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Prevalence of mental health conditions and pain in adults with skeletal dysplasia

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Abstract

Purpose We sought to examine the prevalence of depression and anxiety in adults with skeletal dysplasias, and to assess any correlations with pain.

Methods Participation was via an anonymous REDCap survey, which consisted of sociodemographic questions followed by the brief pain inventory-short form (BPI-SF), patient health questionnaire-8 (PHQ-8), and generalized anxiety disorder-7 (GAD-7) questionnaires. These assessed pain, depression, and anxiety respectively.

Results Of the 336 usable responses, 16.1% scored 10 or greater on the PHQ-8 consistent with current depression while 17.3% scored 10 or greater on the GAD-7 consistent with current anxiety. The majority of participants (76.2%) experienced pain, which was significantly associated with prior mental health diagnoses ($p < 0.05$). A total of 34% reported either a prior diagnosis of depression or scored 10 or greater on the PHQ-8, and 31% reported either a prior diagnosis of anxiety or scored 10 or greater on the GAD-7.

Conclusions This study identified a substantial percentage of individuals with mental health concerns as well as pain in the adult skeletal dysplasia population. Further research is warranted to investigate barriers to service or treatment of mental health disorders as well as pain management.

Keywords Skeletal dysplasia · Mental health · Depression · Anxiety · Pain

Introduction

There are over 400 described forms of skeletal dysplasias, a vast majority of which involve disproportionate short stature, with the most common being achondroplasia [1]. Chronic pain and orthopedic issues necessitating surgery are common complications of these various diagnoses [2–5]. For example, achondroplasia is a spondylometaphyseal dysplasia, meaning the spine and metaphyseal regions of the bone are involved. Individuals with this condition reach an average final height of approximately 4 feet 2 in. tall. Adults with achondroplasia can develop lower back and lower extremity pain as well as difficulty in ambulation, often arising from spinal stenosis which can necessitate decompression and fusion surgery [4, 6]. Pseudoachondroplasia is a spondyloepimetaphyseal dysplasia, meaning it involves the spine, epiphyseal and metaphyseal regions of the bone, and is also characterized by short stature [7]. A majority of individuals with pseudoachondroplasia are noted to have a predisposition to joint pain that significantly impacts their quality of life, with pain starting prior to significant joint degeneration evident by radiographs [3]. Orthopedic interventions can include cervical spine decompression and fusion due to instability, and some adults with this diagnosis require hip and/or knee replacement for early onset degenerative arthritis [7]. Spondyloepiphyseal dysplasia congenita is another distinct skeletal dysplasia, with its own etiology and associated features, including marked short stature and joint pain. For individuals with this dysplasia, odontoid hypoplasia with

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concomitant atlantoaxial instability is a risk, kyphoscoliosis needing surgical fusion can occur, and hip replacements are also not uncommon in adults [8]. Overall, two studies have identified chronic pain in around three quarters of individuals with skeletal dysplasias, underscoring pain as a frequent issue for this population [2, 5].

Prior studies have measured the quality of life (QOL) of individuals with skeletal dysplasias, and a majority have found overall decreased values compared to the general population, [5, 9–12]. Discomfort was one of the major differences found in QOL subscales between groups in one study, suggesting that chronic pain and other physical symptoms experienced by people with skeletal dysplasias could adversely affect QOL [9]. Johansen et al. investigated the physical and mental health status of a short statured population that was predominately diagnosed with skeletal dysplasias, and compared them to both the general population as well as a group with rheumatoid arthritis who had chronic pain and similar physical function scores [11]. They showed that the short statured predominately skeletal dysplasia group had statistically significant lower scores in the mental health, social functioning, and vitality subscales than both other groups surveyed [11]. Additionally, a study from 2017 found a lower health-related quality of life (HRQOL) in adults with skeletal dysplasias compared to the general American population, with pain contributing to lower physical component scores, and lack of social support contributing to lower mental component scores [5]. In contrast, almost 20 years earlier, the mental component scores of adults with achondroplasia by Mahomed et al. noted no significant difference from the general population, though physical component scores were significantly lower [13].

Given these prior study findings, more research is warranted on the type and prevalence of mental health concerns in this population. Research on the prevalence of depression and anxiety in adults with skeletal dysplasias is currently limited, however there have been some investigations performed with small sample sizes and/or in specific populations. A study in Puerto Rico examined the prevalence of mental health conditions in 22 adults with achondroplasia, finding that 32% of individuals had mild to moderate symptoms of depression and 55% had mild to moderate symptoms of anxiety [14]. Another showed an increased prevalence of depression in 103 adults with chondrodysplasias when compared to a group of their unaffected siblings, but not when compared to the general population; however, a higher prevalence of anxiety was noted both compared to unaffected siblings as well as to the general population [15]. In addition, there is considerable existing literature noting the association between depression, anxiety, and chronic pain among individuals without skeletal dysplasias [16–25]. Given many individuals with skeletal dysplasia experience chronic pain [2, 5], investigating this avenue is important to understand the interplay with any mental health concerns. We sought to advance this topic further by investigating a snapshot of prevalence of anxiety and depression in a large skeletal dysplasia population, and to assess whether there is an association between these mental health conditions and pain, to then be used as a springboard for future studies on this subject.

Methods

Participants

This survey was open to adults aged 18 years and older that were English speaking, comfortable using computers, and had a self-reported diagnosis of a skeletal dysplasia. There were no further exclusion criteria. Individuals were recruited in person at the National Little People of America (LPA) Conference in Boston, MA in July 2016, as well as at a regional LPA conference in Wilmington, DE in October 2016. The LPA organization disseminated the survey link to their members electronically using e-mail and online postings on both their Facebook page and their homepage http://www.lpaonline.org. We additionally advertised the study within skeletal dysplasia clinics across the country by providing clinic coordinators a handout with the anonymous URL to any interested adults with a skeletal dysplasia diagnosis. Given that LPA is based in the United States, we would surmise that a majority of survey participants were American, but we are aware that individuals from other countries were present at the LPA national conference, and could have participated in this study there as well as online.

The study was approved by the Nemours/AI duPont Hospital for Children Institutional Review Board (#880591). This study was also approved by the Executive Board and the Medical Advisory Board of LPA, which is the largest national support group for this population.

Procedures

The survey was conducted online using REDCap with the title "Health and Wellness Survey." There was no mention of mental health, in order to help prevent bias as to who would chose to fill out the survey. At the LPA meetings and at Nemours/A.I. duPont Hospital for Children, Apple iPads were available for participants to access the survey. The survey was open from June 27, 2016 to November 12, 2016.

Measures

The validated study tools in the survey included patient health questionnaire-8 (PHQ-8), generalized anxiety disorder-7 (GAD-7), and brief pain inventory (BPI) short form, to assess the prevalence of depression, anxiety, and chronic

pain in adults with skeletal dysplasias. The PHQ-8 is derived from the PHQ-9, a validated survey to screen for depression [26]. The PHQ-8 consists of 8 questions that ask about the frequency of depressive symptoms in the last 2 weeks. Questions are scored on a scale from 0 to 3 and then totaled. The PHQ-8 has been validated and has a specificity and sensitivity of 88% for major depression with a score greater than or equal to 10 but can also detect other depressive disorders [27–29]. The GAD-7 is a seven question survey that screens for generalized anxiety disorder. Each question asks about symptom prevalence in the past 2 weeks and questions are scored on a scale of 0–3. The scores are then totaled and scores greater than or equal to 10 indicate generalized anxiety disorder with a sensitivity of 89% and a specificity of 82% but the GAD-7 is also able to detect pain, social anxiety, and post-traumatic stress disorders [29, 30]. The Brief Pain Inventory (BPI) is a published, validated survey used to assess pain and describes the location of the pain, intensity of the pain [Pain Severity Score (PSS)], and also the interference of the pain in daily life functioning [Pain Interference Score (PIS)] [31, 32]. The prevalence of pain is assessed through a yes or no answer on the BPI that states: "Throughout our lives, most of us have had pain from time to time (such as minor headaches, sprains, and toothaches). Have you had pain other than these everyday kinds of pain today?" The PSS is scored by taking the mean of the worst, least, average, and current pain in the last 24 h on a scale from 0 to 10. Scores for the PSS are categorized into mild (0–3), moderate (4–6), and severe (7–10). The PIS is calculated from the mean score of the 7 interference questions. These questions assess pain's interference with daily life activities including general activity, walking, work, relationships with others, mood, sleep, and life enjoyment.

The survey contained sociodemographic questions in addition to the three validated questionnaires. Demographic information was self-reported and included questions about age, sex, race, height, membership in Little People of America, and skeletal dysplasia diagnosis. Additional questions were asked as part of a larger study, which were not analyzed at this time. History of anxiety and depression diagnoses was assessed by the question: "Have you ever been diagnosed with a mental health condition? Please check all that apply." Former and current treatment for these diagnoses were also self-reported. Additionally, a free text comment section was added at the end of the survey after the National LPA Conference, due to participant requests.

Data analysis

Parametric and nonparametric statistical analyses were performed using SPSS software to determine the following: (1) the prevalence of depression and anxiety using frequencies, (2) correlation of pain with depression and anxiety within this population using χ^2 analysis and 2-tailed t tests, and (3) relationship of anxiety and depression in this population with various factors, including gender, age, diagnosis, and LPA membership, using χ^2 analyses. Statistical significance was considered when $p < 0.05$, and is emphasized in bold in Tables 3 and 4. Blank or missing answers to questions were excluded from analysis.

Results

A total of 528 survey responses were collected by self-report. Response rate is indeterminable as it was an advertised and not an invited survey. There were 192 surveys that were removed from analysis for the following reasons: 140 for blank surveys, 24 for incompletion of either the GAD-7 or PHQ-8 (as complete surveys are needed for scoring analytics), 16 for no diagnosis listed or a non-skeletal dysplasia listed as a diagnosis, 9 for participants under 18 years of age or no age listed, and 1 each for a duplicate response, a parent filling out the survey for their child, and for contradictory mental health information, leaving 336 usable responses. Demographic and social history information is collated in Table 1. A wide variety of skeletal dysplasia diagnoses were represented, with achondroplasia making up the majority, at 57.1%. Participants were 68.8% female, 90.2% white, and 76.8% were members of LPA for an average of 23.9 years. The average height was 48 inches, with a range of 30–64 inches. The average age of participants was 40.8 years old with a range from 18 to 81 years.

In the survey population, 28.6% of individuals denoted a prior diagnosis of depression and 25.0% listed a prior anxiety diagnosis, with 17.3% having prior diagnoses of both (Table 2). On the mental health questionnaires, 16.1% of adults scored 10 or greater on the PHQ-8 indicating depression, and 17.3% of adults scored 10 or greater on the GAD-7 indicating anxiety disorder, with 10.1% of the survey respondents scoring 10 or greater on both the GAD-7 and PHQ-8 (Table 2). Of the people who stated they had never been diagnosed with depression, 7.9% scored greater than or equal to 10 on the PHQ-8, and 8.3% of people with no noted prior anxiety diagnosis scored greater than or equal to 10 on the GAD-7.

Women were more likely than men to score 10 or higher on the PHQ-8 ($N = 336$; X^2 (1) = 4.9; $p = 0.03$), but no statistically significant sex differences were noted on the GAD-7 using χ^2 analysis ($p > 0.05$) (Table 3). Additionally, women were more likely than men to have a prior diagnosis of either depression ($N = 336$; X^2 (1) = 15.3; $p < 0.001$) or anxiety ($N = 336$; X^2 (1) = 7.8; $p = 0.005$). There was a significant difference found between age and anxiety diagnosis ($N = 336$; $X^2(3) = 9.6$; $p = 0.02$), but no statistical significance seen with age groupings and depression,

Table 1 Demographic information

	N = 336 (%)
Sex	
Men	105 (31.3)
Women	231 (68.8)
Age	
18–24	93 (27.7)
30–44	109 (32.4)
45–59	94 (28.0)
60+	40 (11.9)
Average age	40.8 years
Race[a]	
Hispanic/latino	14 (4.2)
African-American	7 (2.1)
American Indian	5 (1.5)
Asian	11 (3.3)
Pacific islander	1 (0.3)
White	303 (90.2)
Other	11 (3.3)
Little People of America Member (*N* = 335)	
Yes	258 (76.8)
Average length of membership (out of 254)	23.9 years
Diagnosis	
Achondroplasia	192 (57.1)
Cartilage hair hypoplasia	6 (1.8)
Diastrophic dysplasia	24 (7.1)
Hypochondroplasia	9 (2.7)
Metatropic dysplasia	3 (0.9)
Morquio	2 (0.6)
Osteogenesis imperfecta	5 (1.5)
Pseudoachondroplasia	19 (5.7)
Spondyloepiphyseal dysplasia (SED)	37 (11.0)
Spondyloepimetaphyseal dysplasia (SEMD)	2 (0.6)
Spondylometaphyseal dysplasia (SMD)	1 (0.3)
Other	32 (9.5)

[a]Participants could choose more than one option

PHQ-8 scores, or GAD-7 scores via χ^2 analyses ($p > 0.05$) (Table 3). There was no relationship found between LPA membership status and either prior diagnosis of or current symptoms of depression or anxiety, with $p > 0.05$ via χ^2 analyses (Table 3). Statistically significant differences were also not seen between achondroplasia and all other skeletal dysplasia diagnoses combined using χ^2 analyses for the validated survey instruments and for prior diagnoses of anxiety or depression ($p > 0.05$) (Table 3).

Information about pain was obtained via responses to the BPI. 76.2% of survey participants indicated that they experienced pain. There were no statistically significant differences seen in pain levels between men and women ($N = 332$; $X^2(1) = 0.20$; $p > 0.05$). Individuals with achondroplasia appeared to have less pain than the other dysplasia diagnoses in aggregate (72.9% vs. 80.6%), but the data did not reach statistical significance ($N = 332$; $X^2(1) = 2.6$; $p = 0.10$). The overall average Pain Severity Score (PSS) was 3.29, denoting mild to moderate pain on average, and average Pain Interference Score (PIS) was 3.03. Pain also trended upward with age, but did not reach statistical significance ($N = 332$; $X^2(3) = 7.3$; $p > 0.05$). Participants with pain were more likely to have a prior diagnosis of depression ($N = 332$; $X^2(1) = 13.3$; $p < 0.001$) or anxiety ($N = 332$; $X^2(1) = 5.3$; $p < 0.05$) (Table 3). Additionally, Table 4 illustrates that PSS averages were higher in the groups with previous diagnoses of depression ($M = 4.18$, SD = 2.03) versus no prior diagnosis ($M = 2.94$, SD = 2.30); $t(324) = -4.53$, $p < 0.001$, and with previous diagnoses of anxiety ($M = 3.99$, SD = 2.19) versus no prior diagnosis ($M = 3.06$, SD = 2.28); $t(324) = -3.24$, $p = 0.001$. PSS averages were also higher in the groups with scores of 10 or greater on the PHQ-8 ($M = 4.92$, SD = 2.16) versus scores less than 10 ($M = 2.99$, SD = 2.19); $t(324) = -5.78$, $p < 0.001$, and with scores of 10 or greater on the GAD-7 ($M = 4.13$, SD = 2.35) versus scores less than 10 ($M = 3.12$, SD = 2.25); $t(324) = -3.00$, $p < 0.01$.

Table 2 Prior depression/anxiety diagnoses and patient health questionnaire-8 (PHQ-8)/generalized anxiety disorder-7 (GAD-7) scores of 10 or greater

	N = 336 (%)	95% CI
Previous depression diagnosis	96 (28.6)	23.7, 33.4
Previous anxiety diagnosis	84 (25.0)	20.4, 29.6
Previous depression and anxiety diagnosis	58 (17.3)	13.2, 21.3
PHQ-8 score 10 or greater[a]	54 (16.1)	12.1, 20.0
GAD-7 score 10 or greater[b]	58 (17.3)	13.2, 21.3
PHQ-8 and GAD-7 score 10 or greater	34 (10.1)	6.9, 13.3
Previous depression diagnosis or PHQ-8 score 10 or greater	115 (34.2)	29.2, 39.3
Previous anxiety diagnosis or GAD-7 score 10 or greater	105 (31.3)	26.3, 36.2
Depression currently treated, with PHQ-8 score 10 or greater (*N* = 58)	23 (39.7)	27.1, 52.2
Anxiety currently treated, with GAD-7 score 10 or greater (*N* = 53)	23 (43.4)	30.1, 56.7

[a]35 individuals who scored 10 or greater on the PHQ-8 also had a prior depression diagnosis

[b]37 individuals who scored 10 or greater on the GAD-7 also had a prior anxiety diagnosis

Table 3 Relationship of anxiety/depression with various factors

	Prior diagnosis of depression (%)	χ^2 (df) p	Prior diagnosis of anxiety (%)	χ^2 (df) p	PHQ-8 ≥ 10[a] (%)	χ^2 (df) p	GAD-7 ≥ 10 (%)[b]	χ^2 (df) p
Gender		15.3 (1) **< 0.001**		7.8 (1) **0.005**		4.9 (1) **0.03**		1.7 (1) 0.20
Male ($N=105$)	14.3		15.2		9.5		13.3	
Female ($N=231$)	35.1		29.4		19.0		19.0	
Pain ($N=332$)	34.0	13.3 (1) **< 0.001**	28.1	5.3 (1) **0.02**	18.2	3.9 (1) **0.05**	18.6	2.2 (1) 0.14
Diagnosis		2.8 (1) 0.09		2.3 (1) 0.13		1.3 (1) 0.25		0.06 (1) 0.80
Achondroplasia ($N=192$)	25.0		21.9		14.1		17.7	
Other dysplasias ($N=144$)	33.3		29.2		18.8		16.7	
Age groupings		1.1 (3) 0.77		9.6 (3) **0.02**		4.6 (3) 0.20		7.1 (3) 0.07
18–29 ($N=93$)	25.8		23.7		18.3		23.7	
30–44 ($N=109$)	29.4		32.1		15.6		15.6	
45–59 ($N=94$)	31.9		25.5		19.1		18.1	
60+ ($N=40$)	25.0		7.5		5.0		5.0	
LPA membership		0.8 (1) 0.38		0.5 (1) 0.49		2.6 (1) 0.10		0.1 (1) 0.91
Yes ($N=258$)	29.8		26.0		14.3		17.4	
No ($N=77$)	24.7		22.1		22.1		16.9	

[a]Indicates a score of 10 or greater on the patient health questionnaire-8 (PHQ-8)

[b]Indicates a score of 10 or greater on the generalized anxiety disorder-7 (GAD-7)

Also shown in Table 4, PIS averages were higher in the groups with previous diagnoses of depression ($M=4.18$, SD = 2.46) versus no prior diagnosis ($M=2.56$, SD = 2.54); $t(320)=-5.21$, $p<0.001$, and with previous diagnoses of anxiety ($M=4.10$, SD = 2.64) versus no prior diagnosis ($M=2.65$, SD = 2.51); $t(320)=-4.46$, $p<0.001$. PIS averages were also higher in the groups with scores of 10 or greater on the PHQ-8 ($M=5.57$, SD = 2.58) versus scores less than 10 ($M=2.56$, SD = 2.35); $t(320)=-8.20$, $p<0.001$, and with scores of 10 or greater on the GAD-7 ($M=4.39$, SD = 2.61) versus scores less than 10 ($M=2.75$, SD = 2.54); $t(320)=-4.35$, $p<0.001$ (Table 4).

Discussion

To our knowledge, this is one of the largest known studies examining a snapshot of prevalence of anxiety and depression in a skeletal dysplasia population. Overall, 34% of the participants scored either 10 or greater on the PHQ-8 noting symptoms of depression or had a previously noted diagnosis of depression, and 31.3% of individuals scored either 10 or greater on the GAD-7 noting symptoms of anxiety or had a previously noted diagnosis of anxiety (Table 2). The percentage of mood disorders found here is similar to a prior study in an achondroplasia-only group [14].

Moreover, approximately three-fourths of individuals in the skeletal dysplasia population surveyed here noted they experienced pain. Although it has not explicitly been stated that the BPI-SF has been validated for the prevalence of chronic pain, another study has used the BPI to measure prevalence of chronic pain in the skeletal dysplasia population with similar results of 70% [2], and a subsequent study using a different instrument in the skeletal dysplasia population also noted a similar prevalence of 79% [5].

Our data from the PHQ-8 and GAD-7 additionally demonstrate that there are likely a substantial number of individuals in this population with undiagnosed or undertreated mental health conditions, for whom treatment could be advantageous. For example, we identified that 39.7% of individuals noted to be undergoing current treatment for depression still scored 10 or greater on the PHQ-8, and 43.4% of individuals currently undergoing treatment for anxiety still scored 10 or greater on the GAD-7 (Table 2).

We also show an association between mental health conditions and pain in this skeletal dysplasia cohort (Table 3), though due to study design we cannot determine causality. Given the majority of individuals surveyed here are noted

Table 4 Relationship of anxiety/depression with pain

	Prior diagnosis of depression		t (df)[a] p	Prior diagnosis of anxiety		t(df)[a] p	PHQ-8 ≥ 10[b]		t(df)[a] p	GAD7 ≥ 10[c]		t(df)[a] p
	Yes (SD)	No (SD)		Yes (SD)	No (SD)		Yes (SD)	No (SD)		Yes (SD)	No (SD)	
Pain severity score average ($N = 326$)	4.18 (2.03)	2.94 (2.30)	− 4.53(324) **< 0.001**	3.99 (2.19)	3.06 (2.28)	− 3.24(324) **0.001**	4.92 (2.16)	2.99 (2.19)	− 5.78(324) **< 0.001**	4.13 (2.35)	3.12 (2.25)	− 3.00(324) **< 0.01**
Pain interference score average ($N = 322$)	4.18 (2.46)	2.56 (2.54)	− 5.21(320) **< 0.001**	4.10 (2.64)	2.65 (2.51)	− 4.46 (320) **< 0.001**	5.57 (2.58)	2.56 (2.35)	− 8.20(320) **< 0.001**	4.39 (2.61)	2.75 (2.54)	− 4.35(320) **< 0.001**

[a]T-test sig (2-tailed)

[b]Indicates a score of 10 or greater on the Patient Health Questionnaire-8 (PHQ-8)

[c]Indicates a score of 10 or greater on the Generalized Anxiety Disorder-7 (GAD-7)

to have pain, this is an important point that should not be overlooked. We interpret our findings on the PIS and PSS as indicating that pain perceived as more severe, as well as pain that interferes with daily life functioning, is associated with the presence of depression and anxiety (Table 4). This is consistent with other research noting those with depression and anxiety experienced more severe chronic pain and greater disability [16, 21, 33]; this association has also been found in diverse cultures [18]. Bair et al. highlighted that depression and anxiety have additive as well as independent effects on pain interference, pain severity, and health-related quality of life [16]. Ohayan et al. found that pain was rated as more severe in individuals with major depressive disorder (MDD) versus non-depressed individuals and that pain intensity was more likely to be worsened by anxiety in the MDD group [21]. Williams et al. underscored the compelling relationship between pain and depression, with pain severity increasing the odds of depression, and vice versa [33]. Likewise, Kroenke et al. highlighted that a change in pain severity was a predictor of subsequent depression severity, and vice versa [19].

Prior research and current results underscore the importance of investigating both presence of and treatment for pain as well as mental health conditions in any individual [19]. In a free text comment section at the end of the survey, 14 individuals, unprompted, specifically listed untreated pain as a cause of frustration and stress, leading to difficulty with employment and decreased independence. One person also mentioned that once pain was managed appropriately, mental health took a dramatic positive turn. As this was not specifically addressed in this study, future detailed research investigating barriers to pain management in this population could provide more insight. Insurance, work stressors, disability, and social security concerns were all mentioned as sources of stress in the comments section of the survey as well. As we know, there are many contributing factors to an individual's mental health, and further study is also warranted in this area for this population.

Study limitations

This study did have limitations. While there were over a dozen different diagnoses represented in this study, over half of the study participants were diagnosed with achondroplasia. Given the comparatively small sample sizes of the other dysplasias, it was not statistically possible to analyze each of them independently against all the others. However, given there was no statistical difference in the scores between achondroplasia and the other dysplasias in aggregate (see Table 3), we felt it appropriate to combine all for analysis. This survey was completed through self-report, which introduces the possibility of incorrect responses and biased

results. For example, the skeletal dysplasia diagnoses were self-reported and not medically confirmed. The survey participants were predominantly white, and female, so these results may not be representative of the entire skeletal dysplasia population. Additionally, approximately three-fourths of the participants were members of Little People of America, a national support group for individuals of short stature. Though this may introduce biased results as members of a support group may be more "supported" and less likely to have mental health concerns, it is also possible that the opposite is true, as individuals who find they need more support are the ones that reach out to participate in such groups. We also tried to overcome volunteer bias by giving the mental health survey a neutral name; however, since this survey was distributed mainly through LPA, there could be inherent response bias by individuals in LPA telling their acquaintances what the survey was about before they volunteered to complete it. However, via Chi-square analyses, there were no statistically significant differences in the survey tool results between LPA and non-LPA members (see Table 3), so these are likely non-issues.

Practice implications

Skeletal dysplasias are a complex and diverse group of conditions with specific healthcare needs from specialists familiar with these rare diagnoses. The US Preventive Services Task Force recommends screening all adults in the general population for depression [34]. This study underscores that the skeletal dysplasia population is no different. We encourage health-care professionals to screen and subsequently refer adults with skeletal dysplasias for treatment of mood disorders, as well as anxiety, as appropriate. This study also underscores the importance of assessing for chronic pain, and to optimize pain management in concert with any mental health screening and treatment.

Research recommendations

Cause and effect cannot be determined based on the cross-sectional study design; however, the percentage of individuals with mental health concerns as well as pain is significant in the adult skeletal dysplasia population, and this warrants further inquiry. As noted above, we encourage future research into any barriers to service or treatment for mental health conditions that this population may face as well as encourage additional research into some of the likely upstream contributors to these issues such as lack of appropriate management of chronic pain [19]. Given the interplay between depression and anxiety with pain perception and disability, research into optimized integrated approaches to treatment would be ideal [16]. Finally, education and open conversation should be fostered in order to reduce stigma surrounding mental health conditions in general. We hope overall that the study itself encouraged dialogue on this topic and that additional research will ultimately help those in need.

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Compliance with ethical standards

Conflict of interest Colleen P. Ditro, DNP RN CPNP, Michael B. Bober, MD PhD, William G. Mackenzie MD, and Angela L. Duker, MS LCGC, are members of the Little People of America Medical Advisory Board. Otherwise, authors Sarah Jennings, MS LCGC, Colleen P. Ditro, DNP RN CPNP, Michael B. Bober, MD PhD, William G. Mackenzie, MD, Kenneth Rogers, PhD LAT ATC, Laura Conway, PhD MS LCGC, and Angela L. Duker, MS LCGC declare that they have no conflict of interest.

Ethical approval All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000.

Informed consent Informed consent was obtained from all individuals included in the study. The requirement for documentation of informed consent was waived based on the applicable federal regulation.

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