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Caring for a child with Bardet-Biedl syndrome: A qualitative study of the parental experiences of daily coping and support

Deniz Zelihić[a], Finn R. Hjardemaal[b,1], Charlotte von der Lippe[a,*,1]

[a] Centre for Rare Disorders, Rikshospitalet, Oslo University Hospital HF, Norway
[b] University of South-Eastern Norway, Norway

A B S T R A C T

This study aimed to explore the parental experiences of having a child with Bardet–Biedl syndrome (BBS) and how parents managed to cope with this situation. Five parents of children with BBS (0–18 years old) participated in semistructured in-depth interviews. Inductive thematic analysis was used to identify themes. The parents experienced distress due to a lack of knowledge on BBS in their support system (e.g., school staff, clinicians, and family members), and they found it stressful to coordinate with multiple support services. Socialization at work, support from family members, and communicating with other parents who are in a similar situation promoted better coping and adaptations to daily life.

Results highlight the importance of parents receiving adequate support while they face daily challenges. An increased knowledge on how rare disorders impact family life is needed in the support system.

1. Introduction

Bardet–Biedl syndrome (BBS) is a rare autosomal recessive ciliopathy (Forsythe and Beales, 2013) with an estimated prevalence of 1–9/1,000,000 (Orphanet, 2008). BBS is characterized by six primary features: retinal dystrophy, postaxial polydactyly, obesity, genital anomalies, renal anomalies, and learning disorders (Forsythe et al., 2018). Secondary features include speech delay, developmental delay, diabetes mellitus, dental anomalies, congenital heart disease, brachydactyly/syndactyly, ataxia/poor coordination, and anosmia/hyposmia (Forsythe et al., 2018). Over the last two decades, 21 genes that account for 80% of the features associated with BBS were identified (Forsythe et al., 2018). A Danish study reported that patients carrying *BBS1* display fewer learning disorders and obesity signs compared with those carrying the other genes associated with this condition (*BBS2* and *BBS10*) (Hjortshøj et al., 2010). However, individuals with BBS experience both intrafamilial and interfamilial variations in their symptoms (Forsythe et al., 2018).

BBS diagnosis is often delayed (Forsythe et al., 2018). This delay may be explained by the development of retinal dystrophy, which manifests later in childhood (Beales et al., 1999). Moreover, this delay is caused by the shared similarity of some BBS symptoms with other conditions, such as Alström syndrome and Meckel–Gruber syndrome (Iannello et al., 2002; Karmous-Benailly et al., 2005).

Studies of the experiences of parents living with a child with BBS, wherein such experiences are based on the parent's first-person perspectives (Gallagher, 2012), are scarce. One qualitative study revealed that parents experienced stigmatization from healthcare professionals, from family members, and from others as a consequence of having an obese child with BBS (Hamlington et al., 2015). These parents reported that their experiences triggered strong emotions characterized by anger, frustration, and helplessness. Unfortunately, health-related stigmas are well-known phenomena (Hamlington et al., 2015; Weiss et al., 2006).

Rare disorders comprise different conditions that affect a limited number of people (Garrino et al., 2015). In Europe, one criterion used to classify a disorder as rare is when it affects one out of 2000 individuals (Eurordis, 2014). Most rare disorders are genetic (Garrino et al., 2015), and it has been estimated that more than 30 million people in Europe suffer from one of the 7000–8000 known rare conditions (Eurordis, 2014). Efficient treatment methods are developed for some rare disorders, albeit no cure exists for the majority of these cases (Institute of Medicine (US) Committee on Accelerating Rare Diseases Research and Orphan Product Development, 2010).

Many individuals who suffer from rare conditions share common challenges, including chronic afflictions, deteriorating alterations, and severe functional impairment (Garrino et al., 2015). One systematic review of the existing qualitative literature revealed the significant challenges faced by adults living with rare disorders (von der Lippe et al., 2017). These challenges included extensive medical,

* Corresponding author.
E-mail address: uxhelc@ous-hf.no (C.v.d. Lippe).
[1] These authors contributed equally to the article.

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psychological, and social difficulties and a general lack of knowledge about rare disorders, which could result in an incorrect or delayed patient diagnosis (Kole and Faurisson, 2009; Nutt and Limb, 2011).

Parents who have a child with a rare condition report similar challenges. Adapting to new responsibilities and experiencing increased pressure on their personal capabilities and resources may become part of their daily lives (Dellve et al., 2006). In addition, these parents are continuously concerned about their capacity to accommodate their child's needs and to acquire sufficient support for their child in the future (Pelentsov et al., 2015). In other words, they face lifelong commitments as caregivers, caretakers, and providers (Dellve et al., 2006; Pelentsov et al., 2015). Fortunately, many parents manage to cope, to stay positive, and to adapt to their child's condition (Nabors et al., 2018).

Parents may benefit from receiving early advice and information from healthcare professionals on how to cope with and adapt to their child's condition (Lundberg et al., 2016). Support and care from other family members are also essential; for example, siblings may provide protection, support, and emotional support to their brother or sister with a chronic condition (Nabors et al., 2018). Unfortunately, many parents of children with disabilities must learn to manage their situation on their own without receiving sufficient information about their child's condition or support from their support system (Tracey et al., 2018; Brown et al., 2012; Dreyfus and Dowse, 2018). Therefore, it is imperative that healthcare professionals and other service providers understand these parents' needs, especially those related to adaptation and receipt of sufficient information, and they must ensure good dissemination and accessibility of information (Tracey et al., 2018; Biesecker and Erby, 2008). The parents' support sources, both formal (e.g., healthcare professionals and service providers) and informal (e.g., friends and family), are likely to significantly influence adaptation outcomes (Woodman, 2014).

Parents of children with chronic and/or genetic conditions may become anxious with the vulnerability of their children to major changes in their lives, such as their transition from kindergarten to school. A recent study revealed that parents of children with mental and/or physical disabilities had bad experiences with the support services offered by their school systems after their child's transition from kindergarten to first grade (Siddiqua and Janus, 2017). These parents were uncertain about the available services, resources, and aid that they were entitled to. Moreover, some parents reported their school system's inadequate administration of information related to their child's condition, causing unnecessary obstacles post transition (Siddiqua and Janus, 2017).

When parents learn about their child's life-altering condition, they may experience psychological stress (Muscara et al., 2015), uncertainty (Hinton and Kirk, 2017), and the loss of their expectations for having a healthy baby (Biesecker and Erby, 2008). In addition, they may experience despair and other emotional reactions, such as shock and sorrow (Fernández-Alcántara et al., 2015; Beresford, 1994; Dellve et al., 2006). Parents of children with diverse chronic illnesses usually have higher stress levels than parents of healthy children (Cousino and Hazen, 2013; Pinquart, 2018). Therefore, adapting to their child's condition and to the associated daily challenges becomes pivotal in minimizing emotional distress and in facilitating coping efforts.

Parents may respond differently to their child's condition. The transactional theory of stress and coping emphasizes the reciprocal influence between personal characteristics (e.g., appraisals, thoughts, and expectations) and environmental factors (e.g., situations, challenges, and strains) (Lazarus and Folkman, 1984). However, in understanding how a rare disorder may impact a family as a whole, it is valuable to incorporate a framework in which the family is viewed as a system consisting of different parts (Minuchin, 1985; Kazak, 1989; Hoffman, 1981). In essence, a child's chronic condition impacts not only the affected individual but also the whole family. Bronfenbrenner's ecological systems framework considers the importance of continuous interactions between the individual and his/her environment (Bronfenbrenner, 1979, 2005). The framework differentiates the five systems that constitute an individual's environment: The *microsystem* (immediate settings, such as the family), *mesosystem* (relationships between the microsystems), *exosystem* (connections and processes where the individual is not directly present but where the microsystems are affected), *macrosystem* (blueprint of the environment), and *chronosystem* (illustrates how an individual's development is affected by life events and experiences). Accordingly, parents' experiences of having a child with a rare disorder can be understood in the light of the different settings in which they live and work.

BBS is a rare disorder that involves psychosocial and physical challenges, and little knowledge is known as regards the parents' experiences of taking care of their child with BBS. Deeper knowledge of such experiences may act as a guide for healthcare professionals and may serve as impetus for institutions offering clinical and social services to provide adequate help to families with a child with BBS.

1.1. Aim

The main purpose of this study was to answer the following question: "How do parents describe their own experiences of having a child with BBS?" This question was addressed in the light of three specific questions: How do the parents describe their experience in taking care of their child with BBS? Which of their experiences promote coping with their situation? How do the parents describe their relationships with their support system?

2. Methodology

2.1. Ethical considerations

This study was approved by the data protection officer of the Oslo University Hospital (reference number: 2016/9984), and it was conducted in accordance with the Helsinki Declaration. To protect the privacy of the parents and to ensure confidentiality, we refer to the participants as Parent 1–5, and we kept the genders of the parents and their children confidential.

2.2. Study design

This study was conducted based on an exploratory qualitative research approach. This project was initiated by the Center for Rare Disorders of the Oslo University Hospital in Norway. This interdisciplinary competence center provides information, counselling, support, and seminars on roughly 70 rare disorders.

2.3. Recruitment of participants and the study sample

There is no registry for individuals with BBS in Norway. However, the Center for Rare Disorders has a voluntary registry of 50–60 individuals with BBS (Senter for sjeldne diagnoser, 2018). Fifteen children (aged 0–18 years) with BBS were registered in 2016/2017. These children's parents were sent (by post) an invitation, which consists of various documents, namely, informed consent, study purpose, participant's rights, and participation anonymity. We did not send a reminder. The sole inclusion criteria were parents of children with a clinically or genetically confirmed diagnosis of BBS. None of those who declined the invitation gave a reason for not participating. The final sample included five Norwegian-speaking parents with children aged 0–16 years (participation rate: 33%).

2.4. Data collection

Data were collected by telephone using semistructured in-depth interviews; telephone interviews were conducted given that the parents

resided all over the country. A final-year student in psychology, who was unfamiliar with the participants and the BBS diagnosis, conducted the interviews. One parent was interviewed at the Center for Rare Disorders. The average interview duration was 49 min (range: 25–77 min). The participants did not check the data. The interviewer used an interview guide containing guidelines for the questions to be asked and the topics to be highlighted during the interview. The open-ended questions asked about the time of diagnosis, the parents' experiences of any challenges related to having a child with BBS, the kinds of support provided for the child and the family, the effect of the disorder to the family, the information the parents or the children told others regarding BBS, and the parents' experiences regarding the medical and psychosocial support provided for their child.

2.5. Data analysis

Each interview was recorded and transcribed verbatim. The interview transcripts were analyzed using inductive thematic analysis as described by Braun and Clarke (2006). The first author listened to the recorded interviews and read the transcriptions several times. The first author coded the entire material line-by-line in tandem with the last author. All authors discussed the coding and the subsequent analysis. The codes were collected under thematic headings, and the sections with associated headings were grouped into subthemes. Next, the subthemes with mutual thematic associations were combined to form the main themes, which were then reviewed by comparing them with the transcripts and with the research questions. In the end, the analysis constituted three main themes. The subthemes and the main themes were compared with the transcripts to ensure consistency between the findings and the actual raw data. The comprehensive and direct excerpts of the transcripts are included in the results section to associate the final analysis with the parents' own words.

3. Results

The results of the present study illustrate the parents' own lived experiences of having a child with BBS. We present the results of our in-depth analysis of three main themes: the first year of the child's life, challenges of everyday living, and positive experiences.

3.1. The first year of the child's life

All of the parents described the time after childbirth as a demanding period. Inadequate knowledge about the cause of their child's difficulties contributed to the daily challenges and uncertainties. The lack of knowledge about BBS among their local healthcare professionals made the diagnosis difficult, leaving almost all of the parents unsatisfactorily informed about their child's condition. Many of the parents were distressed as described by Parent 3: "*There was no one who could give us any information … or a certain diagnosis.…So we lived with great uncertainty … it was very tough*!!" Moreover, Parent 3 explained that their child was given the wrong diagnosis prior to the confirmation of the BBS diagnosis: "*That time when we initially got the [other rare genetic diagnosis,]…. it was terribly hard.*" For some parents (3/5), it was a shock to learn that their child was born with a rare disorder that neither they nor the hospital had any experience with. The time-consuming assessments at the hospital often resulted in poor sleep, and the hospital became a kind of unwanted "second home":

> I almost lived at the hospital as I remember, and when I go back there today, I almost become like, 'oh I can't bear that smell and those hallways'. I was familiar with the whole [hospital].…when I look back, I think it was incredibly tough. It was incredibly tough. (Parent 4)

Four of the parents explained how their babies were clearly different from the other infants, and they described features associated with autism. They found it difficult not being able to participate in mutual communication with their child. This difficulty was especially challenging when the child could not express his/her own needs, making it very difficult for the parents to know whether their child was in pain or was in need of something:

> Yes, there were some signs of something wrong.…I noticed that [the child] was a bit unusual compared to other children. When [the child] was a baby, [the child] didn't smile like other babies, and didn't play in the same way, [the child] was quite passive—not interested in toys, not interested in other people, and [the child] didn't look us in the eyes. (Parent 1)

3.2. Challenges of everyday living

Most of the parents (4/5) found it difficult to tell about their child's disorder to others and to spend considerable time explaining their child's condition when meeting new people. One parent described feelings of distress, sadness, and guilt in the beginning when they talk about the disorder: "*I felt depressed for several years, and it was difficult thinking about why it happened to us. What did we do wrong?*" (Parent 1).

Several parents (3/5) emphasized the difficulty that other individuals have had in understanding their situation. These parents' concerns were not always taken seriously, and the feeling of not being believed often resulted in feelings of incompetence despite their unwavering efforts:

> A little before [the child] was 2 years old, we asked [the kindergarten] if they could refer [the child] to a speech therapist, but they wouldn't … I had to just calm myself with the fact that kids develop differently.…So eventually, I contacted the educational-psychological service and the speech therapist and referred [the child] myself. (Parent 5)

Unfortunately, this parent reported that he/she was faced with disrespect from a physician after a consultation: "*I remember a physician who said, 'Can't you just accept the fact that your child is immature?*'" (Parent 5). Another parent talked about family members not accepting the child's disorder as a permanent condition, and they preferred not to be open about the disorder.

Moreover, the parents described how having a child with a rare disorder impacted the whole family. It was not easy to find spare time to spend on personal interests or to engage time with friends and other family members. One parent even explained that the opportunity to do daily chores was kind of a blessing:

> If we want something, we have to wait until [the child] is at the respite residence, and it can be something so simple … as, for example, that my husband and I want to go together to the store, then we must wait for [the child] to be at the respite residence.…So it's something ordinary people don't consider to be special, but for us, it is. (Parent 1)

Several parents (3/5) talked about not being properly informed about the available social welfare services and the rights that they were entitled to. The parents had to identify information and support on their own initiative; according to them, they would have had better time to prepare and a chance to provide better care if they had been informed earlier about the disorder's complications. Often, the parents found themselves in a lonely struggle:

> I am very much my own secretary. I feel that I use plenty of time on everything from … applications, finding out who is the coordinator for [my child], getting a physiotherapist.…All the paperwork around [the child] … is a very big burden. (Parent 4)

A common concern was related to the child's rigidness and behavioral problems, together with the challenge of identifying the suitable leisure activities. These concerns often contributed to segregation from

peers, solitude, and isolation from the local community. The burden of continuously learning about new complications associated with the disorder was also emphasized.

3.3. Positive experiences

Two parents mentioned that being at work enabled them to have social interactions through which they receive support from their colleagues: "*It was so ok being at work and being around those who could understand you. Many gave me advice and were there* supporting *me.*" (Parent 4). Being at work represented a "normal" part of the day, and it reduced the feeling of being isolated from the outside world.

All of the parents admitted that being open to others about their child's diagnosis mattered to them, and it had a positive effect. Moreover, two parents who were active in the BBS patient organization emphasized the benefits of being acquainted with other parents who are in the same situation. They discussed their challenges and shared their experiences, and they provided each other information and advice. One parent explained that it was good to "*… get a little more understanding that it was the disorder that makes the [children] that way … it is not necessarily us not doing enough.*" (Parent 2). The parents really appreciated supportive family members who dedicated their time to help and socialize with the child:

> They were with me and supporting me when suddenly, during nighttime [the child] couldn't breathe. [The child] was sick and someone could take us to the emergency unit. My brother or my sister or mom [was] there helping me …; they have always been supportive. (Parent 4)

Parent 4 also emphasized the importance of having a lot of courage and willpower as a caregiver, especially when one has a child with special needs:

> If you have children with special needs, then you know how tough it is, but … how much support one gets and how one deals with it … is what I think should be the aim … not that my child is born with this or that syndrome, you can't do anything about that. (Parent 4)

Although the parents described many challenges, they mentioned positive feedback about the personnel they encountered in support meetings, kindergartens, schools, and healthcare facilities. One parent highlighted a good collaboration with a school that exerted efforts for the child:

> In school, [the child] had the same special educator for six years.… [The child] has a very good relationship with her. She had [the child] in math, Norwegian, and English, and she … teaches [the child] English by singing songs.…They were a good match together. She didn't focus on getting through the entire curriculum; she focused on [the child's] skills of learning and coping. I felt she did it in a good way. (Parent 5)

Another parent pointed to their physician's good efforts:

> The communication with the physician is very good … he didn't know anything about … that disorder, but he was very motivated to learn about it, and he is very interested to help.…We have a very good collaboration with the physician. (Parent 1)

Several parents (3/5) wished they had gotten in touch with the Center for Rare Disorders early on because this facility could have given them more support initially. Specifically, one parent described the need for a permanent counsellor or someone to turn to: "*I would have avoided going through so much tears, sweat, time, energy, and all.*" (Parent 4). Not having a qualified team available and the opportunity to become acquainted with other parents of children with BBS were highlighted as primary obstacles:

> It would have been a dream.…having a person to turn to, instead of. …finding out of things and search[ing] [for yourself], and all that frustration going through all of this. It is … the biggest kind of [challenge] from the beginning. (Parent 4)

It was also important that the personnel/staff (e.g., in schools) could consult a competence center for advice and to learn more about BBS:

> I think it's important to have a close collaboration with the [competence center] … that disorder being so rare, we don't know of any other children having the same [disorder].…I think it's important … that the teacher can get in touch or take some courses. (Parent 1)

4. Discussion

The participants in the current study emphasized that the early period with the child was demanding. They found insufficient knowledge about BBS throughout the Norwegian support system, and they described several challenges associated with having a child with a rare disorder. These challenges included uncertainty about their child's condition, long visiting hours at the hospital, and lack of knowledge about the condition in the support system. In addition, the parents experienced a lack of understanding from professionals in kindergarten and schools and from the support system regarding their concerns.

4.1. Uncertainty and lack of knowledge resulting in parental stress

The important challenges identified by the parents included uncertainty about their child's condition, time-consuming visits to the hospital, and lack of knowledge about BBS. Moreover, several parents described the difficulties associated with poor dissemination of information about the support services and the welfare rights that they were entitled to. The difficulties experienced by these parents contributed to their emotional reactions, including shock, frustration, and grief. Our findings indicated several stressors, as mentioned above, associated with having a child with BBS in the Norwegian context. Our finding is consistent with findings confirming the constant stress experienced by parents of children with rare disorders (Cousino and Hazen, 2013; Pelentsov et al., 2015; Dellve et al., 2006; Senger et al., 2016).

The participants experienced the insufficiency of knowledge about BBS throughout the Norwegian support system, and this lack led to delays in getting an initial diagnosis. The similarities between BBS and other genetic conditions, such as Meckel–Gruber syndrome, can make the initial diagnosis of BBS challenging (Karmous-Benailly et al., 2005). However, the confusion between BBS and Meckel–Gruber syndrome is not necessarily due to a lack of knowledge, although the similarities shared by these conditions (e.g., polydactyly and renal anomalies) contribute in the difficulty in distinguishing one from the other (Karmous-Benailly et al., 2005). Nevertheless, these similarities complicate the diagnosis of BBS and generates stressful experiences for the caregivers. This finding is consistent with that of previous research on rare disorders, demonstrating that a lack of knowledge can create stressful experiences for parents (Cousino and Hazen, 2013; Pelentsov et al., 2015; Goodwin et al., 2017) and can cause delays in the diagnosis of rare conditions (Iannello et al., 2002).

Stressful experiences associated with the uncertainties about a child's condition, such as those reported by the participants, impact the parents' mental health. For example, McConkie-Rosell et al. (2018) pointed out how parents of children with undiagnosed chronic conditions manifested mild to moderate depression and anxiety. In addition, delayed diagnosis can influence medical treatment and may lead to unnecessary assessments, further distressing the parents (Anderson et al., 2013). Some complications associated with BBS (e.g., retinal dystrophy) develop slowly during the first years of the child's life, leading to the extensive variations in the development of these children (Beales et al., 1999; Forsythe and Beales, 2013). As such, slowly

emerging symptoms and developmental variations may create uncertainty, which is frequently experienced by parents of children with rare and congenital disorders; such uncertainty often leave these parents in states of distress (Hinton and Kirk, 2017; Goodwin et al., 2017). In addition, parents of a child with a progressive genetic disorder, such as retinal dystrophy in BBS, may experience anticipatory loss (Rolland, 1990).

In this study, the parents' missed courtesy and understanding from kindergarten/school professionals, from their support system, and from their family members. This pattern is consistent with the findings of recent studies, which showed that scarce knowledge about rare disorders may lead to inadequate public support (Kesselheim et al., 2015; Yanes et al., 2017; von der Lippe et al., 2017), and this pattern can have serious implications for the support and aid offered by a support system and by other social services for both those affected and their parents. Many parents feel that they themselves have gained more knowledge about their child's rare condition than a general practitioner (Kirk and Glendinning, 2004; Kesselheim et al., 2015; Pelentsov et al., 2015), and their experiences with their support system differ from those of parents of children with more common conditions. For instance, in a study by Kesselheim et al. (2015), parents experienced difficulty finding qualified specialists and treatments available for their children's rare conditions. As frequently reported by the parents in our study, parents also feel stigmatized by others because of their child's condition; they have experienced being judged for their child's difficulties (Goodwin et al., 2017; Hamlington et al., 2015).

Several parents in the present study described the challenges stemming from their child's rigidity. Previous studies corroborating these findings revealed the prevalent behavioral problems and the preferences for routines among individuals with BBS (Beales et al., 1999; Khan et al., 2016). This behavioral pattern may be especially unfavorable for children and adolescents in terms of managing friendships with peers, which are important for one's quality of life in the long run (Sigstad, 2016). Parents' may also find it difficult to manage satisfactory relationships with their own network of friends and family and to find dedicated personal time. Worse, this may lead to a poor quality of life that affects the parents' social functioning and their relationships with family, friends, and colleagues (Lazarus and Folkman, 1984).

4.2. Experiences that promote coping

The participants emphasized that their workplace was an arena that enabled fellowship and participation in a community network. Their workplace promoted emotion-focused coping strategies, such as seeking social support to reduce emotional distress. Previous findings demonstrated the being beneficial of these strategies and highlighted the crucial role of social support (e.g., marital relationships, family, friends, and support groups) for parents of children with chronic conditions and its effect on their social functioning (Lazarus and Folkman, 1984; Yanes et al., 2017; Kelso et al., 2005). Moreover, several studies emphasized the importance of family members as a source of support, as well as highlighted their contributions in increased coping and in relationships between siblings (Pelentsov et al., 2015; Fletcher et al., 2010; Patterson et al., 2004).

Several parents in the present study stated the importance of openness about their child's condition and how it contributed in promoting positive emotions. Consultation with other caregivers of children with similar challenges contributed to an increased insight into and understanding of their child's difficulties, reminding them that these difficulties were not caused by poor nurturing. Similar findings from another study also demonstrated that openness about a child's condition with family members, healthcare professionals, and others can sustain positivity and hope in the parents and at the same creates stronger social support (Rafferty et al., 2019). Moreover, parents can better adapt to their situations by seeking support and advice from other caregivers through problem-focused coping (Folkman, 2013). This beneficial interaction was revealed by one study wherein parents of children with different chronic conditions received emotional support and information through their interaction with other parents who are in similar situations, and they learn from one another's experiences (Rafferty et al., 2019). Openness and support-seeking may also engage parents in the process of normalization, which involves the acknowledgment and acceptance of having a child with a rare disorder (Knafl and Deatrick, 1986; Cohen and Biesecker, 2010).

4.3. The support system

The main challenges for the participants revolved around the lack of knowledge about their child's disorder in various microsystems (e.g., kindergarten, school, and healthcare system), in addition to the inefficient communication between these institutions in their mesosystem (e.g., relationships between family and school staff or healthcare professionals). They highlighted the importance of collaborating early on with specialized professionals who are equipped with relevant and updated knowledge about rare disorders. This collaboration is especially important because seeking and coordinating for care and support services were some of the main challenges for the parents. The unavailability of professionals to consult is very unfortunate because these parents carry an enormous responsibility for their child's physical and emotional well-being, often at the expense of their own health. As such, competence centers, which offer information about rare disorders through counselling and seminars, seem to be of great importance for the parents.

Existing connections between microsystems, referred to as the mesosystem (Bronfenbrenner, 1979), are likely to contribute in how parents are helped by their support systems. For example, Anderson (2009) found that the mothers of children with chronic conditions experienced insufficient communication between their healthcare professionals and school personnel. Poor communication between the microsystems may also have a negative impact on the transition from kindergarten to first grade of children with different chronic conditions (Siddiqua and Janus, 2017), further distressing their caregivers. Therefore, improved communication among parents, schools, and support systems (i.e., the mesosystem) will likely enhance each microsystem's understanding of a child's condition (Gallo et al., 2008).

4.4. Limitations and strengths of this study

This study was limited by its sample size. The number of parents who were invited to participate was restricted in the first place, and this number was even reduced because some parents did not consent to participate. One limitation of this study is that we did not send a reminder to the prospective participants. As presented in this study, taking care of a child with BBS is demanding, which may have hindered some of the parents from participating in research projects. Another limitation of this study involves the use of telephone interviews, which may have affected the quality of the study. However, telephone interviews are more convenient for parents residing in remote locations, and this method enabled the researcher to establish a personal connection with the parents (Heath et al., 2018). Another possible weakness is that parents were asked whether they experienced having any challenges of having a child with BBS. This approach may have created a bias; however, the intention of this study was to investigate any challenges, which, based on our clinical experience, prompt many parents of children with BBS to contact a competence center. One strength of this study is that the findings are not only applicable to the parents of children with BBS; they may also be applicable to caregivers of children with other rare genetic and chronic conditions.

5. Conclusions

The results of this study highlighted the need for more knowledge about the experiences of parents with a child with BBS. Moreover, the findings demonstrated how the microsystems and the interplay between them in the mesosystem significantly affect the support offered to caregivers. Professionals and other individuals at the microsystem level should be attentive to the possibility of parents experiencing grief and guilt toward their child's condition. These parents may also be feeling despair because they do not know how to deal with such condition. Therefore, it is necessary to deepen the knowledge about BBS at all levels of the support system (i.e., kindergarten, school, and healthcare system). Furthermore, it is important for parents to collaborate with a competence center as early as possible. A competence center can provide both parents and other institutions with specialized knowledge related to rare disorders, and it can provide support through counselling and seminars. More knowledge is needed on how to strengthen the relationships of the parental microsystem as early as possible in order to promote better support for the parents of children with BBS and other rare disorders. This approach will in turn provide better support for children with rare disorders and their families.

CRediT authorship contribution statement

Deniz Zelihić: Data curation, Formal analysis, Writing - original draft, Writing - review & editing. **Finn R. Hjardemaal:** Formal analysis, Writing - original draft, Supervision. **Charlotte von der Lippe:** Conceptualization, Formal analysis, Resources, Writing - original draft, Writing - review & editing, Supervision.

Declaration of competing interest

The authors report no potential conflicts of interest.

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Appendix A. Supplementary data

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