Brief Report

Shaping and managing the course of a child’s disease: Parental experiences with osteogenesis imperfecta

Maman Joyce Dogba, M.D., Ph.D. a,*, Frank Rauch, M.D. a, Ghislaine Tre, M.D., Ph.D. b, Francis H. Glorieux, M.D., Ph.D. a, and Christophe Bedos, D.M.D., Ph.D. c

a Shriners Hospital for Children, 1529 Cedar Avenue, Montreal, QC, Canada H3G 1A6
b Center of Laval University Research Hospital 10, rue de l’Espinay, QC, Canada G1L 3L5
c Faculty of Dentistry, McGill University, 3550 University Street, Montreal, QC, Canada H3A 2A7

Abstract

Background: Osteogenesis imperfecta (OI), a rare genetic disease, causes increased bone fragility. The course of childhood chronic conditions particularly rare genetic diseases can be modified by both child and parents. However with limited research, shaping and managing that process is not well understood.

Objective: Here we examine how parents of children with severe and mild OI have shaped and managed the condition of their child over time. Our goal is to provide a in-depth understanding of parental responses to OI.

Methods: This study was carried out in a pediatric orthopedic hospital located in Montreal, Canada. Using the principles of interpretative description, we conducted semi-structured interviews with 48 parents of children diagnosed with OI.

Results: We found that parental responses to their child’ disease are constituted by their feelings and their actions. These responses changed over time. We can report four successive phases: an initial reaction, acceptance, normalization and passing the baton. Each stage affected subsequent stages. Every stage was influenced by the severity of OI, parents’ individual characteristics, their day-to-day experiences and the entourage.

Conclusion: Our study contributes to increased understanding of parental responses to OI and to improved parental responses and ultimately the child’s coping. © 2014 Elsevier Inc. All rights reserved.

Keywords: Osteogenesis imperfecta; Parental responses; Children; Illness and rare disease; Research; Qualitative; Psychosocial issues; Genetics; North America

The trajectory model of chronic illnesses postulates that the course of chronic conditions including rare genetic diseases can be influenced by interactions, persons and their environments. Biomedical or psychosocial interventions, known as shaping and managing, interact with the natural history of the disease to alter the course of chronic conditions over time.

The course of rare genetic diseases can be modified by coping strategies of both the child and the parents. Parental coping is closely related to child development. Higher family functioning, higher self-esteem and a greater sense of parental control correlate with better psychological and physical health in children with chronic conditions, both directly and indirectly through better social support and stress management. While parental coping in rare genetic diseases is receiving increased attention, longitudinal design or life course approaches are limited. We therefore aimed at an in-depth understanding of the progress of parental responses to a child’s disease over time with the aim of informing the development of support interventions.

Osteogenesis imperfecta (OI), a rare genetic disease of increased bone fragility, affects approximately 1 in 10,000 newborns. Apart from fractures, short stature, limb and spine deformities and restricted mobility, extraskeletal symptoms like blue sclera, abnormalities of the teeth and hearing impairment can be present. Disease severity
varies widely, which has led to a classification that currently comprises seven clinically defined types (OI types I—VII), and an expanding number of genetically defined OI types.\textsuperscript{11,12} The occurrence of fractures in young children may lead to a suspicion of child abuse and neglect, especially in children with milder forms of OI.\textsuperscript{13,14}

The psychosocial consequences of OI can include social isolation, long delays for definitive diagnosis and difficult access to knowledgeable health professionals.\textsuperscript{15—23} The parental responses over time are not fully understood. The severity of the disease as a determinant of parental responses has been insufficiently accounted for.\textsuperscript{11,18} To address these knowledge gaps, the present study provides an in-depth understanding of how parents of children with severe and mild OI can alter shape and manage the condition of their child over time.

Methods

The study was performed at a pediatric orthopedic hospital in Montreal, Canada, from July 2011 to May 2013, when approximately 400 children and adolescents with OI, mainly from Canada, the United States, Latin America and Europe, were actively followed. The study is an interpretative description which is a qualitative research design that examines a phenomenon poorly understood in a clinical setting by identifying explanatory patterns among individuals.\textsuperscript{24,25} Ethical approval was obtained from the Institutional Review Board of McGill University.

Using purposeful maximum variation sampling, we recruited parents of children who varied widely regarding disease severity and trajectory.\textsuperscript{26,27} Variability in disease trajectory was accounted for by the recruitment of parents near the time when their children were diagnosed with OI, when the incidence of fractures is typically high, and then further along the timeline, when symptoms are usually more stable. To ensure variation in the sickness trajectory, we invited both parents who encouraged their children to functional independence, and parents who, often for fear of fractures, tended to discourage their children from the physical activity required for functional independence. Illness trajectory was accounted for by including both parents with a negative discourse around their experiences with OI and parents with a more positive outlook as revealed in previous discussions with hospital staff.

We invited parents of children diagnosed with OI and followed at our institution if they were fluent in English or French. A total of 48 parents (34 mothers and 14 fathers) of 39 children were recruited (Additional file 1 www.disabilityandhealthjnl.com). Face-to-face semi-structured interviews were conducted using an interview grid developed by the research team (Additional file 2 www.disabilityandhealthjnl.com). The 30—60 min interviews were digitally recorded, transcribed verbatim and stripped of identifying information. Between 3 and 12 months after the initial interviews, parents were met again to check for accuracy of their input and to hear our core findings.

Using NVivo 10 (QSR International) we performed a semantic thematic analysis.\textsuperscript{28} Two authors independently compared their line-by-line transcript coding. The percentage of agreement between codes was 93%—100%. Coding disagreement was resolved by discussion. As patterns became more apparent, dominant categories inside the corpus were identified and considered themes. Coding ended by establishing and checking relationships between categories by generating matrices that systematically compared themes across different groups of parents (parents of children with severe OI versus mild OI, parents of young versus older children, parents with no family history of OI versus with a family history).\textsuperscript{29}

Results

We conceptualized parental responses as a series of feelings and actions chronologically organized into four phases: Initial reaction, acceptance, normalization, and “passing the baton.” The phases respectively match the following life developmental stages: neonatal period, childhood and socialization in daycare and school, adolescence and early adulthood. Elements likely to influence parental responses were identified and grouped into four categories: Severity of the disease, day-to-day experiences of parents, parents’ personal characteristics and parents’ entourage. Table 1 outlines parents’ feelings and actions at each of the four stages and influencing elements. There are commonalities and differences across the four stages. Table 2 provides selected illustrative quotes.

Phase I: Initial reaction

This begins with the diagnosis, when parents focus on the medical characteristics of OI.

How parents felt

Most parents of severely affected children were devastated by the diagnosis. However, parents who waited a long time before obtaining a definite diagnosis generally also felt relieved, as many of them had been suspected of child abuse (Quotation 1). Some reported anger because of the genetic origin of the disease and guilt for having “caused” the suffering to their child. Most importantly, many parents described having different, sometimes contradictory feelings at the same time: devastation and denial, and shock and relief (Quotation 2).

What parents did

They actively sought information about the consequences of OI, its prognosis and cure through the internet and social media (e.g., OI parents’ group on Facebook).
Elements likely to influence parental responses at phase 1

The severity of the disease and day-to-day experiences. Parental responses were influenced by circumstances of the diagnosis (e.g., delay before confirmation or quick diagnosis), prior knowledge of OI and the communication of the diagnosis to parents by health professionals (Quotation 3, 4).

Personal characteristics and environment. Parents’ temperaments and personal life situations sometimes explained their reactions in Phase I. For example, the diagnosis of OI interrupted the professional and family plans of many. However, the initial reaction depended on the personal environment as revealed by embarrassing situations when their child became an “attraction,” because of short height and deformities (Quotation 5).

Phase II: period of acceptance

As this phase coincided with the socialization stage of daycare to entering school, parents focused on the social life of their families and the sickness trajectory.
sequences added to the burden of care depending on acceptance of the disease. Parents’ perception of these consequences (e.g., loss and social isolation) was an important step to the psychosocial consequences of the diagnosis (e.g., job adjustments to a type of hope and we would have liked to be given a chance to hope. 1. They can live, be happy and have a productive life. Yes. And it is important to let parents see that they have hope. She communicates, she can talk to me. She can laugh. What else do you want from your child? 2. We were told that she will not survive, we were told that there was an 80% chance that she will not survive and nobody discussed with us but if she survived. Nobody discussed what we’re going to do. Nobody discussed what the treatment options were. It was all about the negative part… As a parent, you are searching for a type of hope and we would have liked to be given a chance to hope. 1. Most often, parents reported trying to prepare their child for a better future either by getting involved with the school to ensure individual temperament and how much control parents could exert on life events. This was illustrated by the report of mothers who reduced their professional activities to care for their children. While some came to be satisfied with their decisions, others were frustrated for losing control over their lives by choosing to be the “stay-home parent” (Quotation 8, 9).

The surroundings of families of severe versus mild disease burden differed. Most parents of severely affected children were pitied by friends and neighbors, whereas parents of mildly affected children were shocked that others questioned the diagnosis because of the lack of deformities and normal height (Quotation 10).

Phase III: normalization
How parents felt
Most parents redefined what their “new normal” would be and educated their children accordingly. They stressed that previous phases influenced the normalization stage. Among parents who stressed being able to focus on the sickness trajectory of OI, some defined their children as normal with one added challenge, that of living with OI (Quotation 11). Other parents who focused on the disease trajectory described the course of OI as periods of ups and downs punctuated by the occurrence of fractures and other complications (Quotation 12).

What parents did
Parents reported trying to prepare their child for a better future either by getting involved with the school to ensure


better integration, or by participating in research to improve care options for OI. Some regretted becoming overprotective; however most were confident about being able to prepare their child for independent living.

**Elements likely to influence parental responses**

The ability of the child to cope with his condition was a core component to parental responses and the resilience of their child was often mentioned as a great inspiration to many parents.

**Personal characteristics and the environment**

Parents tended to react according to their temperament and their sources of support were mainly their own inner resources, church, family and hospitals.

**Phase IV: passing the baton**

This final stage, corresponding to adolescence and young adulthood is called “passing the baton” to the child with OI. Here, when severely affected patients transition from the pediatric setting to adult health care institutions, parents expect their child to manage his own life. Usually, patients with mild OI will live independently. For severely affected patients, parents adopted either a companionship or managerial stance depending on the child’s level of autonomy.

**How parents felt**

Most parents reported feeling a sense of achievement once their children have a successful professional and family life. Meanwhile, parents with severely affected children experienced worry about the future (Quotation 13).

**What parents said they would do**

By research design, none of the children had achieved complete independence and were accompanied by their parents during the study visit. Therefore parents speculated on phase IV. However, parents emphasized that starting from the normalization stage they prepared their children for independent management of their own health. Parents were most concerned about their child’s physical setting and their emotional life. During their emotional life, the genetic nature of OI will resurface. When the person with OI chooses to have a child, the parents (becoming grandparents) felt morally bound to help their children face the challenges (Quotation 14).

Fig. 1, a schematic representation of our findings summarizes the four phases, with each influenced by the previous and the next. Elements likely to influence each stage are outlined.

**Discussion**

In this study we assessed how parents of children with OI react to and shape the condition of their child over time and confirmed huge differences between parental responses to OI in a child with mild OI and severe OI. In all cases, in Phase I a mosaic of concurrent or sequential contradictory feelings was reported, which subsequently crystallized into a clearer pattern. While parents focused on the diagnosis in
the initial phase, they entered into a new routine at the acceptance and normalization phases before experiencing worry again in Phase IV. In Phase I, parents mainly searched for information about OI and the search of treatment centers, whereas Phase II was characterized by parental assessment and adaptation to the consequences of the diagnosis. Phases III and IV were used to prepare the child for a better future. At all phases, the severity of the disease and the reaction of the familial environment were influential. However, the trajectory focus differed depending on the phases. At Phase I, parents focused on the disease trajectory while physical impairments were obvious. The focus at Phase II was the social life and sickness trajectory while most parents were able to reflect on their experience with OI (illness trajectory) in Phase III.

Diagnostic uncertainty and a prolonged period of waiting for a definite diagnosis can be difficult to manage for parents of children with a rare genetic disease, explaining why some can feel relieved. In the case of OI, parental relief was more related to the prior suspicion of child abuse and neglect that threatens all personal and societal definitions of good parenting.

Our conceptualization of the parental responses to OI could help to design adequate interventions for patients and their families over the life course. For example, medical staff should be sensitized to the lifelong influence of the experience of diagnosis on latter parental responses. Thus health professionals should be trained for improved communication of the diagnosis to patients. This might include adopting an interprofessional approach to communicating the diagnosis with psychologists, genetic counselors, social workers and other professionals in the care team.

Our findings also advocate for a tailored parental support by early parental focus on the social trajectory rather than the physical impairments. While some coping strategies such as positive thinking are to be assessed by professionals, this study reveals that support from friends and families are likely to positively influence parental coping. Besides, as the child’s coping influence parental responses at Phase III, efforts should also be made to obtain better child patient coping, emphasizing the need for a family-centered approach to care, especially in the case of rare genetic diseases.

Despite the theoretical understanding and practical implications of our study, several limitations could impede application to other contexts. First, we have only considered one genetic rare disease that has specific features: OI is non-progressive and in most cases non-lethal and does not affect cognitive development. Studies about other heritable skeletal diseases with a non-progressive course such as achondroplasia or hypophosphatemic rickets could gain insight from our findings. However further investigations are needed to test the relevance of our findings to rare genetic diseases that have developmental impairments. Second, by research design, Phase IV was insufficiently documented in this study and needs to be addressed in parents of young adults at different stages of life: making reproductive choices, professional integration and independent living.

Conclusion

Parental feelings and actions to the diagnosis of OI in their child shape and manage the trajectory of the disease over time. This shaping and managing process is very complex and non-linear. Parental and child responses are inter-related and dependent on their temperaments, the familial environment, and the severity of the disease. Our findings point to the need for a greater understanding of parental responses in other settings. However, practical implications were derived such as the need for a tailored life-span and family-centered psychosocial support for families; and increased awareness and training among health professionals about the complexity of these parental experiences.

Acknowledgments

We are very grateful to the families who participated in this study. We acknowledge the assistance of Michaela Durigova, Kathleen Montpetit, Trudy Wong, Colette Désilets, Susan Lempriere and Judith Kashul in conducting the study. We also want to thank two anonymous reviewers for providing suggestions and constructive criticism.

Supplementary data

Supplementary data related to this article can be found at http://dx.doi.org/10.1016/j.dhjo.2014.03.002

References