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Review Article

Exploring the impact of Osteogenesis Imperfecta on families: A mixed-methods systematic review

Melissa Hill, PhD [a, b, *], Celine Lewis, PhD [a, b], Megan Riddington [c], Belinda Crowe [c], Catherine DeVile [c], Cecilia Götherström [d], Lyn Chitty [a, b]

[a] North East Thames Regional Genetics Service, Great Ormond Street Hospital for Children NHS Foundation Trust, London, UK
[b] Genetics and Genomic Medicine, UCL Great Ormond Street Institute of Child Health, London, UK
[c] Osteogenesis Imperfecta Service, Great Ormond Street Hospital for Children NHS Foundation Trust, London, UK
[d] Department of Clinical Science, Intervention & Technology, Karolinska Institutet, Stockholm, Sweden

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ABSTRACT

Background: Osteogenesis Imperfecta (OI) is a rare genetic condition whose key characteristic is increased bone fragility. OI has the potential to impact upon all family members, making it important to consider the challenges families face, how they cope and their support needs as the affected individual moves from childhood through to adult life.
Objective: To conduct a mixed-methods systematic review investigating the experiences of families when a family member is affected with OI.
Methods: A systematic search of seven electronic databases, relevant patient organisation websites and reference lists was conducted. Data extraction was performed for all studies that met the eligibility and quality criteria. Results were synthesised following the principles of thematic analysis.
Results: One mixed-method, six qualitative and six quantitative studies were included in the review. Three overarching themes were identified through thematic analysis: Impact of OI on the psychosocial wellbeing of families, impact on family life and evolving roles and relationships. Fear of fractures and the uncertainty of when the next fracture will occur are key issues that permeate all areas of family life and impact upon all family members.
Conclusion: The experiences, coping strategies and support needs of families affected by OI were highly variable and changed over time. Future research should address the need for adaptive health and education interventions that support all family members.

Osteogenesis Imperfecta (OI) is characterized by bone fragility that results from low bone mass. Common features include bone fractures, osteopenia, varying degrees of short stature and atypical skeletal development.[1,2] Brittle translucent teeth, hearing loss and hypermobile joints can also occur. OI is a rare disease affecting approximately 1 in 10,000—20,000 births.[1–3] OI can be inherited or occur due to a *de novo* mutation. Mutations in the *COL1A1* and *COL1A2* genes that result in abnormal collagen microfibril assembly are the most common cause.[4] Classification into types has been traditionally used to define OI as mild (type 1), moderate (type IV), severe (type III) or lethal (type II). More recently these types have been combined with new genetic classifications.[4] Severity can vary within the OI types and significant variation in clinical course can even occur within families where several members have the same genetic mutation.[5]

Current treatments aim to reduce fractures, provide pain relief and improve mobility and function.[5] Bisphosphonates, that increase bone mineral density and improve bone strength to reduce fracture risk and pain,[6,7] are frequently used for children with moderate to severe OI, but require intravenous infusions, sometimes over 3 days every 2—6 months. Surgical interventions, such as intramedullary rodding, maintain limb alignment and reduce fracture risks when bowing or recurrent fractures in the long bones occur.[5] Physical and occupational therapy aim to improve mobility and function, which in turn will encourage independence.[8]

OI has the potential to impact on the whole family throughout life.[9] Two recent systematic reviews focusing on individuals living

* Corresponding author. Great Ormond Street Hospital, Level 5 Barclay House, 37 Queen Square, London, WC1N 3BH, UK.
E-mail address: Melissa.hill@ucl.ac.uk (M. Hill).

with OI have highlighted how the characteristic clinical features of OI can significantly impact on quality of life (QoL)[10] and are linked with psychosocial implications such as fear of fractures and feelings of isolation and being different.[11] Here we describe a systematic literature review investigating the experiences of families when a family member has OI. By collating and integrating the available research we can begin to look at the challenges families face, how they cope and their support needs as the affected individual moves from childhood through to adult life.

Methods

Design

A systematic review was undertaken to allow the identification, evaluation and synthesis of research findings in a structured and reproducible way. We used an integrative approach to data synthesis and have included qualitative, quantitative and mixed-methods studies.[12]

Search question

The SPIDER acronym was used to guide our systematic search:[13]
Sample: People affected with OI/people with experience of living with someone affected with OI/health professionals and advocates who work with people affected with OI.
Phenomenon of **I**nterest: Experiences and psychosocial impact of OI on families and caregivers.
Design: Interview, focus group or questionnaire.
Evaluation: Lived experience of OI.
Research type: Qualitative, quantitative or mixed-methods.

Search strategy

A systematic search for relevant studies was conducted across five electronic databases (CINAHL, PsychArticles, PsycINFO PubMed, EMBASE). We also searched the Joanna Briggs Institute, Cochrane library and Google Scholar websites. Three relevant patient support group websites were searched (Brittle Bone Society (www.brittlebone.org), Osteogenesis Imperfecta Foundation (www.oif.org) and Osteogenesis Imperfecta Federation Europe (oife.org)) but no unique articles meeting the inclusion criteria were identified. A manual search of the reference lists of included studies and relevant original and review articles was also performed. No time-limit was set on database searches. The last search was conducted on 15/01/2018. Search results were managed using the reference manager EndNoteX7 (Clarivate Analytics, USA).

The search terms used were; 'Osteogenesis Imperfecta' OR 'brittle bone*' AND 'qualitative' OR 'interview' OR 'focus group' OR 'quantitative' OR 'survey' OR 'questionnaire' OR 'attitude' OR 'view' OR 'opinion' OR 'psychosocial' OR 'experience' OR 'family' OR 'relationship' OR 'social' OR 'sibling' OR 'parent' OR 'mother' OR 'father' OR 'caregiver'.

The study selection process followed the PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses) guidance (Fig. 1).[14] After duplicate articles were removed, titles and abstracts were independently reviewed by two researchers (MH and CL). The full-text of any potentially relevant articles were obtained and considered against the inclusion and exclusion criteria by MH and CL independently. Any discrepancies regarding study inclusion were discussed between MH and CL until consensus was reached.

Criteria for the inclusion of studies were:

- Original research articles using qualitative, quantitative or mixed-methods approaches.
- Published in peer-reviewed journals.
- Includes the perspectives of children and adults affected with OI, their parents or carers, siblings, the general public, health professionals or patient advocacy groups.
- Includes family experiences of living with OI including; views, attitudes, beliefs, experiences, actions, expectations, friendships and social life, emotional/psychosocial impact, education and career, healthcare, transition from child to adult health services and hospitalisation.

Criteria for study exclusion were:

- Article not available in English.
- Unable to obtain the full-text.
- Abstracts, editorials, commentaries, letters, and study protocols.
- Non-peer reviewed articles such as reports, books or thesis.
- Quantitative measurements of QoL and studies that focus solely on the psychosocial experience of individuals with OI were excluded, as two recent systematic reviews have addressed these topics.[10,11]

Quality assessment

All eligible original research articles were critically appraised using the quality assessment criteria devised by Kmet et al.,[15] which allows assessment of both qualitative and quantitative research using separate, but equivalent checklists. Checklists for qualitative (10 criterion) and quantitative (14 criterion) studies are scored as "met" (2 points), "partially met" (1 point), "not met" (0 points) or "not applicable". The total score is converted to a percentage. For the mixed-methods studies both the qualitative and the quantitative checklists were used and a combined score given. We used a low cut-off point of 55%, described as liberal by Kmet et al.,[15] that follows the approach used in other mixed-methods systematic reviews.[16,17] The researchers critiqued the included studies to further consider potential limitations such as sample size, recruitment strategies (such as single site versus multiple site recruitment), types of participants (for example; mothers, fathers, siblings; OI types; and parents with or without OI).

Data extraction and synthesis

Study details, including the aim, design, sample characteristics, analysis, and findings were extracted into a table by MH (Table 1). As methodologies included quantitative, qualitative and mixed-methods studies, a narrative synthesis of findings was considered appropriate. NVivo10 (QSR International Pty Ltd. Australia) software was used to facilitate coding and analysis. Research findings were synthesised using a data-based convergent synthesis design.[18] The quantitative and qualitative data were analysed following the principles of thematic analysis[19] and meta-ethnography, which allows findings across different study designs to be integrated and interpreted.[20] The results section of each of the studies was coded. For qualitative studies direct quotes from participants, themes and descriptions were coded. For quantitative studies tabulated data and descriptions of findings were coded. For our thematic analysis, the initial codes were drawn from each of the reviewed studies. Similar codes were then sorted into broad categories that were refined and then grouped into overarching themes.
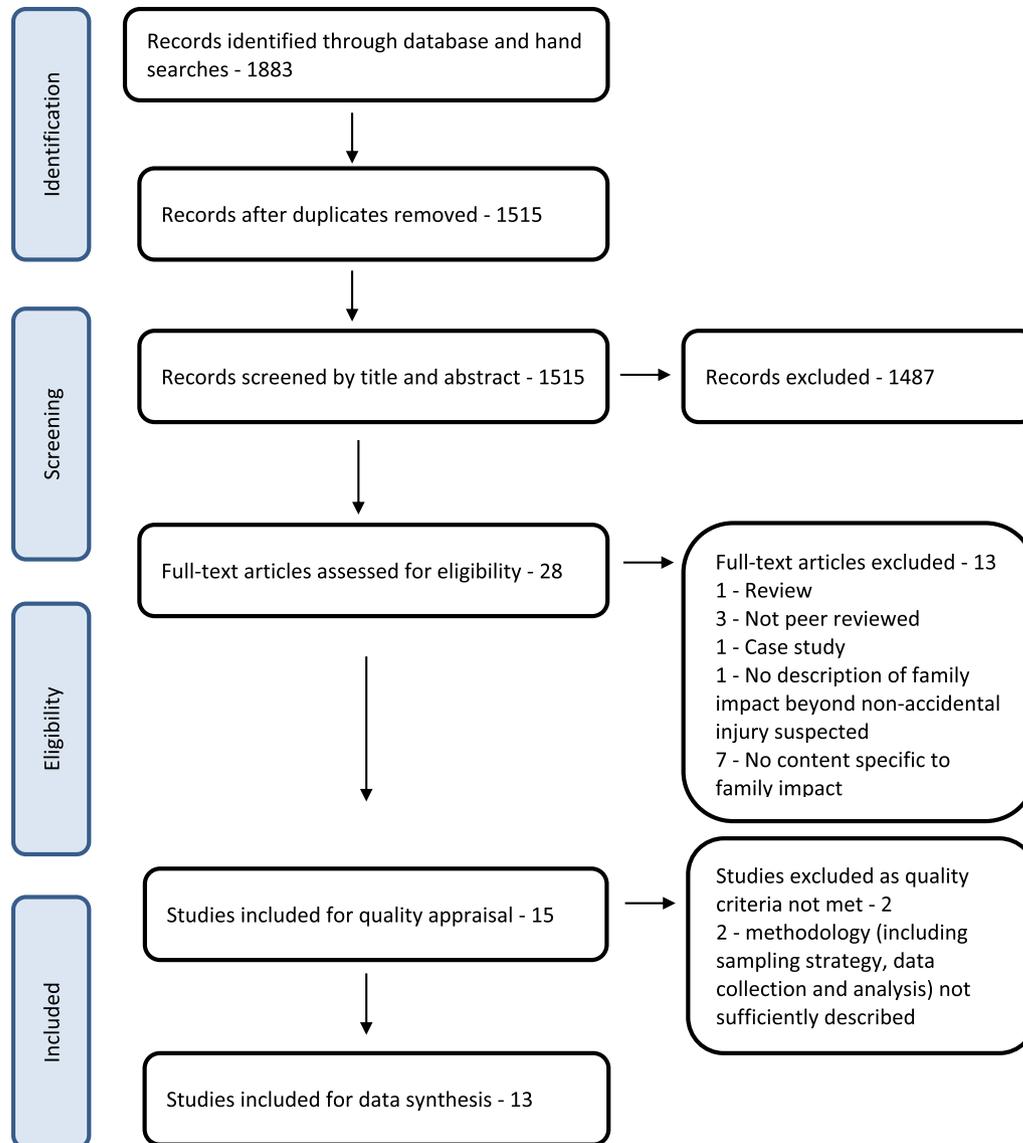
Fig. 1. Study selection process.

Results

Titles and abstracts for 1515 studies were reviewed by MH and CL. The full-text of 28 articles were reviewed against the inclusion criteria, and 15 were included in the quality assessment. Two studies did not meet the cut-off for the KMET quality appraisal checklist and were excluded. Thirteen studies were included for data extraction and synthesis. Details of the study selection process are provided in Fig. 1.

Study characteristics

A summary of findings from each of the thirteen studies included in the review is provided in Table 1. There were three studies from Canada[21–23] and the USA,[24–26] two studies from Turkey[27,28] and individual studies from Brazil[29] Poland,[30] Portugal,[31] Sweden[32] and the UK.[33] Methodological approaches included six qualitative studies,[21,23,24,26,31,33] six quantitative studies,[22,25,27–30] and one mixed-methods study.[32] The KMET tools were used to assess the quality of the studies, the cut-off score for

inclusion in this review was 55%. The individual KMET scores of the included studies are presented in Table 1.

The KMET scores of the included qualitative studies ranged from 75% to 100%. The main limitation was recruitment from single sites in four of the studies.[21,23,26,33] Interviews were used for data collection in all qualitative studies. Interview participants included parents and caregivers,[21,23,26,31,33] adults with OI,[24] children with OI,[21,26,31,33] siblings[31] and health professionals.[33] Participants in some studies were parents of children with OI, who had OI themselves.[23,29,33] These studies focused on; the stereotypes of high intelligence and euphoric personality in individuals with OI,[24] the experiences of adolescents and their parents,[21] parents responses to having a child with OI,[23] the impact of OI on the quality of life of children with OI and their families,[33] the situations that give families with OI the most distress[31] and the experiences of daily life for children and their families when bisphosphonates are used.[26]

For the six quantitative studies KMET scores ranged from 67% to 89%. Sample sizes were generally small in these studies, recruitment was from single sites, some studies did not include detailed descriptions of methodology and approaches to statistical analysis

Table 1
Summary of included studies.

Reference/Country	Purpose	Methodology	Participants	Findings relating to families and caregivers (key themes and concepts)	KMET summary score (%)
Ablon (2003)[24] USA	Examine stereotypes of high intelligence and euphoric personality attributed to persons with OI	Qualitative Anthropological	55 adults with OI (30 female, 25 male) Age: 19–67 years OI Types: III, IV	Parents focus on academic activities over physical activities. A positive approach to life is a response to parental expectations. Note: This study primarily focused on individual experiences of OI.	90
Arabaci et al. (2015)[27] Turkey	Identify the difficulties experienced by caregivers of children with OI	Quantitative Questionnaire (16 open ended/19 closed ended questions)	46 caregivers (43 mothers 3 fathers) Mean age: 35.52 ± 6.65 years	Caregivers of children diagnosed with OI experience psychological and social difficulties Response to diagnosis: 46 (100%) felt anxious, 44 (95.7%) felt sad/sorrow, 40 (87.0%) felt frightened, 39 (84.8%) felt disappointed, 29 (63%) felt shocked, and 28 (60.9%) felt depressed. Difficulties experienced: 26 (56.5%) physical, 45 (97.8%) psychological, 45(97.8%) social, 35 (76.1%) economic changes, 24 (52.1%) no social support, 38 (82.6) inadequate information.	67
Bozkurt et al. (2014)[28] Turkey	Investigate the impact of a psycho-educational program (10 semi-structured three-hour training sessions) for caregivers of children and adolescents with OI	Quantitative Questionnaire (9 open ended/19 closed ended questions plus 4 validated scales: Burden Interview, Coping Strategies Scale, Problem-Solving Inventory, Psychosocial Adjustment to Illness Scale) Completed pre-education, post-education, follow-up	16 caregivers (10 mothers, 5 fathers, 1 aunt) Mean age: 35.25 ± 6.79 years	Prior to training, participants were unable to cope with many aspects of their caregiver responsibilities and lack of knowledge created an emotional burden. Following the training programme, the majority of participants reported positive life changes.	83
Claesson and Brodin (2002)[32] Sweden	Explore the consequences of OI in daily life with a focus on families need for support	Mixed-methods Questionnaire (54 questions) and interviews with a subset Thematic analysis	30 families completed the questionnaire 10 families were interviewed Child age: 0–24 years Child OI type: I (n = 9), III or IV (n = 13)	Topics discussed: 1) First information 2) Parents experiences of the disease 3) Technical devices 4) Influence and participation in decision-making 5) Support in family relations and other relations	61
Dogba et al. (2013)[21] Canada	Explore the impact of severe OI on the lives of adolescents with OI and their parents	Qualitative Interpretive description/thematic analysis	12 young people with OI (9 female, 3 male) Age: 15–21 years OI type: III (n = 4), IV (n = 5), V (n = 3) 12 parents (8 mothers, 4 fathers)	Themes: 1) Starting at the time of diagnosis, a series of stages shaped life and the return to every day "normal" 2) Living with OI was full of "ups and downs" throughout life 3) Every day "normal" life with OI consisted of significant changes for parents and challenges for the whole family 4) Living with OI generated some positive experiences	75
Dogba et al. (2014)[23] Canada	Provide an in-depth understanding of parental responses to OI	Qualitative Interpretive description/thematic analysis	48 parents (34 mothers, 14 fathers) Child OI type: I (n = 7), III, IV or V (n = 32)	Phase: Key determinant of parental responses 1) Initial reaction: Experience of diagnosis 2) Acceptance: Burden of care 3) Normalisation: Child reaction and resilience 4) Passing the baton: Feelings of uncertainty about the future	90
Dogba et al. (2016)[22] Canada	Involve families of children living with OI in the development of a tool to assess the impact of OI on the lives of patients and their families	Quantitative Integrative knowledge translation Pilot Questionnaire: Impact of OI, Experiences, Challenges and Expectations of patients and families (I-OI/ECE)	27 families (17 completed by mother only) Child OI type: I, III, IV, V, VI, Other	29% received psychological support at diagnosis (7 of 8 who received psychological support thought it should be provided routinely, starting with diagnosis and throughout the life span) 30% were suspected of non-accidental injury 70% had high expectations of specialist services 40% reported no effect on marital life, 11% reported a negative effect on marital life 8% experienced a drop in net family income	89
Hill et al. (2014)[33] UK	Determine how OI impacts on the quality of life and well-being of children and their families	Qualitative Phenomenology	10 children (5 female, 5 male) Age: 6–17 years OI type: mild (n = 3), moderate (n = 4), severe (n = 3) 10 parents (8 female, 2 male)	Key themes identified: 1) Being safe and careful 2) Reduced function 3) Pain 4) Fear 5) Isolation 6) Independence	95

Table 1 (*continued*)

Reference/Country	Purpose	Methodology	Participants	Findings relating to families and caregivers (key themes and concepts)	KMET summary score (%)
Santos et al. (2017)[31] Portugal	Explore the situations perceived to be the most distressing and stressful for individuals with OI and their families	Qualitative Discourse analysis Disability Index of the Childhood Health Assessment Questionnaire (CHAQ)	Age: 28—52 years 5 health professionals (4 female, 1 Male) Age: 28—54 years 7 families 7 children with OI Age: 5—16 years OI type: I (n = 7) 4 Siblings Age: 9—16 years 7 mothers Age: 23—39 years 6 fathers Age: 25—45 years	Key times of stress for families: 1) Time of diagnosis 2) Fractures and pain 3) Hospitalisation 4) Recovery at home 5) Back to school All children scored as mild to moderate difficulties on the CHAQ scale	95
Suskauer et al. (2003)[25] USA	Determine if parental over-protection and parent coping are related to the motor performance and temperament of their children with OI	Quantitative Carey Temperament Scale Vulnerable Child/ Overprotecting Parent Scale (VCOPS) Childhood Health Assessment Questionnaire (CHAQ) Parent Daily Hassles Scale (PDH)	34 mothers Child with OI age: 1—12 years OI type: III (n = 18), IV (n = 17)	Parents reports of a higher frequency and intensity of daily hassles correlated with child traits of negative mood, more intensity, less adaptability, and less predictability. No significant correlation between parental overprotection and temperament or physical performance.	82
Szczepaniak-Kubat et al. (2012)[30] Poland	Assess QoL in parents of children or adolescents with OI	Quantitative Cross-sectional design WHOQOL-BREF QoL questionnaire	25 families 25 mothers Median age: 34.3 ± 7.1 years 24 fathers Median age: 38.1 ± 8.5 OI type: I (n = 7), III (n = 14), IV (n = 4)	56% - Reported their global quality of life as good 60% - Reported their health status as good QoL scores were highest in the psychological domain and lowest in the physical health domain. Parents of children with severe OI (Type III) had a lower score on the environmental domain than parents of children with mild OI (types I and IV). (P = 0.025)	77
Vanz et al. (2015)[29] Brazil	Assess QoL in caregivers of children with OI	Quantitative Cross-sectional design WHOQOL-BREF QoL questionnaire	24 Caregivers (16 mothers, 1 stepmother, 6 fathers, 1 grandmother) Mean age: 39 ± 9.1 years 2 caregivers had OI (I, IV) Child OI type: I (n = 10), III (n = 4) IV (n = 13)	All mean domain scores were significantly lower than those of healthy individuals Brazil QoL scores were highest for the social relationships domain and lowest for the environmental domain. QoL scores did not differ significantly according to OI type or number of fractures	73
Wiggins and Kreikemeier (2017)[26] USA	Describe the experience of daily life for children diagnosed with OI (who are receiving intravenous cyclic bisphosphonate therapy) and their parents	Qualitative Human science approach	6 children with OI (4 females, 2 males) Age: 6—18 years OI types: I (n = 2), III-IV (n = 1), IV (n = 3) 6 mothers	Key themes identified: 1) Living daily life in stride 2) Normalcy is living with uncertainty 3) Renewal with infusions 4) Making choices and living with the consequences	100

were primarily descriptive. Participants were parents and caregivers of children with OI.[22,25,27–30] Aims and methodologies were diverse. Two studies used a validated scale (WHOQOL-BREF) to consider the Qol of parents of children with OI to population controls.[29,30] In the remaining studies; one used validated measures to consider the relationship between child temperament and motor performance and parental over-protection and coping,[25] one used a descriptive questionnaire to identify the difficulties experienced by caregivers of children with OI,[27] one used a descriptive questionnaire and validated scales to examine the value of an education program for carers[28] and one described the development and piloting of a questionnaire to assess the impact of OI on families.[22]

The sole mixed-methods study had a KMET score of 61%. The key limitations were a small sample size and minimal description of methodology. The reported findings were descriptive. Participants were parents and caregivers or children with OI recruited through a national association. The aim of the study was to explore the impact of OI on daily life with a focus on support needs for families.[32]

Although individual study aims and approaches were diverse, the findings gave insights into the day-to-day life, experiences and psychosocial impacts of living with OI. Study findings are discussed below within the three overarching themes that were identified. Table 2 outlines the themes and sub-themes and indicates which studies contributed to the derivation of the themes.

Table 2
Summary of themes and sub themes.

Themes and sub-themes	Quantitative						Qualitative						Mixed
	Arabaci 2015[27]	Bozkurt 2014[28]	Dogba 2016[22]	Suskauer 2003[25]	Szczepaniak-Kubat 2012[30]	Vanz 2015[29]	Ablon 2003[24]	Dogba 2013[21]	Dogba 2014[23]	Hill 2014[33]	Santos 2017[31]	Wiggins 2017[26]	Claesson 2002[32]
Impact of OI on the psychosocial wellbeing of families													
Diagnosis	X	X	X					X	X		X		X
Fear of fractures	X							X	X	X	X	X	
Emotional challenges	X	X	X				X	X	X	X	X	X	X
Coping and resilience	X	X		X			X	X	X			X	X
Impact on family life													
Impact on family functioning	X	X	X		X	X		X		X	X	X	X
Impact on professional life, finances and the home	X	X	X		X			X	X	X			X
Fracture management							X			X		X	X
Evolving roles and relationships													
Overprotective parenting: an internal battle for parents	X								X	X			
Parents support the child's growing independence							X		X			X	
Transition to adulthood								X	X				

Impact of OI on the psychosocial wellbeing of families

Diagnosis

Parents describe diagnosis as an intensely difficult time.[21–23,27,28,31,32] Diagnosis was often sudden and unexpected and frequently described as devastating.[21–23,27,28,31,32] Parents experience multiple and sometimes contradictory emotions; shock, relief, anger, guilt and denial.[21,23,27] In some circumstances, diagnosis was a relief as it decreased uncertainty, ruled out lethal conditions, and explained frequent fractures.[23,31] Relief was particularly apparent after a long period of searching for a diagnosis or if there had been a suspicion of abuse.[21,23,31] Dogba et al.[23] discussed two factors influencing parents' responses to a diagnosis of OI; health professionals who focus only on the negatives of OI when discussing the diagnosis and prior knowledge of OI through their family history. Only one paper touched on parents' experiences of diagnosis during pregnancy.[32] Parents described the devastating impact of seeing ultrasound pictures or being told something was wrong with the baby.[32] Feelings of anxiety and distress at the time of diagnosis were amplified when health professionals were not OI specialists or when parents felt health professionals were not listening to their opinions.[31] In several studies, parents described being unsatisfied with the information and support they received and sometimes had to seek information on their own.[21,23,28,31,32] In one quantitative study with 27 parents, the majority (70%) reported that they did not receive psychological support following diagnosis.[22]

Fear of fractures

Fear of fractures permeated all areas of family life.[21,26,27,31,33] The types of activities the child can undertake are restricted[21,23,33] and all family members take an active role in keeping the child safe from fractures.[31] Early in the child's life parents face worries that even basic handling and care could result in a fracture.[26,33] Notably, some parents doubted their ability to parent[26] and some reported feeling unable to care for a fragile child and wanted additional support.[31] Fear of handling the child was also felt by the extended family.[33] Anxiety is intensified as both parents and children understand that being careful can't always protect them from fractures as external factors come into play.[31] Parents fear being separated from the child, and transition from sole care at home to shared care at nursery or school can be daunting.[33] Parents sometimes restrict their own activities and social interactions so that they can stay close and keep the child safe.[27]

Emotional challenges

The emotional "burden" of being a caregiver for a child with OI was raised by parents in several studies.[23,24,26–28,31] Daily care, multiple fractures, frequent health care appointments and the effort required to stay safe take an emotional toll[26] and those who had OI themselves also felt guilty for passing on the gene.[23,33] Many emotional burdens discussed, including feelings of lack of control, uncertainty and helplessness, related to the child's perceived vulnerability.[26,27,31] Some parents felt socially isolated as a result of their child's OI.[23,27] Parents reported finding it distressing to see their child in pain[31,33] and found their child's tiredness and fatigue challenging.[26,33] Concerns about the future were also evident, particularly for parents of children with severe OI.[23]

Daily life was sometimes described as frustrating and exhausting and parents sometimes described themselves as being anxious or depressed as a result.[23,26–28,31] Dogba et al.[23] noted that the disease burden was greater for parents of children with severe OI, who had more frequent fractures and clinical appointments, and when children were younger and when more than one child was affected.[23] Several studies demonstrated the ups and downs that living with OI brings for families[21,23,26,31,33] and key times of additional stress such as diagnosis, fractures and hospital stays after a fracture or surgery and recovery at home have been highlighted.[31] In addition, new settings and times of transition required parental advocacy, proactive planning, and additional support from health professionals.[26,31–33]

Coping and resilience

The ability of families to cope with, and be resilient to, the additional challenges OI brings were evident in many studies.[21,23,24,26,27] Over time the diagnosis and its consequences are accepted. Families adapt, the child's needs are merged into daily routines and families endeavour to keep their outlook positive and maintain a normal family life regardless of the challenges faced and the energy required.[21,23,26,27] Wiggens et al.[26] found that the improvements from bisphosphonate treatment on their child's wellbeing (reduced fractures, pain and tiredness) had a positive impact on parents' confidence and strengthened their ability to take daily life in their stride. Another factor that allowed parents' to remain

positive was focusing on social or academic activities over physical activities.[23,24]

Child temperament can impact on how parents cope.[23,25] One quantitative study specifically looked at the impact of child temperament on parent coping using the Parent Daily Hassles Scale.[25] Aspects of the child's behaviour, such as negative mood, high intensity, lower predictability and persistence of expression increased parental perception of daily hassles. Disease severity and motor performance were not significantly related to daily hassles.[25] The ability of the child to cope with OI influenced parental responses and the resilience of their child was often regarded as a great inspiration.[23] The relationship between child and parent coping is complex and one study indicated that children hide their pain to help their parents and make up for causing them pain and frustration.[24]

Sources of support included friends, family, health professionals, religious faith and other families affected by OI.[21,23,27,32] Information seeking through the internet, social media and other parents of children with OI appears to play a major part in coping, particularly at the time of diagnosis.[23,27,32] In turn, sharing their experiences with other parents was viewed positively.[21] In two studies the need for routine psychosocial support for families was emphasised.[22,28] In one questionnaire study with 27 families, 70% reported high expectations for specialized services to help them care for their children, plan and coordinate services from multiple specialists, manage transitions, train professionals and build awareness of OI.[22]

One small study from Turkey described the evaluation of a psycho-educational training programme for caregivers of children with OI that consisted of 10 three hour training sessions.[28] Prior to the study the 16 participants had stated that OI services were inadequate and 94% reported that they had not received enough information about OI. Following the training, 94% reported having adequate information and 75% reported positive changes in their lives.

Impact on family life

Impact on family functioning

In addition to accompanying the child to routine visits, treatment appointments and daily care routines, family life is disrupted by the need to respond to fractures and the uncertainty of when a fracture might occur. To allow all family members to be involved, the types of family activities undertaken are sometimes restricted and additional planning and organisation is often needed.[32,33] Tiredness can also limit the types and timing of activities.[33] Family activities are affected by uncertainty as it is impossible to predict when a fracture could mean that family plans are cancelled.[26]

The attention required by the parents for the child with OI impacts on siblings[21,26–28,31] and can influence the parents' relationship.[21,22,27,28,32] It was noted in one study that there is less time and energy for other family members and siblings can feel abandoned.[21] At times when the focus on the child with OI is more pronounced, such as hospital stays, the roles and routines of family members are disrupted and siblings can feel lonely, sad or angry and resentful.[31] Siblings are involved in supporting the child with OI and keeping them safe. For example, siblings know to avoid rough play.[26] In addition, siblings often become more caring and compassionate.[21] Impaired partner relationships were discussed, with anger and fighting resulting from the stress of diagnosis and day-to-day care.[21,22,28] Parents could also become closer, as sharing care and dealing with stress drew them together.[21] Claesson et al.[32] noted that it was important for parents to have time together as a couple and that respite care is needed.

Two small studies considered parents' QoL using the WHOQOL-BREF which contains 26 questions distributed in four QoL domains; physical health, psychological, social relationships, and environment. Szczepaniak-Kubat et al.[30] looked at the QoL of 25 mothers and 24 fathers of children with OI in Poland. They concluded that global QoL and health were not affected by having a child with OI. Vanz et al.[29] looked at QoL in 24 caregivers of children with OI in Brazil. Scores were highest for social relationships and lowest for environment (feelings of safety, accommodation, access to information and participation in/opportunity for recreation and leisure). QoL was lower in the physical, psychological and environment domains compared to a Brazilian control group (n = 50).

Impact on professional life, finances and the home

Changes to professional life and financial burdens were discussed in several studies.[21–23,27,28,30,33] Many parents, primarily mothers, worked part-time or did not work outside the home due to daily care needs of the child, large numbers of medical appointments and concerns about safe child care.[21,23,27,30] Some parents were satisfied with the decision not to work or study, while others felt frustrated at the lack of control over their lives.[23] Financial burdens arise from changes to professional life, frequent medical appointments and treatment costs.[21,27] Participants in several studies noted that they had either adapted their home or moved to meet the accessibility needs and promote independence for their child with OI.[21,22,27,32]

Fracture management

Parents take the lead in administering first aid when fractures occur, learning to provide early pain relief and splinting,[26,33] and deciding when hospital care is needed.[32] Parents report that health professionals encountered outside of specialist centres frequently have little or no experience with OI.[24,26,32] Stress and delay can occur when seeking help for fractures[26] and incorrect care has been experienced.[32] Emergency care can, however, be positive when health professionals give plenty of time or if they understand OI and invite input from the child and family.[26]

Evolving roles and relationships

Overprotective parenting: an internal battle for parents

Fear for the child's safety can result in parents becoming very protective of children as they strive to avoid risks and prevent fractures. An internal struggle exists for parents, as they recognise the need to support their child's independence, but at the same time they view keeping the child safe as paramount.[32,33] Parents acknowledge being overprotective, and while some viewed it as necessary,[27] some parents said they regretted being overprotective.[23] Parents also spoke about their inability to let go and finding it difficult to hand over care to school or nursery.[27,32,33] There was also evidence of children struggling for independence from families and school. Children in one study spoke about disliking their parents attempts to keep them safe and doing everything for them.[33]

Parents support the child's growing independence

The way parents view their role and their relationship with their child evolves over time. When the child is young parents take the primary role in the proactive planning required to keep the child safe and frequently act as an advocate for the child in healthcare management and at times of transition, such as starting school. Although it was acknowledged as a struggle and took time to

accept, some parents felt that an important aspect of their role was to help children to be independent and learn that choices have consequences.[26] Older children take a proactive role in staying safe and dealing with uncertainty and co-manage decision-making about staying safe with parents.[26] Parents recognise that they are overprotective and their need to work to support their child's independence.[23] Parents can have a positive impact on independence, and some adults with OI spoke about how their parents raised them to be independent and to have a positive outlook on life.[24]

Transition to adulthood

The struggle faced by parents to let go continues into adulthood and health professionals spoke about the process of letting go as being "nerve wracking" for parents when children reach young adulthood and gain more independence.[33] Parents of children with mild forms of OI expect their child to manage their own life and health as adults and describe preparing their child to live independently.[23] Parents want their child with OI to work and support themselves. This was regarded as a right for young people with disabilities and a question of accessibility.[32] When children had severe OI parents took on a role of either being a companion or manager, depending on the child's ability to be independent.[23] Parents anticipated supporting their child through experiences, such as having a child themselves, and felt "morally bound" to do so.[23]

Discussion

Our review highlights the complexity of family life when a family member has OI. Families face the challenge of integrating frequent medical appointments, restriction of certain activities, response to fractures and time in hospital into daily life. Fear of fractures and the uncertainty and unpredictability of when the next fracture could occur are a constant undercurrent that impacts all areas family life. Parents can feel a heavy burden from care-giving, face feelings of stress, helplessness and loss of control and experience guilt, anxiety, depression and a lack of confidence in parenting skills. The diagnosis is initially devastating for many families, but many are resilient and can, over time, accept the diagnosis, adapt and focus on maintaining normal family life. Similarly diverse psychosocial impacts are seen in individuals affected with OI and although resilience and a positive mind set are common, varying degrees of adjustment and coping are seen and some children and adults with OI report feelings of loneliness and experience depression and anxiety.[11]

There is considerable overlap in the experiences of families affected by OI and those of families affected by other chronic childhood conditions.[34—37] In a review of 34 studies of families where a child was affected with a chronic condition such as diabetes, juvenile arthritis or asthma, Smith et al.[37] identified evidence of resilience where parents gradually accept the diagnosis and then focus on meeting the child's needs and integrating them into normal family life. Smith et al.[37] also found that some families faced psychosocial, emotional and relationship difficulties. A systematic review of 13 quantitative and 96 qualitative studies that specifically considered parenting stress in caregivers of children with chronic conditions such as diabetes, sickle cell disorder and cancer, demonstrated that parents felt greater general parenting stress than caregivers of healthy children.[35] Notably, frequent and/or intense episodes of pain were linked to increased parenting stress for caregivers of children with arthritis and sickle cell disorder.[37] The distress and helplessness felt by parents when the affected child was in pain was also seen for families affected with OI.

Diagnosis was a difficult experience for most families and parents often felt that more support and information was needed at this time. How diagnosis is managed and communicated to parents by health professionals can impact greatly on their ability to cope[23] and this is a key time-point for health professionals to consider the need for additional interventions. Non-accidental injury is a differential diagnosis for OI and the added emotional distress of being suspected of child abuse was touched on by parents in some studies and resulted in families feeling relief when a diagnosis of OI was given. There were, however, no detailed explorations of the short and long-term impact of this experience which may leave parents lacking confidence in their own parenting and distrustful of healthcare providers. Studies reviewing medical records demonstrate the potential for additional emotional distress when non-accidental injury is suspected. For example, Kocher et al.[38] found that in 33 cases of suspected child abuse later diagnosed as OI, 23 parents had the child removed by social services and 20 had older siblings removed as well.

There is a delicate balance for parents to achieve as they work to ensure that a child with OI is not left out whilst the needs of other family members are also met. Finding this balance is important for child development and family functioning.[39] Overall, mothers were the most common research participants reporting on the experience of families in the included studies. An absence of the experiences and views from fathers has been seen in other reviews of family experiences of chronic childhood conditions,[37,40] an important omission that needs addressing as fathers' involvement in managing healthcare when the child has a chronic condition can positively impact on all family members and supports family functioning.[40] Similarly, although experiences of siblings were discussed by parents in several studies, siblings were only included as participants in one study.[31] There is some evidence to suggest that siblings of children with chronic conditions are at risk of negative impacts on psychological functioning (depression/anxiety), peer problems and behavioural difficulties.[34,41] However, in one review siblings were found to be more warm and caring as a result of having a brother or sister with cancer.[42] Positive outcomes for siblings were also seen here, with parents reporting siblings as being more caring as a result of helping to look after their brother or sister with OI. Other studies have found that the emotional experience for siblings is complex and they experienced diverse and contradictory feelings about the implications of the disorder, their family and social life.[43] Siblings may need help to communicate their feelings and could benefit from interventions that help to validate and normalise mixed emotions, decrease anxiety or improve social and emotional functioning.[41,43]

A key challenge for parents of children affected with OI is adapting to their changing roles as the child grows and increases their independence. Many parents struggled against being overprotective and wanted to support their child's independence. Parents of children with other chronic conditions also find it challenging to step back and let children become independent.[44] Transition to adulthood is particularly difficult and parental concern makes it difficult to hand over responsibility for health management.[44] Ultimately, both the young person and his/her parents are at risk for psychosocial and emotional difficulties which may lead to problems with development and health.[16] Transition to adult healthcare is an area that requires more research for families with OI; this process was only touched on in one study.[23] Transition guidelines for young people with OI stress the need for health professionals to support the family and encourage the young person to be their own care-coordinator and advocate.[45]

A multidisciplinary approach is recommended for caring for children with OI[22,46] and specialist services should consider the psychosocial wellbeing of the family as a whole. Approaches to care

need to evolve over time as family challenges will change as the child moves from childhood to adolescence and into adulthood.[21] The studies included here demonstrate that there are key times when families may need extra clinical and psychosocial support; diagnosis, fractures, hospital admissions, starting school and transition to adult health services. Parents and health professionals need to form effective partnerships that allow parents to be heard, valued and actively involved in the development of care plans.[37] Interactions with health professionals when families were seen for emergency care were mixed as health professionals were not always experienced in OI. General practitioners (GPs) in the UK have also reported finding patients with OI difficult to manage due to lack of knowledge.[47] These experiences could be improved through wider education of health professionals about OI and creating comprehensive web-based information that is linked to patient records.[47]

One small study looked at an educational training programme for parents of children with OI and parents reported positive life changes as a result.[28] More research is required in this area. Families in the reviewed studies frequently sought support and information from other families affected with OI and from patient support groups. Thus, support and interventions developed by specialist services working with the wider OI community with web-based support or peer-led interventions may be valuable. For example, approaches such as peer coaching have been shown to be acceptable and feasible for parents of children with other chronic conditions such as diabetes.[48,49] Several areas where support and intervention may have a positive impact were identified through our review: 1) Support following diagnosis that includes practical guidance on how to care for the child as fear of handling the child was raised and how this may impact on attachment and bonding, 2) Building skills and strategies for coping, resilience and adaptation, 3) Normalising and managing fear of fractures and helping parents find a way to avoid overprotective behaviours and support independence in their child, and 4) Strategies to recognise when families are at risk of mental health issues such as anxiety and depression so that interventions or referral to psychology services can be offered.

Strengths and limitations

A key strength of this review was the rigorous and systematic approach to identifying studies which was undertaken independently by two researchers. Another strength was the integration of evidence from qualitative, quantitative and mixed-methods research. Seven studies that included in-depth interviews with parents and other family members were available to provide rich data on the experiences of families. However, the studies included were limited to those available in English. Unpublished studies were not included, and publication bias may be a limitation. The voices of siblings and fathers are missing as only one study included siblings as participants and many more mothers than fathers participated. As studies generally had small participant numbers, there were limited opportunities to compare subgroups such as severe versus mild OI or the experiences of families where OI is a new diagnosis compared to those with a family history of OI. Moreover, the specific experiences of parents who have OI themselves and must additionally manage their own condition were not reported in the included studies. All papers included in the review underwent quality assessment. The included qualitative studies were all of a high standard. The quality of the included quantitative and mixed-methods studies was more varied and limitations included small sample sizes, lack of detail in descriptions of methodology and lack of detailed statistical analysis. Overall, the research literature looking at the experiences of families with OI is primarily descriptive and future research is needed that takes a more rigorous approach.

Conclusions

The findings of this review clearly highlight that OI is a condition that impacts on the whole family. Our review identified several gaps in the literature, such as the under-representation of siblings and fathers, that should be addressed in future studies. Research that considers possible interventions across the lifespan that target all family members is also needed. It is important to provide health and education that aims to support all family members that can be adapted to fit with the unique needs of individual families.

Conflicts of interest

None to declare.

Disclosures

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