A Qualitative Exploration of Patient Perspectives on Psychosocial Burdens and Positive Factors in Adults with Osteogenesis Imperfecta

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Abstract

Osteogenesis imperfecta (OI) is a pleiotropic, heritable connective tissue disorder associated with a wide range of health implications, including frequent bone fracture. While progress has been made to understand the spectrum of these physical health implications, the impact of OI on psychosocial well-being, as well as protective factors that buffer against adverse psychosocial outcomes, remain understudied. This present study relies on a qualitative approach to assess

patient perspectives on both protective and adverse psychosocial factors specific to OI in 15 adults with varying disease status. Semi-structured interviews were conducted, subsequently coded, and themes extracted. Themes concerning psychosocial burdens (i.e. negative affective and behavioral impacts of disease status) and protective factors were identified from cooperatively-coded transcripts (2 coders per transcript). Participants reported experiencing an increase in negative affect and disease-related distress after fracturing a bone and during recovery. Fear and concern specific to the uncertainty of future bone fractures and negative self-image was common. In contrast to these negative impacts, participants additionally described positive orientations toward their disease and attributed positive traits to their lived experience with a chronic disease. While limited due to small sample size and lack of ethno-racial diversity, findings highlight a need for continued research on the relationship between OI disease status and psychosocial outcomes, as well as the development of psychological interventions designed for OI populations. Findings have relevant clinical applications for healthcare providers working with those diagnosed with OI.

INTRODUCTION

Osteogenesis imperfecta (OI) is a connective tissue disorder that is caused by dominant or recessive alleles in 18 different genes that are important in type I collagen production (Tauer et al., 2019; Tosi et al., 2019). Common physical characteristics of OI include bone fragility, short stature, bone deformity, joint laxity, dental and craniofacial pathology, and sclera discoloration. Recent estimates indicate that OI occurs in 1 in 20,000 births within the United States (Etich et al., 2020). The 2015 Nosology and Classification of Genetic Skeletal Disorders proposes four primary clinical presentations of OI that range in severity and impairment, although other types do exist but are less common: Type I (mild), Type IV (moderate), Type III (severe), and Type II (fatal at or shortly following birth) (Mortier et al., 2019). Given that there is no known cure for OI, treatment has largely focused on physical disease symptom management (Marr et al., 2017). There has been limited research, however, regarding the assessment and treatment of the psychosocial needs of adults with OI. Psychosocial intervention research is well-established among other chronic disease populations (Acquati & Kayser, 2019; Caruto & Breitbart, 2020; Hirase et al., 2018; van Bastelaar et al., 2008; Dorstyn et al., 2011), but OI-specific research is clearly needed due to the potential for disease-specific experiences and psychosocial needs.

Despite ongoing efforts to understand the ways in which OI impacts child and family quality of life, research evaluating the psychosocial and mental health repercussions of adults with OI is even more limited (Tosi et al., 2015; Tsimicalis et al., 2016), with few studies assessing the psychosocial experiences of adults with OI. Tosi and colleagues (2015) conducted a large-scale survey to compare health and psychosocial concerns among a sample ($n = 959$) of adults with OI to population-level norms (Tosi et al., 2015). Adults with OI reported greater concerns specific to different organ systems (e.g., musculoskeletal, auditory, and pulmonary subsystems) which was more pronounced for individuals with

more severe OI presentations. Somewhat unexpectedly, individuals with OI reported lower levels of pain intensity and interference relative to the general population (Tosi et al., 2015). Additionally, the OI sample reported lower levels of satisfaction with social roles and greater levels of anxiety and depression compared to population level norms; although statistically significant, clinical significance for anxiety and depression was not observed, suggesting that levels of anxiety and depression for the sample were comparable to the general population. Subsequent studies have provided inconsistent findings regarding mental health, with some finding elevated levels of anxiety and depression (Arponen et al., 2021; Barlow et al., 2022; Tosi et al., 2019), and others suggesting that these concerns may not be elevated within an adult OI population (Bendixen et al., 2018). Research among children and families with OI has also included evaluation of psychosocial challenges, with similarly varied results across studies which provide evidence for both positive and negative psychosocial outcomes as a result of OI status (Dogba et al., 2013; Dogba et al., 2014; Hill et al., 2014; Santos et al., 2018; Wiggins & Kreikemeier, 2017). Commonly reported negative consequences include physical and functional limitations and the impact this may have on the family structure in addition to parents' chronic fear for their child's safety (Brodin, 1993; Claesson & Brodin, 2002; Cole, 1993; Moorefield & Miller, 1980; Suskauer et al., 2003; Wekre et al., 2011). However, as families and children adjust to their diagnosis, positive experiences such as resilience and high achievement have been noted (Ablon, 2003; Alston, 1983; Widmann et al., 2002; Pelentsoy et al., 2016). Further research is clearly needed to better understand factors which may affect mental health in people living with OI.

While the presence of an OI diagnosis has been associated with physical and mental health concerns among adults, children, and families, it is important to recognize both positive and negative impacts of OI on individuals. The use of different coping strategies appears as an important buffer against adverse psychosocial outcomes (Tsimicalis et al., 2016). Research has identified using principles of *acceptance,* or the active embrace of internal psychological experiences without attempting to change them (Hayes et al., 2006), as a set of strategies which may help adults with OI (Balkefors et al., 2013). Additionally, developing and maintaining an optimistic outlook and the use of humor have been found to be beneficial for overall well-being among those with OI (Tsimicalis et al., 2016). Behavioral changes have also been found to be effective for buffering against negative psychosocial outcomes: in one study, 17 out of 19 adults with OI who adhered to a regimen of 30-minutes of daily physical exercise reported improvements in life satisfaction (Balkefors et al., 2013). Lastly, individual resilience can help buffer against adverse outcomes through self-acceptance, adaptation, and willingness to engage in activities regardless of physical limitations (Cole, 1993; Hayes et al., 2006). These findings emphasize the need to delve into the positive life experiences and personal characteristics of individuals with OI, as well as understanding their psychosocial challenges.

Though existing literature does provide evidence of unique psychosocial factors associated with chronic health conditions, research in this area specific to adults with OI has been limited (Tosi et al., 2015; Cole, 1993; Hill et al., 2022). With this in mind, the present study expanded upon the current understanding of psychosocial factors among adults diagnosed with OI, specifically examining factors associated with psychosocial burdens as well as

protective factors that may buffer against negative psychosocial outcomes through the use of qualitative semi-structured interviews. We hypothesized the following:

1. Adult OI participants would report psychosocial burdens associated with anxiety and depression related to their OI status.

2. Adult OI participants would report protective factors that buffer against adverse psychosocial outcomes specific to their OI status.

MATERIALS AND METHODS

Study Design

Semi-structured interviews were conducted with adults with OI ($n = 15$) addressing topics including mental health, pain, future priorities for OI interest stakeholders, and the impact of OI on daily life. The study was approved by the Institutional Review Board of Baylor College of Medicine in accordance with the Declaration of Helsinki. All participants provided informed written consent and were compensated with a $50 electronic gift card.

Sampling and Recruitment

For the present study, eligible participants included English-speaking adults (ages 18 – 85) from the United States or Canada who identified as being diagnosed with OI; all OI subtypes were eligible to participate. Participants were recruited via Facebook and email through the Osteogenesis Imperfecta Foundation. Interested participants contacted the research team via email and were subsequently contacted over phone or email and scheduled for the research study.

Data Collection

An interview guide was developed in collaboration with the Osteogenesis Imperfecta Foundation, collaborating researchers, and clinicians experienced in the treatment of OI to address topics of interest utilizing open-ended questions (see Supplemental Materials for guide). Interview questions focused on general characterization of OI, pain challenges specific to OI, mental health challenges, and priorities for future interventions and services for the OI community. While all topic areas were addressed in each interview, the current manuscript focuses solely on participant responses related to mental health challenges and positive factors, with other topics to be addressed in future manuscripts. All study visits were conducted remotely over Zoom or Microsoft Teams. Demographics (including research categories of race/ethnicity) were self-reported by participants to the research team. Semi-structured interviews were conducted by trained members of the research team and supervised by a senior psychologist (EAS) with extensive clinical and research experience with semi-structured clinical interviewing. Interviews averaged 62-minutes (range: 46–74), and all interviews were audio-recorded.

Data Analysis

Interviews were transcribed and subsequently reviewed by a member of the research team for accuracy (WCR). Transcribed interviews were analyzed in MAXQDA2020 using a codebook developed collaboratively by research team members (WCR, ADW, CNM, KMK)

to reach consensus regarding code definitions (Decuir-Gunby et al., 2011). Two members of the study team then independently coded each interview, after which inconsistencies were addressed by WCR and ADW to reach consensus in code application (Ellis et al., 1992). Code outputs were analyzed using Thematic Analysis to identify salient themes expressed by the study sample during interviews (Braun & Clarke, 2020). This analysis identified themes reflecting common psychosocial needs and potential mitigating factors for adults with OI. Theme frequencies were calculated to identify the highest priority needs and potential protective factors.

RESULTS

Sample Characteristics

The final sample consisted of 15 adults with OI (M = 41.2 years, SD = 17.38, Mdn = 36). Complete demographic data are reported in Table 1.

Thematic Findings

A number of key thematic findings resulting from OI disease status appear to negatively impact psychosocial well-being, including loss of mobility due to fracture, the unpredictability of fracture, and self-image. On the other hand, participants discussed positive outcomes and experiences associated with their OI disease status which may buffer against negative psychosocial effects. Thematic frequency data are provided in Table 2.

Loss of mobility due to fracture poses a significant psychosocial burden.

All participants described several signs throughout their interviews associated with negative psychosocial outcomes, including components of clinically relevant diagnoses such as depression and anxiety. These burdens appeared to have been most severe during periods spent recovering from fractures that caused a loss of mobility. While describing the periods of time immediately following a fracture, 8/15 of participants expressed some type of negative affect; common emotional responses included hopelessness (6/15) and anger (6/15) while behavioral components such as isolation were also frequently reported (11/15). One participant described their experience while recovering from a fracture as, "…that internal voice turns very evil because you have nothing else to do…what seems like six weeks to recover from a fracture can get you into very deep depression waters." (32 y/o, F, type II*) These periods of time appear to have been characterized by intense isolation and loneliness for many participants, who expressed isolation not just in the form of physical disconnection from friends, family, and daily routine during hospitalizations and extended recovery periods at home, but cognitive isolation associated with inward-facing thoughts of individualized suffering.

Feelings of hopelessness may be connected to the number of fractures participants suffered, with one participant with type I OI mentioning that "It's kind of tough sometimes. You question why it happens, you know, or why even have a life." (20 y/o, M, type I) Many participants similarly expressed sentiments of hopelessness (6/15) about the interminable nature of their disease, particularly tying these to dread of future episodes of fracture and the ensuing pain and loss of functional abilities associated with them. The acute impacts of these

repeated, limiting fractures seemed to have given participants who were at their lowest the sense that recovery was futile in the face of all but assured future fractures and setbacks.

This sense of futility also played a role in generating frustration and anger (6/15) as well, with one participant who was diagnosed during adulthood noting, "I was angry... I fought it, and I didn't even know what the hell I was fighting." (65 y/o, M, type I) Other participants described moments of anger and frustration not with their disease but with family members, particularly caregivers, who either "smothered" them or did not attend to their medical challenges with enough care.

Phenotypic characteristics of OI are associated with feelings of isolation.

Several participants, particularly male participants with more severe disease, noted isolation separate from fracture in the area of romantic relationships, which they attributed to more visible aspects of OI (3/15). One participant (53 y/o, M, type not reported) noted his "OI forehead" and "shortness" as sources of difficulty in finding a romantic partner. On the other hand, a different participant (23 y/o, F, type IV) described romantic isolation due to the needs associated with their limited mobility and experiences of frequent fracture, having had a romantic partner leave after a particularly severe fracture. Other participants described time periods across the life course associated with feelings of isolation due to physical differences, including experiences of bullying during childhood and feelings of dissociation from bodily deformities in later life.

Unpredictability of fracture and self-image concerns results in fear and anxiety.

The most commonly expressed sources of anxiety for the participant population involved general anxieties or worry about day-to-day stressors (8/15), followed by fear of fracture (7/15). Several participants viewed their near-constant and excessive worrying as a coping response to a life filled with unpredictable injuries, including one who described "It's made me hyper aware of all my surroundings all the time since I was a child…The moment you drop the ball, the moment you let your guard down, you get hurt." (32 y/o, F, type II) There was, however, variability in the extent to which participants viewed their anxiety as adaptive, with other participants describing OI-related anxiety as "mental fragility" and "making it harder" to perform activities of daily living. While slightly less prevalent, fear of fracture in particular appears to have been a particularly strong impediment: "So basically, I stay more in my home than trying to go out just to avoid anything. Just in case." (26 y/o, F, type III) This anticipatory anxiety toward fracture may have been greater in ambulatory participants who experienced more frequent rates of fracture; several participants who lived in the northern U.S. also noted more severe fracture anxiety during the winter, when sidewalks and stairs presented increased fall risks due to ice.

Although less common than general anxieties and fear of fracture, frequent interactions with the healthcare system due to the chronic nature of OI may have been associated with health-related anxiety in this population (2/15). One participant (32 y/o, F, type II*) described being "hypersensitive" about their liver, while another (23 y/o, F, type IV) reported frequently checking health decisions with their previous physicians and "second guessing everything the doctor tells me."

Highly related to a description of fear of fracture appears to have been the experience of self-described traumatic fracture (5/15) and traumatic fall (3/15). Several participants who described a severe fracture mentioned its lasting impact on their psychosocial well-being, including one who discussed the resulting trauma: "I'll just be sitting there on the couch and I'll get like…PTSD from a fall. Or I'll start having psychological pains in my legs if I'm thinking about it." (23 y/o, F, type IV) This presentation captures the experiences of many participants with experiences of traumatic fractures, who frequently recounted episodes of pain when thinking about the event, experiences of re-living the event, and behavioral avoidance of the occurrence of the event. Participants also described experiencing social anxieties (5/15). This appears to have been primarily related to concerns over body image, which were especially salient in individuals with type III/IV OI whose disease status may have been more visible: "I have pretty bad like social anxiety. I think there's more dudes who just when I walk into a room, everyone stares at me and like, I don't like being the center of attention, but I usually am because I'm different. I mean, I'm in a wheelchair so." (26 y/o, F, type III) Similarly, "In the back of my mind was always hey, I'd like to talk to that girl but I've got my OI forehead." (53 y/o, M, type not reported) Perhaps unsurprisingly, each individual who expressed experiences of social anxiety also expressed feelings of isolation associated with their OI disease presentation, suggesting a strong association between social anxiety and visibility of OI status.

Despite the high prevalence of signs of anxiety amongst this population, only three participants reported receiving psychopharmaceutical treatment for anxiety. Two other participants reported speaking with a counselor about mental health concerns related to anxiety.

Positive orientations towards OI buffer against adverse psychosocial outcomes.

In contrast from the previous accounts of psychosocial burdens and anxieties associated with OI, nearly all participants described resilient attitudes, including an optimistic mindset (13/15), not allowing illness to define their life experience (11/15), and a belief in their own ability to overcome challenges (11/15). One striking example illustrates each of these thematic concepts: "Yeah, OI has definitely made me stronger. It's definitely pushed me to my limits. I've never let my disability define who I am. If I see someone that's really struggling…I give them that strength that OI has kind of given me. Because I mean, I know what it feels like to be down, I know what it feels like to not be able to do things or to fit in." (23 y/o, F, type IV) This participant would go on to say, "I've had over 100 broken bones and 25 surgeries. I've never let it get me down."

Participants expressed the benefits of having OI in countless different ways, extolling the empathy, patience, resilience, and strength they gained through their experience with the disease, amongst other virtues. The same experiences which caused the most distress to participants also seems to have provided them with the most strength and meaning; as one individual said, "a lot of adversity in life can either break your or it can make you strong." (53 y/o, M, type not reported) This sentiment was echoed by four other participants (5/15) whose past experiences of suffering and adversity ultimately became sources of strength.

Sources of strength for this population extended beyond individual characteristics. Through interactions with others in the OI community, many participants gained valuable insight into their own experiences. A commonly expressed sentiment was that because others had more severe OI and succeeded in spite of it, their own psychosocial burdens and physical deficits seemed more manageable.

Other positive factors related to participants' psychosocial functioning included active coping mechanisms, such as cognitive reframing of adverse events (12/15), spiritual coping practices (6/15), and exercise (6/15). Exercise was a common method of active coping in this sample. Participants who described using exercise to cope reported benefits both in their pain management and in their mood as well; two participants (32 y/o, M, type I; 23 y/o, F, type IV) discussed using exercise prophylactically to lower their risk of future fractures. Specific to cognitive reframing, one participant noted: "I mean, I would say like I kind of, I'm used to at this point. So I was, like, I just think about the positives, like, and try to just focus on the good." (26 y/o, F, type III) This cognitive behavior, as it was expressed in this sample, had a close relationship with participant characteristics of optimism and positivity, and was described only among individuals who expressed these characteristics. Spiritual coping appeared to function similarly for participants, who described their various practices as providing perspective and gratitude for the "blessings" they were given.

Finally, though increased frequency of fracture was previously associated with feelings of anger and hopelessness, it may simultaneously provide frequent opportunities for adaptation. The intense self-efficacy and belief in one's ability to overcome any challenge (11/15) was exemplified most by participants who had experienced more functionally impairing fractures, such as "long bone" fractures of the tibia, fibula, and humerus. One participant described how he, having fractured both of his arms, took a college coding exam on a computer by typing with his feet (28 y/o, M, type I).

DISCUSSION

The present study relied on a qualitative approach to better understand the negative psychosocial factors associated with adult OI disease status and identify potential protective factors within the adult OI community. The first salient theme to emerge from these interviews is that adults with OI described experiences consistent with depression and anxiety following disease-related injuries, specifically fracture. This is not to suggest that that individuals with OI are *more likely* to experience depression relative to population norms, as prior evidence demonstrates clinical equivalence of depression among those with and without OI (Tosi et al., 2015). This does suggest that fractures may have important implications in an adult OI population in relation to mood, and that these fractures may facilitate negative affect and behaviors associated with depression. Existing health literature provides evidence that injuries associated with disease status that result in functional impairments can facilitate depressive symptoms (McKnight & Kashdan, 2009; Williams & Murray, 2015). This concern appears present within an adult OI population. Regarding clinical applications, providers should be aware of this relationship between fracture and mood and assess mood immediately after fractures as well as through the course of the injury to provide support as needed.

A second finding to emerge is that adults with OI described anxiety regarding future fractures as well as anxiety specific to negative evaluation from others due to physical appearance related to OI. This future-oriented worry about disease symptom (i.e., fracture) fits within existing OI and broader health psychology literature. Fear of fractures has been documented with younger patients with OI and parents of those with OI (Alston, 1983; Hill et al., 2019; Tosi et al., 2019); however, the presence of this concern within an adult OI sample remains relatively understudied and the present results suggest this concern may extend into adulthood. It is unclear how these fears manifest. For example, are individuals avoiding engagement in life activities and if so, is such avoidance adaptive or excessive and dysfunctional? It is plausible to suggest avoidance of certain activities may be adaptive as it may prevent injury, although additional research is needed to determine when and how these types of behaviors become impairing and worsen symptoms of anxiety through active avoidance. Additional research in this area may also consider examining different age cohorts, as OI has differential impacts throughout the lifespan (Tsimicalis et al., 2016). Fear of fracture may vary across the adult lifespan as OI symptoms and treatments evolve across time.

This fear of fracture is conceptually similar to what has been observed in other populations with chronic diseases, where *fear of progression,* or the future-oriented and realistic worries specific to experiencing worsening symptoms of disease, is well documented in existing oncology literature (Dinkel & Herschbach, 2018; Herschbach et al., 2005; Herschbach and Dinkel, 2014). This construct appears to mirror what was recounted by participants in the present study and suggests that concerns specific to disease are present across different health conditions. Relative to the population at large, those diagnosed with OI are disproportionally more likely to experience fracture (Folkestad et al., 2017), and this greater base rate of fracture appears to facilitate anxiety, just as how those with cancer diagnoses are more likely to experience disease progression relative to the population at large (Bartelink et al., 2001), and in turn report disease-specific concerns. This suggests providers working with patients with OI should be mindful of this future-oriented concern, and refer for psychotherapeutic services as needed, although psychotherapeutics tailored specifically to OI remain limited, to date.

Specific to symptoms of social anxiety, the impact of OI on physical appearance does contribute to anxiety in a substantial number of participants (5/15). Social anxiety symptoms related to disease status have been documented in other health conditions where physical appearance may be different relative to others, including acromegaly (Dimopoulou et al., 2017), multiple sclerosis (Poder et al., 2009), scoliosis (Tones et al., 2006), and obesity (Dalrymple et al., 2011). Much like the aforementioned recommendations, health care providers should be cognizant of these concerns when working with OI patients and refer to psychotherapeutic services as may be indicated. Such interventions may encourage exposure to anxiogenic triggers, reframing of maladaptive cognitions, and development of other adaptive coping skills (e.g., self-advocacy).

Lastly, adults with OI reported helpful ways in which they interpret their OI status that appear to buffer against adverse psychosocial outcomes. This is consistent with prior research, where various coping strategies are employed by individuals with OI, including

children who have noted that they maintain an optimistic outlook across situations, and adults who are "making the best out of a situation" in order to cope with difficulties (Tsimicalis et al., 2016). *Illness cognition,* or the way in which an individual evaluates the experience of their disease, may be the crucial mechanism facilitating these helpful appraisals of OI disease status. This construct has been assessed in other populations with chronic conditions and is positively associated with higher quality of life outcomes, such that individuals who lessen negative experiences of their disease or may attribute positive experiences to their disease status display more favorable psychological outcomes (Gillanders et al., 2015; Evers et al., 2001). For example, studies have found that positive illness cognition played an important role in reducing psychological distress, negative emotions, and physical complaints (McCorry et al., 2013; Watson et al., 2004). In contrast, those who perceive their disease to negatively impact their life are more likely to experience depressive symptoms and engage in avoidant behaviors (Gurkova & Soosava, 2018). Understanding an individual's illness cognitions may play a critical role in identifying physical and psychological functioning, which can promote early psychological intervention (Evers et al., 2001). Consequently, assessment of illness cognition appears more limited in the context of OI. From the present study, it appears various cognitive coping strategies may be used. Further research, however, may seek to assess the benefit of these approaches. Additionally, the applicability of various treatment skills that exist in different empirically supported interventions (e.g., cognitive behavioral therapy) should be assessed within an adult OI population to facilitate adoption of these different interpretations of OI status noted in the present study, including optimism, not allowing disease status to define oneself, and believing in oneself to overcome OI specific challenges.

While the present study does shine light on psychosocial factors associated with OI in adults, it is not without limitations. Firstly, the sample size for the present study may appear small relative to what has been suggested in prior qualitative literature (Marshall et al., 2013). While notable, this may not be a concern given the specificity of the rare disease population and that saturation was reached among the collected data (Malterud et al., 2016). Further, the present sample is similar in size to what has been used in earlier qualitative OI research (Bendixen et al., 2018; Dogba et al., 2013; Hill et al., 2014). OI genotype was not represented equally in the present study and additional research should be conducted to determine whether the themes described in the present study are ubiquitous across OI types. The present sample may also have been non-representative of the OI population due to participant self-selection bias, as individuals who have experienced psychosocial challenges may have been more likely to participate. Additionally, the present sample largely identified as white; more diverse samples should be recruited in future research, as intersectionality between racial identity and disease status has been identified to contribute to unique psychosocial outcomes in other health populations (Franklin-Jackson et al., 2007; Alegria et al., 2004). Future studies may consider examining specific anxiety and depression constructs. Here, we relied on a broad approach to assess mental health symptoms among adults with OI, with injury-specific anxiety and depression appearing as salient themes; future research should tailor clinical interviews to assess these relationships in a more targeted manner. Lastly, while we note concerns of anxiety and depression need to be considered by providers working with OI patients, and referrals provided as indicated,

existing psychological intervention research specific to OI is limited, despite interventions having been tailored for other health populations. Until these interventions are developed, the true benefit of referring to mental health providers remains unknown.

CONCLUSION

The present study involved a qualitative approach to understanding key psychosocial factors among adults with OI, with three main themes emerging from the interviews. Firstly, participants reported negative affective and behavioral impacts specific to fracture and change in functionality. They additionally described feelings of anxiety specific to future fractures as well as negative social judgment from others. Lastly, adults with OI identified multiple protective factors specific to their outlook on their disease status which may help to buffer against adverse psychosocial outcomes. The findings here contribute to a small, but growing, literature designed to advance understanding of psychosocial factors specific to those diagnosed with OI. Future research may seek to build on these findings by adopting different methodologies (e.g., quantitative assessment), as well as eventually develop appropriate psychosocial interventions tailored specifically to an OI population.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Funding/Support:

Data availability:

the data that support the findings of this study may be available upon request to the corresponding author. Data are not publicly available due to privacy and ethical restrictions.

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Table 1.

Demographic information ($N = 15$) of the study sample.

	M (SD)	Min	Max
<u>Age</u>	41.2 (16.66)	20	68
	<u>N (%)</u>		
<u>Gender</u>			
Female	9 (60%)		
Male	6 (40%)		
<u>Race</u>			
White	12 (80%)		
Black/A. American	1 (6.6%)		
Multi-racial	2 (13.3%)		
American Indian/Alaska Native	0 (0%)		
Asian	0 (0%)		
<u>Ethnicity</u>			
Hispanic/Latino	4 (20%)		
Non-Hispanic/Latino	11 (80%		
<u>OI Type</u>			
Type-I	7 (46.7%)		
Type-II	1[*] (6.7%)		
Type-III	3 (20%)		
Type-IV	2 (13.3%)		
Other	1[**] (6.7%)		
Not Reported	1 (6.7%)		

Note.

[*] OI-type was self-reported; individuals diagnosed Type-II with long-term survival are often reclassified as Type-III or VI (Amor et al., 2011).

[**] Type-V

Table 2.

Theme and sub-theme frequency ($n = 15$) of the study sample.

Theme	Sub-theme	N (%)
Psychosocial Burdens Associated With Loss of Mobility	Negative affect	14 (93%)
	Negative affect after fracture	8 (53%)
	Hopelessness	6 (40%)
	Anger	6 (40%)
Phenotypic Characteristics of OI	Isolation	11 (73%)
	Romantic isolation	3 (20%)
Unpredictability of Fracture	General anxieties	8 (53%)
	Fear of fracture	7 (47%)
	Health-related anxiety	2 (14%)
	Traumatic fracture	5 (33%)
	Traumatic fall	3 (20%)
Self-Image Concerns	Social anxieties	5 (33%)
Positive Orientations Towards OI	Optimism	13 (87%)
	Minimizing disease impact on roles	11 (73%)
	Overcoming challenges	11 (73%)
	Resilience gained through adversity	5 (33%)
	Cognitive reframing	12 (80%)
	Spiritual coping	6 (40%)
	Exercise	6 (40%)