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## Original Article

# Living with Osteogenesis Imperfecta: A qualitative study exploring experiences and psychosocial impact from the perspective of patients, parents and professionals

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## ABSTRACT

**Background:** Osteogenesis Imperfecta (OI) is a rare genetic condition characterised by increased bone fragility. Recurrent fractures, pain and fatigue have a considerable impact on many aspects of the life of a person affected with OI and their families.

**Objective:** To improve our understanding of the impact of OI on the daily lives of individuals and families and consider how the condition is managed so that support needs can be better addressed.

**Methods:** Semi-structured qualitative interviews (n = 56) were conducted with adults affected with OI, with (n = 9) and without children (n = 8), parents of children affected with OI (n = 8), health professionals (n = 29) and patient advocates (n = 2). Interviews were digitally recorded, transcribed verbatim and analysed using thematic analysis.

**Results:** Three overarching themes are described: OI is not just a physical condition, parenting and family functioning and managing the condition. Fractures, chronic pain and tiredness impact on daily life and emotional well-being. For parents with OI, pain, tiredness and mobility issues can limit interactions and activities with their children. Specialist paediatric health services for OI were highly valued. The need for more emotional support and improved coordination of adult health services was highlighted.

**Conclusions:** Our findings allow a better understanding of the day-to-day experiences of individuals and families affected with OI. Supporting emotional well-being needs greater attention from policy makers and researchers. Improvements to the coordination of health services for adults with OI are needed and an in-depth exploration of young people's support needs is warranted with research focused on support through the teenage years.

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## Introduction

Osteogenesis Imperfecta (OI) is a rare genetic bone fragility disorder characterised by susceptibility to fractures and variably

associated with skeletal deformity, small stature, joint hypermobility, dentinogenesis imperfecta and hearing impairment.<sup>1,2</sup> OI is a clinically and genetically heterogeneous condition that affects approximately 1 in 10,000 - 20,000 births.<sup>1-3</sup> There are autosomal dominant, autosomal recessive and X-linked forms of the condition and causal variants can occur in a large number of different genes. In 85–90% of cases, OI is caused by variants in the genes that encode type I collagen (*COL1A1* and *COL1A2*).<sup>4</sup> OI varies in severity and classification into types has traditionally been used to define OI

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as mild (type 1), moderate (type 4), severe (type 3) or lethal at birth or soon after (type 2).<sup>5</sup> More recently, other rare OI types have been described and new genetic classifications have been considered.<sup>4</sup> Notably, there is clinical variation within the OI types and family members with the same genetic variant can be affected in different ways.<sup>6</sup>

For patients with OI, the occurrence of fractures declines with age<sup>7</sup> but adults do experience physical dysfunction with varying severity and are often affected by chronic pain and fatigue.<sup>8</sup> There is no cure for OI and management requires a multi-disciplinary approach that aims to reduce fractures and deformity, provide relief from pain and support mobility.<sup>6,9</sup> Management includes non-surgical approaches, such as physical and occupational therapy, medical treatments, such as bisphosphonates, and surgery, such as intramedullary rodding.<sup>6</sup>

Recent reviews have highlighted the many practical and emotional challenges OI poses for individuals and families.<sup>10–12</sup> Qualitative research considering the psychosocial impacts of OI on children and families is growing<sup>13–17</sup> but few studies have considered the experiences of adults affected with OI,<sup>18,19</sup> and a notable gap in our understanding is the experiences of parents who have OI. Here we describe our research considering the day-to-day experiences of individuals and families affected by OI from the perspective of patients, parents and professionals. Our aim was to improve our understanding of the impact of OI on the daily lives of individuals and families, how the condition is managed and how support needs can be better addressed.

## Materials and methods

### Study design

We used a qualitative descriptive design with data collection from one-to-one semi-structured interviews. NHS Research Ethics Committee approval was obtained (16/NS/0084).

### Context

The BOOST Brittle Bones Before Birth (BOOSTB4) programme is examining the safety and efficacy of stem cell administration for severe OI.<sup>20</sup> The research described here is part of a wider study whose primary aim was to examine stakeholder views of stem cell administration for severe OI. Qualitative interviews with patients, parents and professionals explored four topic areas: 1. Experiences of living with OI or working with families affected by OI. 2. Views on current treatments for OI. 3. Views on using stem cells to treat OI. 4. Support and information needs for decision-making about stem cells. Findings from topics 3 and 4 have been reported separately.<sup>21</sup>

### Recruitment and data collection

As previously described,<sup>21</sup> participants included adults affected with OI, with and without children, and parents of children affected with OI who did not have OI themselves (“patients and parents”), and health professionals and patient advocates from relevant patient support groups (“professionals”). Patients and parents were recruited through OI specialist services (four paediatric, one adult) and an advertisement on the Brittle Bone Society (BBS) (UK support group) website and Facebook page. They were asked to contact MH if interested in participating or they could complete a “consent to contact” form if attending a clinic. Patients and parents were offered a £10 gift voucher in appreciation of their time. Professionals were identified by the research team and invited to participate via an email from MH. Semi-structured interviews were conducted face-to-face or via telephone by MH.

### Data analysis

Interviews were digitally recorded, transcribed verbatim and anonymised using pseudonyms. Data were analysed following the principles of thematic analysis.<sup>22</sup> To support rigour, analysis was undertaken by multiple researchers (MH, JH, MS and CL). MH, JH and CL are experienced qualitative researchers, and provided training in qualitative research methods to MS. To address our aim of exploring day-to-day experiences of living with OI, inductive and deductive approaches to analysis were used.<sup>23</sup> Themes were drawn from our aims, topic guides and the existing literature and emerged from the empirical data. NVivo10 (QSR International, Pty Ltd, Australia) was used to facilitate analysis. Transcripts were coded into meaningful units of text, which were then grouped into broader categories. Data from the “patient and parent” and “professional” groups were initially analysed separately. MH coded all professional transcripts and JH independently coded a subset (10%). MS and JH independently coded all patient and parent transcripts. Emergent themes and sub-themes were reviewed and revised by MH, JH, MS and CL. Themes from the two data sets were integrated into a single narrative by MH.

## Results

Interviews were conducted with 56 participants; 25 parents and patients and 31 professionals. Response rates for patients and parents could not be determined as recruitment combined invitations and advertisements. The response rate for professionals was 69%, 45 invited and 31 accepted. Interviews were conducted between September 30, 2016 and February 15, 2018, 19 in person and 36 by phone. Interviews lasted between 15 and 56 min (median = 30 min). Participant characteristics are summarised in Table 1.

### *OI is not just a physical condition*

#### *OI impacts on all aspects of daily life*

The impact of the commonly described physical issues of recurrent fractures, reduced mobility, general pain, back pain from scoliosis, and tiredness echo through daily life. Medical appointments, including routine visits and responding to fractures, are common and participants described managing life with OI as “like a full-time job in itself”. To lower fracture risk, the types of activities undertaken are sometimes restricted and additional planning is needed (Table 2: Q1). Tiredness and pain could also restrict activities (Table 2: Q2). Parents and patients reported adapting their homes or moving to homes with improved accessibility to reduce fracture risks.

#### *Frequent interruptions to school, further education and work*

Participants described repeated interruptions to school and further education due to medical appointments, fractures or recovering from surgery (Table 2: Q3). This had both academic and social implications and it was noted that children “lose a lot of time with play with their peers”. OI also impacts work and adults with OI described working around their pain (Table 2: Q4), not being physically able to do certain jobs, needing to work part-time or not being able to work at all due to fractures, pain and tiredness (Table 2: Q5). Some parents chose not to work outside the home, worked part-time or worked remotely to allow time to care for their child and attend medical appointments.

#### *Life with OI can carry an emotional toll*

Participants frequently spoke about the emotional impact of OI and felt strongly that OI was “not just a physical condition”. Parents

**Table 1**  
Participant demographics.

<b>Parents (N = 25)</b>	
<b>Participant group</b>	
Adult with OI (no children)	8
Parent affected with OI	9
Parent not affected with OI	8
<b>Gender</b>	
Female	21
Male	4
<b>Age</b>	
17-30	9
31-40	6
41-50	8
51-60	1
61+	1
<b>Self-reported ethnicity</b>	
White British	20
Asian British	3
Mixed	2
<b>Highest educational qualification</b>	
High School	5
Degree or equivalent	20
<b>Self-described OI type of self or child</b>	
Type 1 or Mild	16
Type 4 or Moderate	1
Type 3 or Severe	7
<b>Number of children with OI</b>	
<b>Parent affected with OI</b>	
No children with OI	1
One child with OI	3
Two children with OI	4
Three children with OI	1
<b>Parent not affected with OI</b>	
One child with OI	7
Two children with OI	1
<b>Professionals (N = 31)</b>	
<b>Gender</b>	
Female	23
Male	8
<b>Age</b>	
21-30	2
31-40	8
41-50	10
51-60	11
<b>Current role</b>	
<b>OI specialist paediatric team</b>	
Nurse specialist	18
Occupational therapist	3
Physiotherapist	2
Endocrinologist/metabolic bone specialist	5
Psychologist	7
<b>OI specialist adult team</b>	
Nurse specialist	3
Rheumatologist	1
<b>Patient advocate</b>	
Genetics & Fetal medicine	2
Clinical geneticist	8
Genetic counsellor	3
Fetal medicine specialist	2
<b>Years working with families affected with OI</b>	
≤5	3
6-15	13
16-25	11
<b>Current practice location</b>	
England (North)	7
England (Midlands)	4
England (London)	5
England (South)	12
Scotland	8
	2

Note: This table has been adapted for this report from the demographic data tables in Hill et al. 2019.<sup>21</sup>

spoke of their “constant fear of fracturing”. The need to be always aware of possible risks was described as “mentally tiring”. Parents also noted feelings of “guilt” and distress when their child was hurt and feeling “frustrated” or “upset” when children were excluded

from care settings, such as nursery, or left out of social activities (Table 2: Q6). Adults with OI described being cautious to avoid fractures with one participant commenting; “I’m always careful and it’s always in the back of my mind” (*Rameen, adult with OI*). Two

**Table 2**  
OI is not just a physical condition: subthemes and illustrative quotes.

**OI Impacts on all aspects of life**

Q1: "they've just had their year six trip, you have to kind of go through all the activities with a fine-tooth comb, get on the phone with all the people in the residential place and assure them that it's ok to have someone with brittle bones there ... talk to them about what that means. I mean it's a part of our life actually but when you sort of stop and think and speak about it, you realise how different it must be to someone who doesn't do it." *Ameenah, adult with OI and parent of a child with OI*

Q2: "I do get really bad aches and pains and it stops me from doing things or going out." *Rameen, adult with OI*

**Frequent interruptions to school, further education and work**

Q3: "You know, feeling tired all the time, having less energy, being in pain, missing school because of hospital. And if I had, like, my leg in traction for six weeks, that's six weeks off." *Mia, adult with OI*

Q4: "So just sort of taught myself to just sort of deal with [the pain], that's the normality of it, but it is hard because my job ... it's really demanding. Not only sort of emotionally but physically as well." *Lily, adult with OI*

Q5: "I was working as a doctor and then was getting multiple stress fractures from being on my feet and the physical side of things, so it's, it's led to quite a lot of changes in my life." *Celeste, adult with OI and parent of a child with OI*

**Life with OI can carry an emotional toll**

Q6: "At the moment we're having problems getting [child's name] to nursery and it's breaking my heart, because I feel like, well, she should be treated like a normal child." *Olivia, adult with OI and parent of a child with OI*

Q7: "For about 4 months after the injury and discharge, I was still having issues, grappling with that, being in my chair and going out again, I didn't feel safe, I didn't feel comfortable and just getting that emotional support was so hard." *Nathan, adult with OI*

**The emotional impact of feeling different can be heightened for young people and is experienced by adults**

Q8: "undoubtedly, teenage years, their body image, when at that age they all want to be the same as someone else so something that's different is going to be an extra challenge for them." *Maddy, professional from an OI specialist team*

Q9: "And I'd be in hospital for a minimum of three months at a time and there's so many things like that can lead to like loneliness ... there's so much research about how people with disabilities are you know, face chronic loneliness ... And not only loneliness, but isolation as well." *Lily, adult with OI*

Q10: "There was always an element of anger and of frustration about it, of being a bit different, being a bit, you know, not being able to join in everything fully and I think that manifested itself in quite a lot of anger and angst above and beyond normal teenage angst." *James, adult with OI*

Q11: "I suppose at eleven he was really struggling with being sort of different ... and I think there was quite a bit of bullying going on." *Abigail, adult with OI and parent of a child with OI*

Q12: "I think you know I've found it really difficult to get work, because of having a condition you know ... unfortunately there's a massive stigma about employing disabled people." *Lily, adult with OI*

**Coping and resilience: adapting to a new normal**

Q13: "he always makes the best of things so he's been great in terms of helping us deal with it because if he doesn't get upset about it then why should we." *Rachel, parent of a child with OI*

Q14: "how they cope with that, that's very much around, you know, personality based and how they deal with situations generally as a family and other demands on that family, so if they've got older siblings or if there are other things in the family that are also particularly stressful." *Piper, professional from an OI specialist team*

**Perceptions of OI: how I see myself and how others see me**

Q15: "I think a lot of people do kind of make it quite light hearted but I think you have to sometimes." *Laura, adult with OI*

Q16: "yeah that's something that I work hard on actually to make sure that they know there are other avenues, you don't have to be sporty. And you can still be sporty and have OI, you can still go off and do everything else, just I don't want you playing contact sports." *Tessa, adult with OI and parent of a child with OI*

Q17: "You're in a wheelchair, so you must have a learning disability. You must be mentally not quite there ... I find you need to get past that barrier of stereotyping and, and people's fear of it, to see that you're a person, that just happens to sit down quite a lot." *Mia, adult with OI*

adults with OI spoke about having experienced bad falls and fractures that meant their "confidence was knocked". They faced additional anxiety around the possibility of another major fracture (Table 2: Q7). For all patients and parents, living with the uncertainty of when a fracture might occur, organisation and planning to avoid risks, multiple fractures, hospital stays and frequent medical appointments could carry an emotional burden.

*The emotional impact of feeling different can be heightened for young people and is experienced by adults*

Teenage years were highlighted as being particularly difficult by both patients and parents as they or their child felt "different" or felt like they didn't "fit in" because of worries about their appearance, such as their height or scars from surgery, or because they couldn't join in certain activities due to the risk of fractures or limitations in mobility. Professionals also pointed out that feeling different could be particularly challenging for teenagers (Table 2: Q8). Several participants noted that some aspects of feeling different may be "harder as a boy" where expectations about taking part in rough and tumble play or contact sports are greater. Feeling different and periods away from school due to fractures could lead to feelings of "isolation", "loneliness" and being "withdrawn" (Table 2: Q9). Feeling different could also result in "anger" and "frustration" (Table 2: Q10). Being "excluded" and "bullying" were additional issues faced at school (Table 2: Q11). Notably, "discrimination" and "stigma" were also described by adults with OI (Table 2: Q12).

*Coping and resilience: adapting to a new normal*

The ability of individuals and families to cope and be resilient was evident. Parents without a family history of OI described the initial "shock" and "panic" of diagnosis and explained how acceptance grew over time as they became used to caring for their child and life became "a bit more normalised". Despite the challenges, patients and parents held a clear aim for life to be "normal" and frequently made comments like "we just get on with it". This perspective could be tempered by feelings of anxiety; one parent said "I do cope with it because we have to but ... you live with the anxiety on a daily basis" (*Louise, parent of a child with OI*). Some parents noted their child's resilience and how their child's temperament helped the family to remain positive (Table 2: Q13). One professional noted that families frequently differed in how well they coped and how this was dependent on personality and the other demands the family faced (Table 2: Q14). Sources of emotional support for participants included friends, family, support groups, health professionals, religious faith and other families affected by OI.

*Perceptions of OI: how I see myself and how others see me*

Some patients spoke about how OI influences how they see themselves and how they interact with others. This was exemplified by one adult with OI who commented that even though living with OI is challenging, "I haven't let it define me". Another adult with OI noted how she used humour with friends to make light of her recurrent fractures (Table 2: Q15). Some patients also noted

that they had focused on social or academic activities over physical activities and several parents spoke about encouraging their child to “find their voice” through activities they enjoyed or were good at that fell within the limits of what they could do physically without putting themselves at risk of fractures (Table 2: Q16).

Several adults with more severe forms of OI who were reliant on a wheelchair or walking aids, described facing stereotyping and dealing with people’s assumption that their physical disability meant that they must also have a learning disability (Table 2: Q17). In contrast, some adults with mild OI spoke of emotional challenges arising from their OI being an “unseen” or “silent” disability as other people don’t know about the condition and it is “difficult to explain to people how it impacts you, because you can’t see what’s wrong with me” (Rameen, *Adult with OI*).

### Parenting and family functioning

#### Starting a family when you have OI

Adults with OI discussed worries about starting a family, including the possibility of passing OI on to their children (Table 3: Q1), the uncertainty around the possible severity of the child’s OI and the impact on partners of having both a partner and a child with OI. Several participants noted that the potential impact of pregnancy and birth on the body was a major factor to consider (Table 3: Q2). Another concern was uncertainty about their own health, mobility and ability to undertake tasks, such as holding the baby (Table 3: Q3).

#### You just adapt around it: parenting when you have OI

Some parents with OI found pregnancy and birth damaging on their bodies, which could make caring for the baby more difficult. All parents with OI described how aches, pains and tiredness impacted on the day-to-day care of their children and placed limitations on interactions and activities (Table 3: Q4). Restrictions in mobility could mean that parents with OI were dependent on others for some elements of caring for their children. One parent

noted a lack of adaptations or equipment to help with parenting and relying on her partner for some tasks (Table 3: Q5). Having a fracture could make caring for a child more challenging and brought more restrictions. Some parents felt “guilty” about these restrictions and worried that they weren’t able to be a “proper parent”. The view that “you just adapt around it” was common (Table 3: Q6). Benefits of being a parent with OI were discussed; such as empathy, understanding and confidence (Table 3: Q7). One parent with OI described wanting to be a “strong role model” for her children and demonstrate that OI doesn’t hold you back.

#### Common challenges for all parents of children with OI

Learning to care for a baby with OI was a major challenge which could leave parents feeling isolated (Table 3: Q8). Some professionals noted that the extended family may be less likely to be involved in caring for a baby with OI (Table 3: Q9). In addition, several parents described finding it hard to step back and allow others to care for the child (Table 3: Q10). New settings and times of transition, such as starting school or reaching puberty can be difficult for families (Table 3: Q11) and additional advocacy and proactive planning was required for new settings. For example, parents sometimes struggled to find a school where they felt their child would be safe, but also included in school activities. Another challenge for many parents was finding a balance between keeping their child safe and not being overprotective (Table 3: Q12). Some parents noted that an important aspect of their role was to help children to be independent and learn that “choices have consequences”.

#### Impacts on family functioning and siblings

Parents discussed the challenges of keeping the family functioning when a child has OI and there are multiple demands on time and attention (Table 3: Q13). They also highlighted the difficulty of “juggling careers and other small children” when hospital stays were required. Parents noted that siblings receive less attention and are frequently asked to be “careful” and “less

**Table 3**  
Parenting and family functioning: subthemes and illustrative quotes.

#### Starting a family when you have OI

Q1: “[my sibling and I] were always very worried that when we had kids that they got brittle bones just because you want the best for your children, you don’t want them to have any kind of illnesses, so it was always in my head.” *Elsie, adult with OI and parent of a child with OI*

Q2: “I think for a lot of these patients making a decision to have a family can be quite a big thing ... they might decide to have one child because they’re testing their own body out as well and they might end up having a caesarean section” *David, professional from an OI specialist team*

Q3: “So I can’t imagine what it would be like for me to be able to hold a baby and move things, I know I’d have lots of support from everyone but sometimes support doesn’t always help does it, you’ve got to be able to do it yourself.” *Fiona, adult with OI*

#### You just adapt around it: parenting when you have OI

Q4: “the main impact would be pain for me, just chronic pain. I can’t walk as far as I used to ... I can drive but and with two children it’s quite difficult to get out and about very much so we are at home more than I would like.” *Celeste, adult with OI and parent of a child with OI*

Q5: “he’ll get the baby out of her cot because I can’t pick her up, change her because I can’t do that either because she’s really into running away at the moment.” *Ameenah, adult with OI and parent of a child with OI*

Q6: “I don’t even go to the hospital anymore for anything like that [fractures in fingers or toes]. So I just strap them together and on I go ... with having two small children and the rest of it, you just adapt around it. And [child’s name] has become very good at putting her own shoes and socks on shall we say from quite an early age.” *Sophie, adult with OI and parent of a child with OI*

Q7: “I remember just feeling quite calm because I knew that I could, I knew what I was doing.” *Tessa, adult with OI and parent of a child with OI*

#### Common challenges for all parents of children with OI

Q8: “I think certainly the newly diagnosed babies, they probably feel a bit like they’re the only person in the world who’s got an OI baby.” *Ishbel, professional from an OI specialist team*

Q9: “my husband, you know, should be allowed to be a dad without me having to be on his back, you know, worrying about those things.” *Olivia, adult with OI and parent of a child with OI*

Q10: “the extended family will then kind of step back more than they probably would have, because again maybe you know fears around how many people are handling a baby” *Piper, professional from an OI specialist team*

Q11: “So be it transitional from the point of they’re moving from one place to another or transitional in terms of their personal development, such as puberty and all big things. So any psychological transition and also physical transition are huge times for OI families.” *Robert, professional from an OI specialist team*

Q12: “I do try not to bubble wrap them because I think it’s quite important for them to have some element of resilience.” *Tessa, adult with OI and parent of a child with OI*

#### Impact on family functioning and siblings

Q13: “I mean with [child’s name] being as delicate as she was, it was, oh it’s just a complete sort of nightmare really, because you were just worrying all the time and you still have to function with every day, you know, with all the other children and you know with everything else.” *Olivia, adult with OI and parent of a child with OI*

boisterous". Parents also noted that siblings often helped and supported the child with OI and one parent commented that living with a brother with OI had made her daughter "a very caring person actually ... she helps him all the time" (*Imogen, parent of a child with OI*).

#### Managing the condition

##### Treatment with bisphosphonates

Many participants reported that, particularly for children, bisphosphonates reduced fractures, pain and tiredness and improved mobility. For some parents and patients bisphosphonates had been "life changing". Physical improvements had wider positive implications, as more time spent in school, and with friends and family became possible (Table 4: Q1); one professional noted that improvements in response to bisphosphonates could have an emotional impact on parents, bringing "relief and a kind of hope for the future that their lives are going to be a bit better than they'd first feared" (*Elise, patient advocate*).

For many parents, a major ongoing issue with bisphosphonate treatment was their child's dislike of needles, ports and cannulas. Treatments could be "traumatic" and some children feared going to hospital (Table 4: Q2). Treatment frequency could also be a concern if time off school and work, travel, overnight stays or child care were required. Parents were positive about newer types of bisphosphonates that require less frequent treatment regimens and shorter hospital visits. Decisions to start treatment with bisphosphonates were not taken lightly and some participants were wary that the long-term impacts are not known. Bisphosphonate treatment was not always viewed as successful and one patient reported that her pain and bone density had not improved. Professionals noted a lack of knowledge around how well bisphosphonates work for adults, and when and how long to use them (Table 4: Q3).

##### Patients and parents become the experts

Many patients and parents spoke about difficulties arising when dealing with health professionals who were not familiar with OI, including missed fractures and giving incorrect advice regarding treatment (Table 4: Q4). Accordingly, many patients and parents spoke about how "when you have a rare condition, you have to

become the expert" and how they had learnt about their own/their child's bodies and pain thresholds so they knew themselves when a fracture had occurred (Table 4: Q5).

##### Clinical support from health services

Specialist paediatric services were highly regarded and multi-disciplinary support was valued. Parents appreciated having a specialist team who could be readily contacted for support and advice about care, and who would go out and speak to schools. Consistency of care was important and one young adult with OI described the professionals in her care team as being like a "little family". Access to paediatric occupational therapy, physiotherapy and correct equipment was said to be more difficult outside of specialist centres and could be location dependent (Table 4: Q6). It was also noted that it can be difficult to access emotional and psychology support in some areas.

There was substantial concern about the services for adults not being centralised and adults with OI spoke of having to "fight" to get an appointment or sustain long waits for services, such as physiotherapy (Table 4: Q7). As one adult with OI commented, "there's nothing for adults. It's almost like when you hit 18, you don't have OI anymore" (*Mia, adult with OI*). Professionals also felt OI services for adults could be "disjointed" and "depends where you live" (Table 4: Q8) but it was also noted there was now "more interest in developing better protocols, better care pathways across the country" (*David, professional from an OI specialist team*).

##### Transition to adult health services

Adults with OI, parents of older children with OI and professionals were asked about the process of transition from paediatric to adult care. Consistency in contacts was felt to help build confidence in the process of transition and having clear links between the paediatric and adult care providers was reassuring. One professional noted that "support for transition remains patchy" and for some adults, transition had been a single appointment with too much to take in (Table 4: Q9). Even when handled with multiple appointments over time, the transition process could be unclear. As one young adult noted "I'm in the transition clinic but I don't know what I'm transitioning to" (*Rameen, adult with OI*).

**Table 4**

Managing the condition: subthemes and illustrative quotes.

##### Treatment with bisphosphonates

Q1: "[Pamidronate] was quite nice because it allowed me to sort of build up my social circles, it allowed me to have more time in school, build up my peer relations which I was lacking when I was younger because I was away so often. So yeah, pamidronate has really helped. Now we know it's nice that I can go a year or two without fracturing and when I do it's not over the longer period of healing as it was before." *Nathan, adult with OI*

Q2: "So the minute we walk through the hospital doors she starts crying ... The last time it was two hours. And that was before she had the cannula put in and then she doesn't like the doctor coming to her. She doesn't like people in uniform ... And then because it's over two days we have to do that again. So it's quite traumatic for her and that's what I'm not looking forward to because it's every, she has it every eight weeks." *Olivia, adult with OI and parent of a child with OI*

Q3: "It's just long term. I mean concerns about long term bisphosphonates ... the difficulty of your patients in their 20s, 30s, 40s – you can't leave somebody on a bisphosphonate forever." *Lesley, professional from an OI specialist team*

##### Patients and parents become the experts

Q4: "Sometimes they didn't quite know how to treat him and he nearly had the wrong operation once." *Liz, parent of a child with OI*

Q5: "I know my body very well. I know when I've got a fracture and they send me home it's like I really know I've fractured my femur, they're like there's nothing on the X-ray and you get a call two days later and they're like oh sorry we missed it." *Ameenah, adult with OI and parent of a child with OI*

##### Clinical support from health services

Q6: "they're not specialists they don't see that many children with OI ... they're not up to speed with stuff as I said they just don't have the specialist equipment actually in terms of even getting a walking frame." *Annie, parent of a child with OI*

Q7: "The whole service and support you get is nowhere near the same as paediatrics. You know, you should be able to have a team. I don't mean a team dedicated to you, just a large team that's working hopefully together for physio, and those kinds of things." *Jade, parent of a child with OI*

Q8: "I think maybe the adult ongoing care falls down a bit ... the adults I've seen with OI are very rarely having follow-up care" *Emma, Genetic Counsellor*

##### Transition to adult health services

Q9: "when they were handing me over to adult services and I went to one appointment it's like oh yeah this might happen to you and this might happen to you and this might happen to you. I was like oh my gosh why have you told me now? I don't know, it just left me feeling really confused, really sort of like oh my God, what if this does happen to me?" *Lily, adult with OI*

## Discussion

Qualitative interviews have been used to explore the impact of OI on the daily lives of patients and families. An important strength of our study was the integration of the views and experiences of patients, parents and professionals. In addition, our participants included nine parents who had OI themselves, allowing us to explore experiences of parenting when also managing their own condition, which has not been a focus of previous studies. Overall, our findings clearly highlight that OI is not just a physical condition and demonstrate the importance of considering both the clinical and psychosocial impacts of OI when addressing the support needs of patients and families.

OI reverberates through all areas of daily life and can impact on emotional well-being. Patients and parents found ways of accepting OI, adapting and coping with the challenges of the condition. They frequently described working to maintain a sense of normality in their lives. Other qualitative studies considering experiences with OI report similar themes that reflect gradual acceptance, coping and aiming for normality.<sup>14,16,17</sup> Times of transition, including external changes, such as starting school, and internal changes, such as navigating teenage years, could be especially challenging for individuals and families. In addition, we found that fractures and hospital stays were particularly difficult for families, as routines were disrupted and parents sometimes struggled to balance care needs, management and planning with the rest of family life. These findings are consistent with other studies<sup>13,17,24</sup> and systematic reviews<sup>11,12</sup> that highlight that there are certain times when additional support and contact with psychosocial services may be needed, including diagnosis, fractures, hospital admissions and transition to adult health services. Offering additional resources to support emotional well-being at specific times may be of great benefit for individuals and families. However, emotional support from health services was noted as being sometimes difficult to access in both paediatric and adult settings.

Some of the psychosocial implications of OI that we identified, such as feelings of isolation, loneliness and being different, have been seen in previous studies with individuals and families affected by OI and other rare conditions.<sup>11,12,25</sup> Notably, we found that feelings of being different were heightened for teenagers and additional psychosocial support may be beneficial. Research looking at participation from the perspective of children and young people with chronic disease suggests caregivers and professionals have a role in supporting children to build a sense of belonging, active involvement and a role within a peer group.<sup>26</sup> More research with young people with OI is needed to understand what supports would be most helpful.

A unique feature of our study was the exploration of experiences with parents who have OI. They described how their condition brought positives to parenting a child with OI, such as their understanding of the condition and confidence in caring for their child. Challenges for parents managing their own OI were centred around the required modifications to daily routines and restrictions on interactions and activities because of reduced mobility, pain and tiredness. Some needed help from partners and others when caring for their children, particularly when a lack of adaptations or equipment was reported. Other studies considering the support needs of parents with physical disabilities highlight that there are both physical and emotional challenges and that parents would benefit from adaptations to make the environment more accessible alongside emotional support and guidance.<sup>27,28</sup> Parents with OI clearly have unique needs for support and adaptations and more research is needed to underpin the development of these resources. Research is also needed to explore the experiences of partners to gain a broader understanding of parenting with OI, taking into

consideration the views and roles of both parents.

Exploration of paediatric service provision indicated high levels of satisfaction with specialist services but suggested there were gaps in accessing psychologists and local therapy services. Consistent with research in other countries, parents felt well supported by specialist paediatric services.<sup>16,24</sup> Our study has, however, highlighted the lack of coordination of services for adults with OI which impeded participant's ability to successfully manage their condition and meet their healthcare needs. Concerns about the transition processes from paediatrics to adult healthcare were also raised, particularly around a lack of clarity of what participants were transitioning to, and the limitations of current adult services may be an additional obstacle in this process. A systematic review considering parents' role in transition for children with long-term health conditions found gaps in transitional healthcare and that differences between paediatric and adult services were barriers to effective transition.<sup>32</sup> Research on best practice for transition for OI is growing<sup>19,29–31</sup> and the use of passports<sup>31</sup> or tools to summarise healthcare history and provide peer mentorship<sup>19</sup> have been suggested to improve facilitation of transition processes.

## Limitations

The patient and parent group included individuals with a wide variety of experiences of life with OI, including differing OI severities, a range of ages and parents with and without OI. In addition, by including the views of professionals we have gained a broader perspective of the impact of OI on individuals and families. Study generalisability has limitations as the majority of participants in the patient and parent group were female, white British and well educated. Another important limitation was that participants were self-selected and recruited based on their willingness to participate in an interview discussing views on stem cells alongside their experiences of OI. As such, care must be taken when interpreting findings. This study was conducted in the UK and findings are not necessarily transferable to other healthcare settings.

## Conclusion

Our findings allow a better understanding of the day-to-day experiences of individuals and families affected with OI and identified key challenges and areas where there is a need to improve the support provided. This knowledge may help clinicians and healthcare systems offer improved services to these families. An in-depth exploration of young people's support needs is warranted with research focused on support through the teenage years and how best to facilitate transition processes. As changes are made to the provision of adult health services for OI, it will be also important to examine their effectiveness in improving delivery of care.

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## Declaration of competing interest

The authors declare that they have no conflict of interest.

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