplasia exists.

Applying the APLES questionnaire in research or in medical practice can help to better understand HrQoL, wellbeing and functioning of children and adolescents with Achondroplasia, since so far little is known about it. Health professionals and doctors but also teachers can use the questionnaire to better understand the burden of the disease and wellbeing of children and adolescents with Achondroplasia and to assess their individual needs, stressors and resources. Hence, this information can help to optimize health care in this patient group and to design care more individually in order to fulfill the needs of the patients. The results show, that besides physical aspects of the disease psychosocial issues are relevant as well. Therefore, clinical care should be expanded towards the integration of psychosocial aspects, especially with regard to social and psychosocial support.

This study describes only the development process of the first condition-specific patient reported outcome questionnaire for children and adolescents with Achondroplasia. Furthermore, it contains results of the pilot test, which results in a refined version of the questionnaire that will be used in a subsequent field- and retest. Within a follow-up study a field and retest should be conducted to psychometrically test the new instrument. The field test should be conducted in a larger sample with 50 children/adolescents (8-14 years) with Achondroplasia and 75 parents of children with Achondroplasia aged 5-14 years. Besides the field version of the APLES questionnaire, all participants should fill out the disease specific QoLISSY questionnaire, the generic KIDSCREEN questionnaire the chronic-generic DISABKIDS questionnaire and the EQ-5D-Y for validation purpose. Two weeks after the field test, the retest should be conducted to test the reliability of the instrument. The retest should be conducted with a minimum of 10 patients per age group, using the same questionnaires as in the field test. The larger sample size will also allow to draw age and gender specific conclusions regarding the children’s and adolescents’ HrQoL, wellbeing and functioning. After data analysis of the field- and retest there will be a psychometrically pre-tested, practically feasible and conceptually appropriate instrument available in the age group 8-14 for patient report and parent report and only parent report for children from 5-7 years.

The APLES can be used both on a group and individual level. Individual and group diagnostics include application of the questionnaire to single children or patient groups for diagnostic, predictive or evaluative purposes. The uncomplicated application of the new instrument is very practical and facilitates the use in medical practice in order to identify needs and resources of children and adolescents with Achondroplasia. Moreover, it can be used in further studies on Achondroplasia to assess pediatric HrQoL outcomes in clinical trials or health economic research. Furthermore, it could be used as a tool to assess HrQoL, functionality and burden of disease in children and adolescents with other forms of skeletal dysplasia, since all forms of skeletal dysplasia are based on disorders in the bone development.

7. Literature

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Annex

Annex 1: Table 18. Original statement list of FG discussions in German

Note: For illustration purposes the table is divided in 4 elements with each two parts

a) Element 1, part 1

ICF-CY Code	Example statement 1	Example statement 2	Example statement 3
e410 Individual attitudes of immediate family members	„Ich mache einen klaren Unterschied. Muss ich mir von meinem Mann auch manchmal vorwerfen lassen. Ich bin in manchen Sachen ein bisschen übervorsichtig, also, wo ich ehm einfach denke kann ich es ihr zutrauen, kann ich es ihr nicht zutrauen.“	„Er versucht sich oft auf seinen Kleinwuchs zurück zu ziehen wenn es um solche Verantwortungen innerhalb der Familie geht. Und wir fordern das eigentlich auch immer wieder ein, dass er altersgerecht Verantwortung übernimmt.“	„Meine Grundstimmung dazu ist immer das ich froh bin, dass ich ihn hab. Das Kind hat mir bis jetzt nur Freude bereitet ich kann es auch nur ganz genauso sagen.“
b180 Experience of self and time functions	„Nee, ich bin so wie ich bin, bin ich in Ordnung.“	„Also das ist natürlich auch das Thema: Ich will unbedingt größer sein.“	„Also ich komme eigentlich sehr gut mit meinem Kleinwuchs zurecht. Man wächst damit auf.“
e445 Individual attitudes of strangers	„Das Anstarren ist auch das was uns am Meisten begegnet.“	„...die Leute fragen mich ständig.“	„Bemerkungen werden auch manchmal gemacht.“
d240 Handling stress and other psychological demands	„Die macht Musik. Sie spielt dann entweder Geige oder Klarinette.“	„Sie guckt dann halt genauso zurück, damit die einfach mal merken, wie das ist, wenn man so angegafft wird. Sie ignoriert das dann auch teilweise.“	„Computer ganz super, um Aggressionen los zu werden . Musik hört er gerne.“
e115 Products and technology for personal use in daily living	„Einlagen hat sie in den Schuhen.“	„Wir haben ne CPAP Maske.“	„Wie gesagt, Hocker mein bester Freund.“
e310 Immediate family	„Ich schätze das größte Hilfsmittel sind wir zu Hause weil wir eben dann oben die Teller rausholen.“	„Ich denke auch, es hilft bloß reden, Sachen aufarbeiten wo einst Probleme gab ja aber sonst.“	„Ansonsten also mit der Toilette das haben wir seit so einem halben Jahr ehm, wo das eigentlich gut klappt. Bis dahin habe ich eigentlich auch noch helfen müssen.“
s750 Structure of lower extremity	„Ein bisschen Knieprobleme einfach, weil sie so ein bisschen instabil ist. Hat da diese Eightblades.“	„Epiphyse Verödung bekommen, wegen der zu langen Wadenbeine.“	„Operation nämlich am Unterschenkel, eine Achsenkorrektur.“
b280 Sensation of pain	„Ja, das wir vielleicht mehr Schmerzen haben als andere.“	„...weil sie jetzt auch sechs Wochen lang Kopfschmerzen hatte.“	„Letztes Jahr hatte sie verstärkt Rückenschmerzen.“
d920 Recreation and leisure	„Und gut ein Vorteil hat man irgendwie beim Völkerball oder beim Spiel weil man kleiner ist und nicht so gut getroffen wird.“	„Und dann schwimm ich auch noch beim DLRG und ich brauch halt auch einfach bei den Bahnen viel länger.“	„...sie reitet.“
e580 Health services, systems and policies	„Haben natürlich Krankengymnastik und solche Geschichten sowieso gemacht und Osteopathie.“	„Mandeln wurden mal rausgenommen.“	„Logopädie und so.“

a) Element 1, part 2

ICF-CY Code	Example statement 4	Example statement 5	Example statement 6
e410 Individual attitudes of immediate family members	„Aber wie gesagt, die Grundeinstellung bei uns ist immer so wenig, wie möglich und so viel , wie nötig, weil draußen das Leben ist auch nicht für sie eingerichtet.“	„Ist ein ganz normales quietschgesundes Kind.“	„Und ich hab es nie gemocht und das haben meine Eltern auch nie gemacht das der Kleinwuchs so ein besonderes Thema war ich bin einfach so ich war einfach so wie ich bin, so das war’s.“
b180 Experience of self and time functions	„Wir fallen eigentlich immer auf. Also wir als Kleinwüchsige, wir fallen immer auf, ob wir wollen oder nicht.“	„Ja ich bin behindert.“	„Oberschenkel sind natürlich ein bisschen dick.“
e445 Individual attitudes of strangers	„Die trauen sich nicht uns anzusprechen.“	„Manchmal Probleme gehabt mit dem Auslachen.“	„Ich werd auch ganz oft wenn ich alleine rum fahre mit dem Fahrrad oder so werde ich auch ganz oft von Älteren so angesprochen, ob meine Eltern mir das denn schon erlauben das ich weil mich alle für drei oder fünf halten.“
d240 Handling stress and psychological demands	„Also die zieht sich oft zurück.“	„Entweder rausgehen oder mit der Katze schmusen.“	„Bei mir ist es dann halt mit meiner Familie. Versuch zu kommunizieren, Kontakt aufzunehmen. Mit Mama, Papa und meinen Brüdern.“
e115 Products and technology for personal use in daily living	„Zweistufigen Tritt vom Baumarkt.“	„Trip Trap, der ist ja super für alle Kinder, ob groß oder kleinwüchsig.“	„Ich nehme mir ganz einfach einen Schuhlöffel und tu da diesen Hacken da oben irgendwie auf den Kasten so drauf und mach mir das immer so auf.“
e310 Immediate family	„Naja, wenn wir spazieren gehen wollen, dann achten sie schon drauf, dass ich nur so weit gehe, wie ich kann halt.“	„Und ja die versuchen ja auch so wenig wie möglich zu machen.“	„Ja, also meine Mutter unterstützt mich auch, sie kennt das ja selber auch, da weiß sie was man da Beantragen muss.“
s750 Structure of lower extremity	„...weil ich O -Beine hatte.“	„Die Oberschenkel sind ein bisschen dick.“	„Wachstumsfugen veröden lassen im Wadenbein.“
b280 Sensation of pain	„Als Kind immer wieder Ohrenschmerzen Entzündungen aber jetzt mittlerweile geht es gut. Das waren die einzigen zwei.“	„Weil ich im Knie und Sprunggelenk Schmerzen hatte beim Gehen, das ist dadurch beseitigt.“	„Und jetzt natürlich während der OP-Zeit, da hatte sie natürlich schon Schmerzen.“
d920 Recreation and leisure	„Die gehen jetzt ständig auf die Skaterbahn ja und dann fahren die eben Skateboard oder BMX-Rad auf diesen Halfpipes da kann er natürlich nicht mithalten.“	„Ja und weil ich auch nie mit den Armen ans Spielfeld kommen da muss mich manchmal hinlegen damit ich drankomme.“	„Beispiel im Europa Park ne Achterbahn darf ich nicht fahren, nur ab ner bestimmten Größe.“
e580 Health services, systems and policies	„Operation an den Ohren.“	„Sie ist in den Ferien operiert worden, hat diese Eightblades bekommen.“	„Dekompressions-OP am kraniozervikalen Übergang.“

b) Element 2, part 1

ICF-CY Code	Example statement 1	Example statement 2	Example statement 3
d760 Family relationships	„Ich glaub schon, dass wir der Tochter ein bisschen mehr Zeit gewidmet haben, als dem Sohn, ja er hat eigentlich nie sehr geklagt, erst jetzt, je älter das er wird, da kommen manchmal so Dinge, wie ja ich brauch sozusagen auch noch ein bisschen was von euch.“	„Aber ja wahrscheinlich verwöhnen wir sie mehr oder tragen wir ihr mehr hinterher als eigentlich sein müsste mit dem Argument Kleinwuchs und wahrscheinlich stimmt das gar nicht, das ist einfach, weil sie die Nette, Kleine, Süße ist.“	„Ich versuch natürlich die Kinder alle gleich zu behandeln.“
e420 Individual attitudes of friends	Für den Freundeskreis, das ist völlig normal, dass darüber gar nicht mehr gesprochen wird und das ist selbstverständlich.“	„Sie zieht mit ihren Freundinnen los und die gehen auch sehr offensiv damit um und sprechen dann Leute, die so ganz blöde starren auch eben auch an mit sowas, wie: Wollen Sie ein Foto von uns?“	„Ja genau, und meine Freunde vergessen das manchmal auch das ich kleinwüchsig bin. Also die schicken mich in die Küche und ich soll aus dem obersten Schrank was holen, also das haben sie auch schon gemacht, weil sie einfach nicht dran denken. Die sehen mich als normalen Menschen, was wir ja auch sind.“
s710 Structure of head and neck region	„Als Säugling siehst du das ja so extrem das der Kopf so groß ist.“	„...tiefliegende Nasenwurzel.“	„Dem Kopf mit dieser Balkonstrirn.“
d445 Hand and arm use	„Sie kann nicht an alles rankommen.“		
b126 Temperament and personality functions	„Mir geht es nicht schlecht weil ich Kleinwüchsig bin. Mir geht es gut.“	„Also ich denke von Grund auf ist er schon ein fröhliches und selbstbewusstes Kind.“	„Ich find das gut, also ich find auch ich bin wirklich selbstständig.“
e155 Design, construction and building products and technology of buildings for private use	„Ein niedrig gesetztes Waschbecken für sie.“	„Lichter halt alle tief.“	„ein Bad wird umgebaut.“
s299 Eye, ear and related structures, unspecified	„...chronische Mittelohrentzündungen.“	„Loch im Trommelfell.“	„Ich hab einmal Röhrchen bekommen in die Ohren.“
e425 Individual attitudes of acquaintances, peers colleagues, neighbours and community members	„Im Kindergarten ist er sehr gut aufgenommen worden.“	„Da hat sie, genau ein Klassenkamerad mal gefragt, ob sie noch Windeln trägt. Der Tag war dann ganz schlecht, ja, der wollte sie eigentlich ärgern, ne, der weiß, dass sie keine trägt.“	„Das man halt von Mitschülern manchmal gehänselt wird.“

b) Element 2, part 2

ICF-CY Code	Example statement 4	Example statement 5	Example statement 6
d760 Family relationships	„Mit meinen Eltern verstehe ich mich ganz gut.“	„Nee nicht ordentlich, aber mal Zankereien, also das gehört glaub ich zu jeder Geschwisterbeziehung dazu, das es irgendwann mal Reibereien gibt, aber nichts weltbewegendes.“	„Also, ich hab Geschwister, eine Schwester die ist acht Jahre älter als ich. Wir verstehen uns jetzt blendend, denk ich mal, also besser, als denn je.“
e420 Individual attitudes of friends	„Also mir fällt das gar nicht mehr auf und meine Freunde regen sich dann halt zwischendurch mal darüber auf.“		
s710 Structure of head and neck region	„...der Kopfumfang.“	„...diese ganze Enge im Mittelgesicht.“	„da wurde das Hinterhauptsloch vergrößert.“
d445 Hand and arm use			
b126 Temperament and personality functions	„Und jetzt, also er ist cool, absolut, also die können ihn angaffen oder so, das interessiert ihn überhaupt nicht.“		
e155 Design, construction and building products and technology of buildings for private use	„Ja bei uns ist das zum Teil etwas niedriger alles.“		
s299 Eye, ear and related structures, unspecified			
e425 Individual attitudes of acquaintances, peers colleagues, neighbours and community members	„Ja, als wäre ich, als wäre ich ... ich krieg keine Sonderbehandlung.“	„Ja am Anfang ist es glaub ich immer komisch oder da gucken sie einen an oder fragen, so such in ner Diskothek oder so, wenn du immer neue Leute kennenlernst und so. Dann sagen sie ohh ist cool, dass du hier bist und dass du auch feierst und so und dann gibt’s auch ein paar Fragen und warum bist du so und so.“	„Manchmal werde ich in der Schule auch ausgelacht weil ich so klein bin.“

c) Element 3, part 1

ICF-CY Code	Example statement 1	Example statement 2	Example statement 3
e450 Individual attitudes of health professionals	„Hat der Prof. uns verabschiedet, hat gesagt "naja, ähm also da ist schon irgendwie was, aber warten sie mal ab und vieles verwächst sich ja noch" und so. Müssen wir Krankengymnastik machen, irgendwas? Arzt:" Nein, nein, es ist nichts erforderlich." O.k., dann sind wir da wieder raus und haben uns nur angeguckt und haben gesagt, also das stimmt hier nicht, also die machen hier so eine Wolke und das passt einfach nicht, man lässt uns nicht wieder rankommen und erzählt nichts, irgendwas stimmt hier nicht.“	„Aber von den Ärzten erwarte ich einfach, wenn man mit dem Kind hinkommt und sagt schon bei der Anmeldung, mein Kind hat Achondroplasie, dass wenn dann der Termin stattfindet, dass der Arzt wenigstens weiß, was das ist, aber das erschüttert mich immer, dass wir den Ärzten sagen, was zu diesem Krankheitsbild dazugehört.“	„Die Krankenschwestern sind mir so ein bisschen aus dem Weg gegangen das war eine ganz blöde Situation, die sind alle um mich rumgeschlichen. Obwohl, wo ich dachte die wissen schon was aber sagen es mir nicht.“
d730 Relating with strangers	„Ja, zum bsp. jetzt im Geschäft. Wir sind jetzt einkaufen und sie möchte sich jetzt von ihrem Taschengeld irgendwas extra kaufen und die Plätzchen stehen halt da oben, dann spricht sie auch Jemanden an und sagt: Hier, können Sie mir mal bitte die Plätzchen geben?“	„Nur er selber mag nicht antworten und da geht er lieber weg und sagt halt, lass die nur reden oder fragen aber das ist halt seine Strategie, das finde ich auch okay.“	„Fremden ist es auch von der Tagesform abhängig, also manchmal marschiert sie dadurch und wenn irgendwas ist, das perlt bei ihr dann ab wie Wasser an Öl und manchmal dreht sie sich um und fixiert die Leute von oben nach unten und dann drehen die sich um und gehen. Also sie hat dann auch manchmal ein Blick drauf, also da wissen Sie genau, was los ist, dann ist es kein Thema.“
e320 Friends	„Sie hat eine gute Freundin, da wird auch einiges ausgetauscht.“	„Es sind halt immer auch die Freunde die einem auch sozusagen Schutz geben. Die kennt man hier man weiß wie die so sind und so und das gibt einem auch Schutz	„Ja, sie nehmen halt auch Rücksicht, das ist klar.“
d455 Moving around	„Und wenn ich was brauche tendiere ich eben mehr so zum Klettern. Ich bin da dann eher so der aktive, der dann einfach hochklettert und sich die Sachen holt.“	„...die halt vergessen das ich nicht so schnell laufen kann.“	„Also, ich komm ja aus dem läuferischen Bereich und ich merk's jetzt das meine Beine sehr, sehr viel einfacher sind vom Laufen, dass ich viel einfacher bewegen kann.“
d710 Basic interpersonal interactions	„Auch Respekt, die sollen nicht immer so haha lachen halt.“	„Also ich würde mir einfach wünschen, dass einfach die Bevölkerung mehr ja sich mehr beschäftigt mit uns und vielleicht einfach auch mehr es so hinnimmt und uns akzeptiert.“	„Aber ansonsten offener werden einfach auf einen zukommen und fragen was ist los? Nicht dieses nur gucken einfach kommen und fragen.“

c) Element 3, part 2

ICF-CY Code	Example statement 4	Example statement 5	Example statement 6
e450 Individual attitudes of health professionals	„Ein Freund von mir dessen Frau ist OP Schwester war bei der Sektio dabei und hat da am Tisch gestanden und der Operateur oder der zuständige Kollege hat dann gesagt also, rausgeholt und das ist ein Liliput. Das war so die erste Aussage.“	„Bei mir waren die alle immer super freundlich.“	„Weil die mir ja einen im achten Monat noch eine Abtreibung angeboten hatten. Diese Ärzte ich sollte noch abtreiben, weil es sollte kein Arm und kein Bein haben im Ultraschall und es war ja alles so schlimm, und da hab ich gesagt: Sag mal sind sie nicht mehr ganz dicht. Ich sag ich bin im achten Monat schwanger ich treib doch jetzt kein Kind ab. Also ich war so was von entsetzt.“
d730 Relating with strangers	„Aber ich hab auch überhaupt kein Problem, auf Leute zuzugehen und zu sagen: Hey, könnte mal jemand die und die Taste drücken.“	„Ja halt dieses mit weil auch zum Beispiel wenn so Fremde wenn ich gerade meine Familie oder Freunde da sind halt dann muss ich immer fragen und das mag ich nicht so gern. Dauernd so immer zu fragen.“	J“a also die gucken mich dann halt auch so an. Und dann frag ich: Hast du irgendwie Fragen über mich und dann sagen sie immer so Nein weil ich denke die haben immer so fragen aber die trauen sich nicht, die zu stellen.“
e320 Friends	„Sie wollen erst einmal sehen ob ich es alleine schaffe und dann wenn die sehen ahh der braucht Hilfe dann sind sie da ohne das ich fragen muss.“	„Früher als wir immer im Bürgerhaus beim Sport waren und ahmm mit meiner besten Freundin, da kam ich halt noch nicht richtig aufs Klo da gab es halt auch keinen Hocker und da hat sie sich immer wie so einen Hocker gemacht und dann bin ich auf den Rücken und dann hab ich meine Füße darauf abgestellt und dann ja dann hab ich halt immer geschafft.“	„Aber was ihm dann am Meisten hilft, ist wenn er wirklich dann einen Freund einen guten Freund aus der Nachbarschaft treffen kann, wenn einer von denen da ist ehm, das ist dann eher so seine Ebene.“
d455 Moving around			
d710 Basic interpersonal interactions			

d) Element 4, part 1

ICF-CY Code	Example statement 1	Example statement 2	Example statement 3
s760 Structure of trunk	„An der Wirbelsäule Spinalkanal mit Versteifung.“	„Ja und ahmm ich hab auch einen Knick in der Wirbelsäule und deswegen muss ich jeden Mittwoch ins Reha.“	„Dabei hat sich dann herausgestellt, dass da ein Wirbel verrutscht war.“
d450 Walking	„Noch etwas Probleme beim Gehen.“	„Nicht so, also nach längerer Zeit laufen da kommen auch Fußschmerzen oder so aber wenn ich so normales etwas längeres ... langsameres Tempo laufe so für mich so laufe, dann ist ist das eigentlich in Ordnung.“	„Also ich konnte auch wieder weite Strecken laufen ohne Probleme etc.“
d540 Dressing	„Aber so breit, es gibt keine Schuhe für weibliche Wesen, die breite Füße haben und das ist glaub ich so mitunter das größte Problem.“	„Ja mit der Kleidung ebenfalls nicht jeder 18 jährige will halt Lillyfee, Hannah Montana oder sonst so was und Hosen und so ist schon nervig.“	„Kopf irgendwas zum Anziehen weil T-Shirts, weil mir gefällt ein T-Shirt und ja dann geht es nicht.“
d770 Intimate relationships	„Naja, wenn ihr kleinwüchsig wärt, dann wüsstest ihr auch, dass das nicht so einfach ist, mit 15 eine Freundin zu haben.“	„Beziehungen sag ich mal, weil ich denke für einen kleinwüchsigen Mann ist es schwieriger eine große Frau zu bekommen als für eine kleinwüchsige Frau einen großen Mann.“	„...mit der Partnerschaft ist es ein Problem.“
b560 Growth maintenance functions	„Ja und ich will, auch eigentlich auch ein bisschen größer sein.“	„Ich würde gerne mittel sein.“	„...dass sie hoch schauen muss zu den anderen Kindern, also ihre Freundinnen sind zum Teil auch wirklich sehr groß für ihr Alter und sie hat dann auch, dass war wirklich ihr Ausdruck, dass sie wirklich gesagt hat: Ich bin nie auf Augenhöhe mit den anderen Kindern.“
d620 Acquisition of goods and services	„Was sie halt nicht so kann ist einkaufen.“	„Ja, mit ihren Freundinnen geht sie shoppen, liebend gerne.“	„Und ja Kaufhallen, irgendwo man braucht Hilfe, ist das eben ein Problem.“
e120 Products and technology for personal indoor and outdoor mobility and transportation	„Da fahr ich immer mit dem Elektroscooter.“	„...einen Roller hat sie.“	„Ja, und dann halt auch Auto, als wir uns eins neu angeschafft haben, haben wir eins mit Schiebetür genommen, damit sie leichter rein kommt und ja.“

d) Element 4, part 2

ICF-CY Code	Example statement 4	Example statement 5	Example statement 6
s760 Structure of trunk	„Jetzt werden die Probleme mit dem Rücken immer häufiger."		
d450 Walking	„Also ich versuch noch genug zu laufen, aber um das mit dem Rücken zu vermeiden fahr ich jetzt viel damit."		
d540 Dressing	„Die Hosen, da hab ich auch ein Problem."	„Beim Shoppen passt nichts und die Mützen sind ganz oft zu eng."	
d770 Intimate relationships	„Auch völlig normal, also ich hab einen normalwüchsigen Freund und das ist auch eigentlich, ja auch alles normal."		
b560 Growth maintenance functions			
d620 Acquisition of goods and services	„Probleme beim Einkaufen halt, so wenn die meisten Sachen oben stehen, das stört mich eigentlich am meisten."		
e120 Products and technology for personal indoor and outdoor mobility and transportation	„Also mein Auto ist gerade im Umbau, also ich hab's gerade zu einer Firma geschickt, wo der Sitz angepasst wird und die Pedale eingebaut werden."	„...hat ein Therapiefahrrad."	„...jetzt auch halt einen Rolli"

Annex 2: APLES Pilot Version for Children and Parents

a) APLES pilot version for children (8-14 years)

ID:

APLES Children

Date: ______________________________

Name: ______________________________

Last Name: ______________________________

Date of birth: ______________________________

Hello!

How are you?

We would like to know about how you are doing and how you feel about yourself. Therefore we invite you to answer the following questions with regard to your life in general, your height, appearance, stature, and your strengths, difficulties and thoughts.

- Please read each question carefully. Choose the circle that represents your answer most closely.
- Note: This is not an exam! There is no right or wrong answers.
- It is just important that you please answer EACH question and that we can see your marks clearly.

This is how it works:

Example:

If you think about your favorite meal...

I like ice cream.	**I don´t agree at all** ○	**I agree somewhat** ○	**I agree** ○	**I agree a lot** ○	**I totally agree** ○

If you absolutely love eating ice cream then you would mark the box that reads “I totally agree“.

First, we would like to begin with how you feel about yourself.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
1.	I feel okay the way I am.	○	○	○	○	○
2.	I always draw attention from others because of my body.	○	○	○	○	○
3.	I can accomplish more than others might think.	○	○	○	○	○
4.	I find myself to be too fat.	○	○	○	○	○
5.	I am independent in daily life.	○	○	○	○	○
6.	I like my body.	○	○	○	○	○
7.	I think I am just like others and just look somewhat different.	○	○	○	○	○

Now, we would like to learn more about your family.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
8.	My short stature is a big deal to my family.	○	○	○	○	○
9.	I am not treated differently from others by my family.	○	○	○	○	○
10.	I am allowed less than other children my age.	○	○	○	○	○
11.	My family shows consideration for me.	○	○	○	○	○
12	Members of my family help me in daily life.	○	○	○	○	○
13	Because of my short stature my family is less confident in me.	○	○	○	○	○
14	I am treated pretty normally by my family.	○	○	○	○	○

In this part, we would like to learn, how it is for you to be with friends.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
15.	My friends think my short stature is pretty normal.	○	○	○	○	○
16.	I get no special treatment by my friends.	○	○	○	○	○
17.	My friends are considerate of me.	○	○	○	○	○
18.	My friends help me.	○	○	○	○	○
19.	My friends stand up for me.	○	○	○	○	○
20.	My friends protect me.	○	○	○	○	○
21	My friends think that my height and appearance is pretty normal.	○	○	○	○	○
22.	I have problems to find a friend/boyfriend/girlfriend.	○	○	○	○	○

These questions refer to how you feel about recreation and time off school.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
23.	I am able to do the sports that I want do.	○	○	○	○	○
24.	In my free time I can do what I like.	○	○	○	○	○
25.	I find clothes that I like.	○	○	○	○	○
26.	Shopping is difficult for me (e.g. because of not being able to reach things).	○	○	○	○	○

How is kindergarten/school for you?

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
27.	My short stature is a big topic in school/kindergarten.	○	○	○	○	○
28.	I am treated differently in school/kindergarten than other kids.	○	○	○	○	○
29.	Because of my height I am teased or laughed at by others in school/kindergarten.	○	○	○	○	○
30.	Other kids at school/kindergarten help me.	○	○	○	○	○
31.	I am treated considerately at school/kindergarten.	○	○	○	○	○
32.	I am supported by my teachers.	○	○	○	○	○

Thinking about your medical treatment…

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
33.	I have already had many surgeries.	○	○	○	○	○
34.	I need to go to the doctor/therapist quite often.	○	○	○	○	○
35.	I need to see many different doctors/therapists.	○	○	○	○	○
36.	I spend too much time at the doctors/therapists.	○	○	○	○	○

Now we would like to learn more about the reactions of other people towards you (e.g. strangers in the streets).

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
37.	I am treated by strangers like other kids my age.	○	○	○	○	○
38.	Strangers often stare at me.	○	○	○	○	○
39.	I am willing to answer questions of strangers about my short stature.	○	○	○	○	○
40.	Reactions of strangers bother me.	○	○	○	○	○
41.	Strangers often make fun of me or laugh at me.	○	○	○	○	○
42.	If strangers stare at me I talk to them.	○	○	○	○	○
43.	I dare to ask strangers for help.	○	○	○	○	○
44.	Attracting attention by strangers because of my short stature hurts me.	○	○	○	○	○
45.	A lot of people are not familiar with short stature.	○	○	○	○	○
46.	I am open minded towards strangers.	○	○	○	○	○

These questions are about things that help you in daily life.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
47.	With the help of my tools I am able to master daily life by myself (e.g. stool).	○	○	○	○	○
48.	With the help of assistive tools I am able to move around.	○	○	○	○	○
49.	At home a lot of things are adapted to my size (reconstruction, furniture).	○	○	○	○	○

These questions are about your body …

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
50.	The shape and length of my legs causes problems for me.	○	○	○	○	○
51.	I am not able to run fast.	○	○	○	○	○
52.	It is hard for me to move long distances.	○	○	○	○	○
53.	The shape and length of my arms causes problems for me.	○	○	○	○	○
54.	I am able to reach out to everything.	○	○	○	○	○
55.	I am often in pain.	○	○	○	○	○
56.	I experience physical complaints (e.g. my legs falling asleep).	○	○	○	○	○
57.	I had/have physical problems with my ears.	○	○	○	○	○

58.	I find my head to be too big.	○	○	○	○	○
59.	I like my face.	○	○	○	○	○

If you have further comments, please note them in the space below:

Thank you very much for your help!

b) APLES pilot version for parents (of children with ACH aged 5-14 years)

ID:

APLES Parents

Date: ____________________

Name: ____________________

Full name of your child: ____________________

Date of birth of your child: ____________________

Dear parent,

We would like to know how your child is doing and how your child feels about him/herself.

Therefore we invite you to answer the following questions with regard to your child’s life in general, his/her height, strengths, difficulties and thoughts.

Please read each question carefully. Choose the circle that fits best to your answer and mark it. There are no right or wrong answers. It is important that you answer ALL the questions and also that we can see your marks clearly.

This is how it works:

Example:

If you think about the favorite meal of your child…

I like ice cream.	**I don´t agree at all** ○	**I agree somewhat** ○	**I agree** ○	**I agree a lot** ○	**I totally agree** ○

If your child loves eating ice cream, then you would mark the box that reads “I totally agree“.

First, we would like to begin with your child itself

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
1.	My child feels okay the way (s)he is.	○	○	○	○	○
2.	My child always draws attention to him/herself because of his/her body.	○	○	○	○	○
3.	My child can accomplish more than others might think.	○	○	○	○	○
4.	My child finds him-/herself to be too fat.	○	○	○	○	○
5.	My child masters daily life all by him-/herself.	○	○	○	○	○
6.	My child likes his/her body.	○	○	○	○	○
7.	My child thinks (s)he is just like others and just look somewhat different.	○	○	○	○	○

Now, we would like to learn more about your family.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
8.	My child’s short stature is a big issue in our family.	○	○	○	○	○
9.	My child is not treated differently from others in our family.	○	○	○	○	○
10.	My child is allowed less than other children his/her age.	○	○	○	○	○
11.	My family shows consideration for me.	○	○	○	○	○
12	Members of our family help my child in daily life.	○	○	○	○	○
13	Because of my child’s short stature we are less confident in him/her.	○	○	○	○	○
14	We treat out child pretty normal.	○	○	○	○	○

In this part, we would like to learn, how it is for your child to be with friends.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
15.	My child's friends think his/her short stature is pretty normal.	○	○	○	○	○
16.	My child gets no special treatment by his/her friends.	○	○	○	○	○
17.	My child's friends are considerate of him/her.	○	○	○	○	○
18.	My child's friends help him/her.	○	○	○	○	○
19.	My child's friends stand up for him/her,	○	○	○	○	○
20.	My child's friends protect him/her.	○	○	○	○	○
21	My child's friends think that his/her height and appearance is pretty normal.	○	○	○	○	○
22.	My child has problems to find a friend/boyfriend/girlfriend.	○	○	○	○	○

These questions refer to how your child feels about recreation and time off school.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
23.	My child is able to do the sports that (s)he wants do.	○	○	○	○	○
24.	In his/her free time my child can do what (s)he likes.	○	○	○	○	○
25.	My child finds clothes that (s)he likes.	○	○	○	○	○
26.	Shopping is difficult for child (e.g. because of not being able to reach things).	○	○	○	○	○

How is kindergarten/school for your child?

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
27.	My child’s short stature is a big topic in school/kindergarten.	○	○	○	○	○
28.	My child is treated differently in school/kindergarten than other kids.	○	○	○	○	○
29.	Because of his/her height my child is teased or laughed at by others in school/kindergarten.	○	○	○	○	○
30.	Other kids at school/kindergarten help my child.	○	○	○	○	○
31.	My child is treated considerately at school/kindergarten.	○	○	○	○	○
32.	My child is supported by his/her teachers.	○	○	○	○	○

Thinking about your child’s medical treatment…

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
33.	My child already had many surgeries.	○	○	○	○	○
34.	My child needs to go to the doctor/therapist quite often.	○	○	○	○	○
35.	My child needs to see many different doctors/therapists.	○	○	○	○	○
36.	My child spends too much time at the doctors/therapists.	○	○	○	○	○

Now we would like to learn more about the reactions of other people towards your child (e.g. strangers in the streets).

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
37.	My child treated by strangers like other kids his/her age.	○	○	○	○	○
38.	Strangers often stare at my child.	○	○	○	○	○
39.	My child is willing to answer questions of strangers about his/her short stature	○	○	○	○	○
40.	Reactions of strangers bother my child.	○	○	○	○	○
41.	Strangers often make fun of my child or laugh at him/her.	○	○	○	○	○
42.	If strangers stare at my child (s)he talks to them.	○	○	○	○	○
43.	My child dares to ask strangers for help.	○	○	○	○	○
44.	Attracting attention by strangers because of his/her short stature hurts my child.	○	○	○	○	○
45.	A lot of people are not familiar with short stature.	○	○	○	○	○
46.	My child is open minded towards strangers.	○	○	○	○	○

These questions are about things that help your child in daily life.

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
47.	With the help of his/her tools my child is able to master daily life by him-/herself (e.g. stool).	○	○	○	○	○
48.	With the help of assistive tools my child is able to move around.	○	○	○	○	○
49.	At home a lot of things are adapted to the size of my child (reconstruction, furniture)	○	○	○	○	○

These questions are about your child's body …

		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
50.	The shape and length of my child's legs cause problems for him/her.	○	○	○	○	○
51.	My child is not able to run fast.	○	○	○	○	○
52.	It is hard for my child to move long distances.	○	○	○	○	○
53.	The shape and length of my child's arms causes problems for him/her.	○	○	○	○	○
54.	My child is able to reach out to everything.	○	○	○	○	○
55.	My child is often in pain.	○	○	○	○	○
56.	My child experiences physical complaints (e.g. his/her legs falling asleep).	○	○	○	○	○
57.	My child had/has physical problems with his/her ears.	○	○	○	○	○

58.	My child finds his/her head to be too fat.	○	○	○	○	○
59.	My child likes his/her face.	○	○	○	○	○

If you have further comments, please note them in the space below:

____

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Thank you very much for your help!

Annex 3: APLES Field Test Version for Children and Parents

a) APLES field test version for children (8-14 years)

ID:

APLES Children

Date: ______________________________

Name: ______________________________

Last Name: ______________________________

Date of birth: ______________________________

Hello!

How are you?

We would like to know about how you are doing and how you feel about yourself. Therefore we invite you to answer the following questions with regard to your life in general, your height, appearance, stature, and your strengths, difficulties and thoughts.

- Please read each question carefully. Choose the circle that represents your answer most closely.
- Note: This is not an exam! There is no right or wrong answers.
- It is just important that you please answer EACH question and that we can see your marks clearly.

This is how it works:

Example:

If you think about your favorite meal...

I like ice cream.	**I don´t agree at all** ○	**I agree somewhat** ○	**I agree** ○	**I agree a lot** ○	**I totally agree** ○

If you absolutely love eating ice cream then you would mark the box that reads “I totally agree“.

First, we would like to begin with how you feel about yourself.

Self-perception		**I don´t agree at all**	**I agree somewhat**	**I agree**	**I agree a lot**	**I totally agree**
1.	I feel okay the way I am.	○	○	○	○	○
2.	I always draw attention from others because of my short stature.	○	○	○	○	○
3.	I can accomplish more than others might think.	○	○	○	○	○
4.	I find myself to be too heavy.	○	○	○	○	○
5.	I master daily life all by myself.	○	○	○	○	○
6.	I like my body.	○	○	○	○	○

Now, we would like to learn more about your family.

Family		**I don´t agree at all**	**I agree somewhat**	**I agree**	**I agree a lot**	**I totally agree**
7.	My short stature is a big issue in my family.	○	○	○	○	○
8.	Members of my family help me in daily life.	○	○	○	○	○
9.	Because of my short stature my family is less confident in me.	○	○	○	○	○
10.	I am treated pretty normally by my family.	○	○	○	○	○

In this part, we would like to learn, how it is for you to be with friends.

Friends		**I don´t agree at all**	**I agree somewhat**	**I agree**	**I agree a lot**	**I totally agree**
11.	My friends think my short stature is nothing special.	○	○	○	○	○
12.	I am not treated differently from others by my friends.	○	○	○	○	○
13.	My friends are considerate of me.	○	○	○	○	○
14.	My friends help me.	○	○	○	○	○
15.	My friends stand up for me.	○	○	○	○	○
16.	My friends protect me.	○	○	○	○	○
17.	My friends think that my height and appearance is pretty normal.	○	○	○	○	○

These questions refer to how you feel about recreation and time off school.

Recreation		**I don´t agree at all**	**I agree somewhat**	**I agree**	**I agree a lot**	**I totally agree**
18.	I am able to do the sports that I want do.	○	○	○	○	○
19.	In my free time I can do the activities I like.	○	○	○	○	○
20.	I find clothes that I like.	○	○	○	○	○
21.	Shopping is difficult for me (eg. because of not being able to reach things).	○	○	○	○	○
22.	I spend too much time at the doctor/therapist.	○	○	○	○	○

How is kindergarten/school for you?

Kindergarten/School		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
23.	Other kids at school/kindergarten help me.	○	○	○	○	○
24.	I am treated considerately at school/kindergarten.	○	○	○	○	○
25.	I am supported by my teachers.	○	○	○	○	○

Now we would like to learn more about the reactions of other people towards you (e.g. strangers in the streets).

Strangers		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
26.	I am treated by strangers like other kids my age.	○	○	○	○	○
27.	Strangers often make fun of me or laugh at me.	○	○	○	○	○
28.	I dare to ask strangers for help.	○	○	○	○	○
29.	Attracting attention by strangers because of my height is annoying to me.	○	○	○	○	○

These questions are about your body …

	Physical	**I don´t agree at all**	**I agree somewhat**	**I agree**	**I agree a lot**	**I totally agree**
30.	The shape and length of my legs cause problems for me.	○	○	○	○	○
31	It is hard for me to move long distances.	○	○	○	○	○
32.	The shape and length of my arms causes problems for me.	○	○	○	○	○
33.	I experience physical complaints (e.g. my legs falling asleep).	○	○	○	○	○
34.	I find my head to be too big.	○	○	○	○	○
35.	I like my face.	○	○	○	○	○

If you have further comments, please note them in the space below:

____

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Thank you very much for your help!

b) APLES field test version for parents (of children with ACH aged 5-14 years)

ID:

APLES Parents

Date: ________________________________

Name: ________________________________

Full name of your child: ________________________________

Date of birth of your child: ________________________________

Dear parent,

We would like to know how your child is doing and how your child feels about him/herself.

Therefore we invite you to answer the following questions with regard to your child's life in general, his/her height, strengths, difficulties and thoughts.

Please read each question carefully. Choose the circle that fits best to your answer and mark it.There are no right or wrong answers. It is important that you answer ALL the questions and also that we can see your marks clearly.

This is how it works:

Example:

If yout think about the favorite meal of your child...					
My child likes ice cream.	**I don´t agree at all** ○	**I agree somewhat** ○	**I agree** ○	**I agree a lot** ○	**I totally agree** ○

If your child loves eating ice cream, then you would mark the box that says “I totally agree“.

First, we would like to begin with your child itself.

Self-percepetion		**I don´t agree at all**	**I agree somewhat**	**I agree**	**I agree a lot**	**I totally agree**
1.	My child thinks (s)he is okay the way (s)he is.	○	○	○	○	○
2.	My child always draws attention from others because of his/her short stature.	○	○	○	○	○
3.	My child can accomplish more than others might think.	○	○	○	○	○
4.	My child finds him-/herself to be too heavy.	○	○	○	○	○
5.	My child masters daily life all by him-/herself.	○	○	○	○	○
6.	My child likes his/her body.	○	○	○	○	○

Now, we would like to learn more about your family.

Family		**I don´t agree at all**	**I agree somewhat**	**I agree**	**I agree a lot**	**I totally agree**
7.	My child´s short stature is a big deal for my family.	○	○	○	○	○
8.	Members of our family help my child with daily activities.	○	○	○	○	○
9.	Because of my child’s short stature we are less confident in him/her.	○	○	○	○	○
10.	We treat our child pretty normal.	○	○	○	○	○

In this part, we would like to learn, how it ist to be with friends for your child.

Friends		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
11.	My child's friends think his/her short stature is nothing special.	○	○	○	○	○
12.	My child is not treated differently from others by his/her friends.	○	○	○	○	○
13.	My child´s friends are considerate of him/her.	○	○	○	○	○
14.	My child's friends help him/her.	○	○	○	○	○
15.	My child's friends stand up for him him/her.	○	○	○	○	○
16.	My child's friends protect him/her.	○	○	○	○	○
17.	My child's friends think that his/her height and appeareance is pretty normal.	○	○	○	○	○

These questions refer to the leisure time of your child.

Recreation		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
18.	My child is able to do the sports, that (s)he wants to.	○	○	○	○	○
19.	In his/her leisure time my child can do, what (s)he likes.	○	○	○	○	○
20.	My child finds clothes, (s)he likes.	○	○	○	○	○
21.	Shopping is difficult for my child.	○	○	○	○	○
22.	My child spends too much time at the doctor/therapist	○	○	○	○	○

How is kindergarten/school for your child?

Kindergarten/School		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
23.	Other kids at school/kindergarten help my child.	○	○	○	○	○
24.	My child is treated considerately at school/kindergarten.	○	○	○	○	○
25.	My child is supported by his/her teachers.	○	○	○	○	○

In this part, we would like to learn more about the reactions of strangers (e.g. people at the street).

Strangers		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
26.	My child is treated by strangers like other kids his/her age.	○	○	○	○	○
27.	Strangers often make fun of my child or laugh at him/her behind his/her back.	○	○	○	○	○
28.	My child dares to ask strangers for help.	○	○	○	○	○
29.	It hurts my child, that (s)he attracts attention by strangers because of his/her height.	○	○	○	○	○

If you think about the body of your child…

Physical		I don´t agree at all	I agree somewhat	I agree	I agree a lot	I totally agree
30.	The shape and length of my child´s legs cause problems for him/her.	○	○	○	○	○
31	It is hard form my child to move long distances.	○	○	○	○	○
32.	The shape and length of my child´s arms causes problems for him/her.	○	○	○	○	○
33.	My child experiences problems with his/her body (e.g. due to bad blood circulation in extremities).	○	○	○	○	○
34.	My child finds his/her head to be too big.	○	○	○	○	○
35.	My child likes his/her face.	○	○	○	○	○

If you have further comments, please use the space below:

Thank you very much for your help!

Annex 4: Table 19. Complete Item Correlation Analysis with Children Data

Item		Cprop	Cheight	Ctot100
I feel okay the way I am.	Pearson Correlation	-.189	.268	.326
	Sig. (2-tailed)	.761	.608	.476
	N	5	6	7
I always draw attention from others because of my body.	Pearson Correlation	-.954*	.203	.747
	Sig. (2-tailed)	.012	.699	.054
	N	5	6	7
I can accomplish more than others might think.	Pearson Correlation	.478	.319	-.597
	Sig. (2-tailed)	.416	.538	.157
	N	5	6	7
I find myself to be too fat.	Pearson Correlation	-.436	-.063	.586
	Sig. (2-tailed)	.463	.906	.157
	N	5	6	7
I am independent in daily life.	Pearson Correlation	-.948*	.203	.603
	Sig. (2-tailed)	.014	.699	.152
	N	5	6	7
I like my body.	Pearson Correlation	-.085	-.814*	.107
	Sig. (2-tailed)	.892	.049	.820
	N	5	6	7
I think I am just like others and just look somewhat different.	Pearson Correlation	.179	-.224	-.341
	Sig. (2-tailed)	.773	.669	.454
	N	5	6	7
My short stature is a big deal to my family.	Pearson Correlation	.481	-.154	-.763
	Sig. (2-tailed)	.519	.805	.078
	N	4	5	6
I am not treated differently from others by my family.	Pearson Correlation	-.615	.715	.325
	Sig. (2-tailed)	.270	.110	.477
	N	5	6	7
I am allowed less than other children my age.	Pearson Correlation	-.638	.620	.364
	Sig. (2-tailed)	.247	.190	.422
	N	5	6	7
My family shows consideration for me.	Pearson Correlation	.085	.872*	-.058
	Sig. (2-tailed)	.892	.023	.901
	N	5	6	7
Members of my family help me in daily life.	Pearson Correlation	.918*	-.031	-.690
	Sig. (2-tailed)	.028	.953	.086
	N	5	6	7
Because of my short stature my family is less confident in me.	Pearson Correlation	-.364	-.319	.515
	Sig. (2-tailed)	.547	.538	.236
	N	5	6	7
I am treated pretty normally by my family.	Pearson Correlation	-.342	-.548	.504
	Sig. (2-tailed)	.573	.260	.249
	N	5	6	7
My friends think my short stature is pretty normal.	Pearson Correlation	-.259	.087	.554
	Sig. (2-tailed)	.675	.870	.196
	N	5	6	7

(Table 19 cont.)

Item		**Cprop**	**Cheight**	**Ctot100**
I get no special treatment by my friends.	Pearson Correlation	-.567	-.034	.764*
	Sig. (2-tailed)	.319	.949	.045
	N	5	6	7
My friends are considerate of me.	Pearson Correlation	-.490	-.363	.796*
	Sig. (2-tailed)	.402	.479	.032
	N	5	6	7
My friends help me.	Pearson Correlation	-.490	.184	.723
	Sig. (2-tailed)	.402	.728	.061
	N	5	6	7
My friends stand up for me.	Pearson Correlation	-.880*	.518	.847*
	Sig. (2-tailed)	.049	.292	.016
	N	5	6	7
My friends protect me.	Pearson Correlation	-.410	-.159	.746
	Sig. (2-tailed)	.493	.764	.054
	N	5	6	7
My friends think that my height and appearance is pretty normal.	Pearson Correlation	-.461	.543	.427
	Sig. (2-tailed)	.435	.265	.339
	N	5	6	7
I have problems to find a friend/boyfriend/girlfriend.	Pearson Correlation	-.085	-.089	.327
	Sig. (2-tailed)	.892	.867	.473
	N	5	6	7
I am able to do the sports that I want do.	Pearson Correlation	-.442	.144	.718
	Sig. (2-tailed)	.456	.785	.069
	N	5	6	7
In my free time I can do what I like.	Pearson Correlation	-.446	-.185	.667
	Sig. (2-tailed)	.452	.726	.102
	N	5	6	7
I find clothes that I like.	Pearson Correlation	-.989**	.278	.933**
	Sig. (2-tailed)	.001	.594	.002
	N	5	6	7
Shopping is difficult for me (e.g. because of not being able to reach things).	Pearson Correlation	-.302	-.612	.347
	Sig. (2-tailed)	.622	.196	.446
	N	5	6	7
My short stature is a big topic in school/kindergarten.	Pearson Correlation	-.261	.621	-.013
	Sig. (2-tailed)	.672	.188	.978
	N	5	6	7
I am treated differently in school/kindergarten than other kids.	Pearson Correlation	-.305	.421	-.110
	Sig. (2-tailed)	.618	.406	.815
	N	5	6	7
Because of my height I am teased or laughed at by others in school/kindergarten.	Pearson Correlation	.517	-.037	-.185
	Sig. (2-tailed)	.373	.944	.691
	N	5	6	7

(Table 19 cont.)

Item		Cprop	Cheight	Ctot100
Other kids at school/kindergarten help me.	Pearson Correlation	-.153	-.017	.560
	Sig. (2-tailed)	.806	.975	.191
	N	5	6	7
I am treated considerately at school/kindergarten.	Pearson Correlation	-.200	.188	.505
	Sig. (2-tailed)	.747	.722	.248
	N	5	6	7
I am supported by my teachers.	Pearson Correlation	-.580	.616	.208
	Sig. (2-tailed)	.305	.193	.654
	N	5	6	7
I have already had many surgeries.	Pearson Correlation	-.665	.625	.382
	Sig. (2-tailed)	.221	.185	.398
	N	5	6	7
I need to go to the doctor/therapist quite often.	Pearson Correlation	-.451	-.546	-.068
	Sig. (2-tailed)	.446	.262	.885
	N	5	6	7
I need to see many different doctors/therapists.	Pearson Correlation	-.665	.625	.374
	Sig. (2-tailed)	.221	.185	.409
	N	5	6	7
I spend too much time at the doctors/therapists.	Pearson Correlation	-.664	.665	.318
	Sig. (2-tailed)	.222	.149	.487
	N	5	6	7
I am treated by strangers like other kids my age.	Pearson Correlation	-.918*	-.541	.494
	Sig. (2-tailed)	.028	.268	.260
	N	5	6	7
Strangers often stare at me.	Pearson Correlation	-.131	.124	.084
	Sig. (2-tailed)	.833	.814	.858
	N	5	6	7
I am willing to answer questions of strangers about my short stature.	Pearson Correlation	.153	-.427	.085
	Sig. (2-tailed)	.806	.398	.856
	N	5	6	7
Reactions of strangers bother me.	Pearson Correlation	.391	-.480	-.161
	Sig. (2-tailed)	.515	.335	.731
	N	5	6	7
Strangers often make fun of me or laugh at me.	Pearson Correlation	-.850	.116	.547
	Sig. (2-tailed)	.068	.827	.204
	N	5	6	7
If strangers stare at me I talk to them.	Pearson Correlation	.368	-.298	-.181
	Sig. (2-tailed)	.542	.567	.697
	N	5	6	7
I dare to ask strangers for help.	Pearson Correlation	.774	-.445	-.754
	Sig. (2-tailed)	.124	.377	.050
	N	5	6	7
Attracting attention by strangers because of my short stature hurts me.	Pearson Correlation	.220	.663	-.396
	Sig. (2-tailed)	.722	.152	.380
	N	5	6	7

(Table 19 cont.)

Item		**Cprop**	**Cheight**	**Ctot100**
A lot of people are not familiar with short stature.	Pearson Correlation	-.555	.536	.380
	Sig. (2-tailed)	.332	.273	.400
	N	5	6	7
I am open minded towards strangers.	Pearson Correlation	-.375	-.457	.389
	Sig. (2-tailed)	.534	.362	.388
	N	5	6	7
With the help of my tools I am able to master daily life by myself (e.g. stool).	Pearson Correlation	-.075	-.595	.164
	Sig. (2-tailed)	.905	.212	.725
	N	5	6	7
With the help of assistive tools I am able to move around.	Pearson Correlation	.272	-.806	.132
	Sig. (2-tailed)	.658	.053	.778
	N	5	6	7
At home a lot of things are adapted to my size (reconstruction,furniture).	Pearson Correlation	-.430	.231	.050
	Sig. (2-tailed)	.470	.660	.914
	N	5	6	7
The shape and length of my legs causes problems for me.	Pearson Correlation	-.476	-.216	.595
	Sig. (2-tailed)	.417	.682	.159
	N	5	6	7
I am not able to run fast.	Pearson Correlation	.330	-.309	.161
	Sig. (2-tailed)	.588	.551	.731
	N	5	6	7
It is hard for me to move long distances.	Pearson Correlation	.091	-.561	.164
	Sig. (2-tailed)	.884	.247	.725
	N	5	6	7
The shape and length of my arms causes problems for me.	Pearson Correlation	-.417	-.698	.390
	Sig. (2-tailed)	.485	.123	.387
	N	5	6	7
I am able to reach out to everything.	Pearson Correlation	-.422	-.107	.386
	Sig. (2-tailed)	.479	.840	.393
	N	5	6	7
I am often in pain.	Pearson Correlation	-.833	-.526	.308
	Sig. (2-tailed)	.080	.284	.501
	N	5	6	7
I experience physical complaints (e.g. my legs falling asleep).	Pearson Correlation	.857	.107	-.638
	Sig. (2-tailed)	.064	.840	.123
	N	5	6	7
I had/have physical problems with my ears.	Pearson Correlation	-.059	.484	.053
	Sig. (2-tailed)	.925	.330	.910
	N	5	6	7
I find my head to be too big.	Pearson Correlation	.436	.282	-.553
	Sig. (2-tailed)	.463	.588	.198
	N	5	6	7
I like my face.	Pearson Correlation	-.444	-.752	.404
	Sig. (2-tailed)	.454	.084	.369
	N	5	6	7

* The correlation is significant at a level of 0.05 (2-tailed) / ** The correlation is significant at a level of 0.01 (2-tailed)

Annex 5: Table 20. Complete Item Correlation Analysis with Parent Data

Item		Pprop	Pheight	Ptot100
My child feels okay the way (s)he is.	Pearson Correlation	.457	.101	.428
	Sig. (2-tailed)	.101	.709	.068
	N	14	16	19
My child always draws attention to him/herself because of his/her body.	Pearson Correlation	-.065	-.091	.052
	Sig. (2-tailed)	.818	.727	.827
	N	15	17	20
My child can accomplish more than others might think.	Pearson Correlation	.236	.087	.036
	Sig. (2-tailed)	.417	.748	.884
	N	14	16	19
My child finds him-/herself to be too fat.	Pearson Correlation	.338	.615**	.060
	Sig. (2-tailed)	.217	.009	.802
	N	15	17	20
My child masters daily life all by him-/herself.	Pearson Correlation	.825**	.777**	-.053
	Sig. (2-tailed)	.000	.000	.830
	N	14	16	19
My child likes his/her body.	Pearson Correlation	.238	.088	.395
	Sig. (2-tailed)	.393	.738	.085
	N	15	17	20
My child thinks (s)he is just like others and just look somewhat different.	Pearson Correlation	.363	.211	.422
	Sig. (2-tailed)	.183	.416	.064
	N	15	17	20
My child’s short stature is a big issue in our family.	Pearson Correlation	-.424	-.192	.042
	Sig. (2-tailed)	.115	.460	.860
	N	15	17	20
My child is not treated differently from others in our family.	Pearson Correlation	.387	.288	-.003
	Sig. (2-tailed)	.154	.262	.990
	N	15	17	20
My child is allowed less than other children his/her age.	Pearson Correlation	-.326	-.322	.309
	Sig. (2-tailed)	.235	.208	.185
	N	15	17	20
My family shows consideration for me.	Pearson Correlation	-.742**	-.519*	.118
	Sig. (2-tailed)	.002	.033	.621
	N	15	17	20
Members of our family help my child in daily life.	Pearson Correlation	-.615*	-.481	-.019
	Sig. (2-tailed)	.015	.051	.935
	N	15	17	20
Because of my child’s short stature we are less confident in him/her.	Pearson Correlation	-.610*	-.842**	.037
	Sig. (2-tailed)	.020	.000	.884
	N	14	15	18
We treat out child pretty normal.	Pearson Correlation	.360	.489	-.032
	Sig. (2-tailed)	.206	.054	.900
	N	14	16	18
My child’s friends think his/her short stature is pretty normal.	Pearson Correlation	.161	.368	.284
	Sig. (2-tailed)	.566	.146	.225
	N	15	17	20

(Table 20 cont.)

Item		Pprop	Pheight	Ptot100
My child gets no special treatment by his/her friends.	Pearson Correlation	.288	.202	.276
	Sig. (2-tailed)	.297	.437	.239
	N	15	17	20
My child's friends are considerate of him/her.	Pearson Correlation	.047	.070	.065
	Sig. (2-tailed)	.867	.790	.786
	N	15	17	20
My child's friends help him/her.	Pearson Correlation	.109	-.055	.179
	Sig. (2-tailed)	.699	.833	.451
	N	15	17	20
My child's friends stand up for him/her,	Pearson Correlation	.356	.117	.463*
	Sig. (2-tailed)	.193	.654	.040
	N	15	17	20
My child's friends protect him/her.	Pearson Correlation	.440	.121	.411
	Sig. (2-tailed)	.101	.644	.072
	N	15	17	20
My child's friends think that his/her height and appearance is pretty normal.	Pearson Correlation	-.016	.278	.201
	Sig. (2-tailed)	.956	.279	.395
	N	15	17	20
My child has problems to find a friend/boyfriend/girlfriend.	Pearson Correlation	-.275	-.412	.137
	Sig. (2-tailed)	.321	.113	.577
	N	15	16	19
My child is able to do the sports that (s)he wants do.	Pearson Correlation	.398	.366	.086
	Sig. (2-tailed)	.142	.149	.718
	N	15	17	20
In his/her free time my child can do what (s)he likes.	Pearson Correlation	.426	.222	.193
	Sig. (2-tailed)	.113	.391	.416
	N	15	17	20
My child finds clothes that (s)he likes.	Pearson Correlation	-.408	-.101	.065
	Sig. (2-tailed)	.131	.700	.787
	N	15	17	20
Shopping is difficult for child (e.g. because of not being able to reach things).	Pearson Correlation	-.195	-.097	-.122
	Sig. (2-tailed)	.486	.711	.609
	N	15	17	20
My child's short stature is a big topic in school/kindergarten.	Pearson Correlation	-.403	-.298	-.020
	Sig. (2-tailed)	.136	.246	.933
	N	15	17	20
My child is treated differently in school/kindergarten than other kids.	Pearson Correlation	-.356	-.199	-.012
	Sig. (2-tailed)	.193	.443	.961
	N	15	17	20
Because of his/her height my child is teased or laughed at by others in school/kindergarten.	Pearson Correlation	.369	.368	-.316
	Sig. (2-tailed)	.175	.161	.188
	N	15	16	19

(Table 20 cont.)

Item		Pprop	Pheight	Ptot100
Other kids at school/kindergarten help my child.	Pearson Correlation	-.186	-.132	-.067
	Sig. (2-tailed)	.508	.615	.780
	N	15	17	20
My child is treated considerately at school/kindergarten.	Pearson Correlation	.091	.411	-.170
	Sig. (2-tailed)	.746	.101	.474
	N	15	17	20
My child is supported by his/her teachers.	Pearson Correlation	.056	.459	.304
	Sig. (2-tailed)	.855	.085	.219
	N	13	15	18
My child already had many surgeries.	Pearson Correlation	-.111	-.206	.127
	Sig. (2-tailed)	.707	.443	.606
	N	14	16	19
My child needs to go to the doctor/therapist quite often.	Pearson Correlation	-.598*	-.459	.005
	Sig. (2-tailed)	.018	.064	.983
	N	15	17	20
My child needs to see many different doctors/therapists.	Pearson Correlation	-.441	-.572*	.164
	Sig. (2-tailed)	.100	.016	.488
	N	15	17	20
My child spends too much time at the doctors/therapists.	Pearson Correlation	-.438	-.542*	.243
	Sig. (2-tailed)	.103	.024	.301
	N	15	17	20
My child treated by strangers like other kids his/her age.	Pearson Correlation	.053	.293	-.030
	Sig. (2-tailed)	.851	.254	.902
	N	15	17	20
Strangers often stare at my child.	Pearson Correlation	-.007	-.130	.229
	Sig. (2-tailed)	.981	.619	.330
	N	15	17	20
My child is willing to answer questions of strangers about his/her short stature	Pearson Correlation	.253	.716**	-.029
	Sig. (2-tailed)	.383	.002	.907
	N	14	16	19
Reactions of strangers bother my child.	Pearson Correlation	-.090	.250	-.332
	Sig. (2-tailed)	.751	.333	.153
	N	15	17	20
Strangers often make fun of my child or laugh at him/her.	Pearson Correlation	.408	.250	-.325
	Sig. (2-tailed)	.131	.333	.161
	N	15	17	20
If strangers stare at my child (s)he talks to them.	Pearson Correlation	-.030	.222	.106
	Sig. (2-tailed)	.916	.392	.658
	N	15	17	20
My child dares to ask strangers for help.	Pearson Correlation	.323	.386	-.271
	Sig. (2-tailed)	.240	.125	.248
	N	15	17	20

(Table 20 cont.)

Item		**Pprop**	**Pheight**	**Ptot100**
Attracting attention by strangers because of his/her short stature hurts my child.	Pearson Correlation	.095	.388	-.323
	Sig. (2-tailed)	.736	.124	.165
	N	15	17	20
A lot of people are not familiar with short stature.	Pearson Correlation	.019	-.158	-.171
	Sig. (2-tailed)	.949	.559	.485
	N	14	16	19
My child is open minded towards strangers.	Pearson Correlation	.151	.286	-.324
	Sig. (2-tailed)	.590	.266	.164
	N	15	17	20
With the help of his/her tools my child is able to master daily life by him-/herself (e.g. stool).	Pearson Correlation	.509	.536*	-.002
	Sig. (2-tailed)	.053	.027	.994
	N	15	17	20
With the help of assistive tools my child is able to move around.	Pearson Correlation	.426	.527*	-.157
	Sig. (2-tailed)	.128	.036	.520
	N	14	16	19
At home a lot of things are adapted to the size of my child (reconstruction, furniture)	Pearson Correlation	.034	.161	-.293
	Sig. (2-tailed)	.903	.537	.211
	N	15	17	20
The shape and length of my child's legs cause problems for him/her.	Pearson Correlation	-.031	-.066	.176
	Sig. (2-tailed)	.912	.802	.458
	N	15	17	20
My child is not able to run fast.	Pearson Correlation	-.333	-.173	-.337
	Sig. (2-tailed)	.225	.506	.146
	N	15	17	20
It is hard for my child to move long distances.	Pearson Correlation	-.289	-.290	.209
	Sig. (2-tailed)	.297	.259	.377
	N	15	17	20
The shape and length of my child's arms causes problems for him/her.	Pearson Correlation	-.050	-.192	-.292
	Sig. (2-tailed)	.860	.461	.212
	N	15	17	20
My child is able to reach out to everything.	Pearson Correlation	-.150	.069	-.197
	Sig. (2-tailed)	.594	.792	.406
	N	15	17	20
My child is often in pain.	Pearson Correlation	.267	.229	-.026
	Sig. (2-tailed)	.336	.378	.913
	N	15	17	20
My child experiences physical complaints (e.g. his/her legs falling asleep).	Pearson Correlation	.323	.172	.589**
	Sig. (2-tailed)	.240	.524	.008
	N	15	16	19

(Table 20 cont.)

Item		**Pprop**	**Pheight**	**Ptot100**
My child had/has physical problems with his/her ears.	Pearson Correlation	-.254	-.118	.025
	Sig. (2-tailed)	.361	.652	.916
	N	15	17	20
My child finds his/her head to be too fat.	Pearson Correlation	.196	.029	.016
	Sig. (2-tailed)	.484	.912	.948
	N	15	17	20
My child likes his/her face.	Pearson Correlation	.439	.103	.200
	Sig. (2-tailed)	.102	.694	.399
	N	15	17	20

* The correlation is significant at a level of 0.05 (2-tailed) / ** The correlation is significant at a level of 0.01 (2-tailed)

Statutory Declaration

I declare that I have authored this thesis independently, that I have not used other than the declared sources/resources, and that I have explicitly marked all material which has been quoted either literally or by content from other sources.

Date	Signature