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Quality of life in osteogenesis imperfecta

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Abstract The purpose of this study was to quantify the physical and mental health of a diverse adult cohort of patients with osteogenesis imperfecta (OI) utilizing a validated health self-assessment questionnaire (SF-36). In addition, a specific demographic questionnaire and a functional questionnaire were utilized to assess more specifically the physical limitations imposed by osteogenesis imperfecta in adulthood. The results of the SF-36 revealed significantly lower physical function scores compared to the U.S. adult norms. However, the SF-36 mental component scores were equal to the U.S. adult norms. The demographic questionnaire revealed high levels of educational achievement, as well as employment, despite significant physical impairments. The functional questionnaire clearly demonstrated limitations mostly related to ambulation.

Résumé Le but de cette étude était mesurer la santé physique et mentale d'une cohorte de malades adultes avec osteogenesis imperfecta utilisant un questionnaire validée de l'estimation de la qualité de vie (SF-36). De plus, un questionnaire démographique spécifique et un questionnaire utilitaire ont été utilisés pour répartir plus spécifiquement les limitations physiques imposées par l'osteogénèse imparfaite à l'âge adulte. Les résultats du SF-36 ont révélé des scores de la fonction physique nettement inférieurs aux normes adultes Américaines. Cependant, le composant mental du score SF-36 était équivalent aux normes adultes Américaines. Le questionnaire démographique a révélé des hauts niveaux d'étude et d'emploi en dépit d'affaiblissements physiques considérables. Le questionnaire utilitaire a montré des limitations principalement liées à l'ambulation.

Keywords Osteogenesis Imperfecta · Quality of Life · SF-36

Introduction

Osteogenesis imperfecta (OI) is a genetically heterogeneous group of congenital disorders of collagen synthesis characterized by generalized osteoporosis and multiple fractures [5]. Other features of this disease include scoliosis, hearing loss, dentinogenesis imperfecta, and generalized ligamentous laxity. The impact of OI on the individual is greatly influenced by the severity of the disease. The spectrum of disease has been outlined by Sillence [13] and ranges from a severe type that results in perinatal death to a minimally affected type that results in mild bone fragility.

It is acknowledged that the physical limitations imposed by OI may influence social development, self-image and independence. These factors were analyzed by Shea-Landry and Cole in 1986 [12] and summarized by Cole in 1993 [4]. Cole, in a study of psychosocial aspects of OI, stated that, "The extent to which these [physical] differences alter lifestyle depend on the severity of the disorder, its natural history, the extent to which it affects social integration, the effect on physical appearance, and the presence of other affected family members." Others have noted anecdotally that individuals with OI have a low incidence of depression and high potential for social achievement [8, 10].

Much has been written about the molecular basis of OI and both medical and surgical treatment for the disorder. Few studies have addressed function and quality of life of individuals with OI in adulthood [8]. We assessed previously the specific interrelationship between spinal deformity, pulmonary compromise, and quality of life patients with OI in a subset of the current study group [17]. The purpose of this study was to attempt to quantify the physical and mental health of a larger diverse adult cohort of patients with OI utilizing the SF-36 and specific functional and demographic questionnaires.

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Material and methods

This is a present cross-sectional survey in which participants completed three different questionnaires. After approval from the Institutional Review Board, every individual 17 years of age or older with OI followed at our institution was invited to participate. Forty-two subjects fit the inclusion criteria. One patient, not followed at our institution, volunteered to participate after hearing about the study and was included. An introductory letter was sent to each participant. Those responding affirmatively received the complete package of questionnaires and consent forms by mail. Thirty people (21 women and nine men) completed the questionnaires and were included in this study. Their average age was 33 years (range 20–50 years).

Three different questionnaires were utilized: (1) The Short-Form 36 health survey (SF-36), (2) a specific demographic questionnaire for OI designed for this study, and (3) one functional questionnaire: the Functional Independence Measure (FIM).

The SF-36 is a self-reported health assessment tool composed of 36 questions in eight different domains that examine aspects of physical and mental health and is widely used to measure health outcomes in medicine. The four main physical domains are Physical Function (PF), Role Physical (RF), Bodily Pain (BP), and General Health (GH). The four main mental domains are Vitality (VT), Social Function (SF), Role Emotional (RE), and Mental Health (MH). Each domain is scored from 0 to 100. A higher score is correlated with better mental and physical health. The physical and mental domains may be combined in two broad scores, the Physical Component Summary (PCS) and the Mental Component Summary (MCS) respectively. These two combined domains are obtained by multiplying each SF-36 scale z-score (obtained from the SF-36 scale score of the U.S. population) by their physical factor score coefficient and adding the products of the eight domains. In summary, these aggregate scores reflect self-assessed physical and mental activity. The SF-36 also has been tested extensively for reliability and validity [6, 7, 14, 15, 16].

The specific questionnaire for OI was designed for this study. This questionnaire included demographic information addressing the timing of first fracture, total number of fractures, comorbid diseases, family history, surgical history, academic and professional achievements, living environment, and the like.

The FIM [18] is a multidimensional functional scale including areas of cognition, speech, mobility, and activities of daily living (ADL). The FIM is widely used in rehabilitation settings. The ADL portion of the FIM was administered to assess traditional ADL skills including eating, grooming, bathing, dressing lower and upper body, toileting, toilet transfers, and in-home transfers. FIM scoring considers all levels of dependence including the need for increased time for ADLs and the use of assistive devices.

Patients were divided into two main clinical groups based on the time of the fracture: (1) those who had their first fracture diagnosed before or during birth were included in the congenita group, and (2) those with their first fracture after birth were placed in the tarda group.

Statistical analysis was performed utilizing the EPI INFO 6 Version 6.0 statistical software package. Statistical significance was considered to be a *P* value equal to or less than 0.05 for a two-tailed test.

Results

Specific questionnaire results

A total of 30 individuals with OI participated in this study. The summary data for the group as a whole is presented in Tables 1 and 2. We utilized a simple classification scheme in which congenita was defined as the presence of fractures at or prior to birth, and tarda was de-

Table 1 Summary of demographic and health data. *UE* upper extremity, *LE* lower extremity, *OI* osteogenesis imperfecta. *Statistically significant difference

	OI study population	Congenita	Tarda
Number of cases	30	18	12
Average age (years)	33.4	32.6	34.4
Other OI siblings	10%	17%	8%
Over 30 fractures	57%	67%	41%
UE surgeries ^a	13%	17%	8%
LE surgeries ^b	37%	39%	33%
Hearing deficit	30%	67%	*25%
Dental problem	60%	72%	42%
Scoliosis	53%	61%	42%
Back pain	50%	50%	50%
Depression ^c	27%	13%	33%

^a Percentage of cases with more than one UE surgery

^b Percentage of cases with more than ten LE surgeries

^c Percentage of cases receiving medical treatment

Table 2 Summary of social data and mobility status. *OI* osteogenesis imperfecta. *Statistically significant difference

	OI Study population	U.S. norm ^d	Congenita	Tarda
Number of cases	30		18	12
Average age (years)	33.4		32.6	34.4
Education ^a				
Not high school	0%	18%	0%	0%
High school	20%	34%	16%	25%
Some college	23%	17%	29%	17%
College	34%	22%	39%	25%
Advanced degree	23%	8%	16%	33%
Marriage				
Married ^b	43%		28%	67%*
Sexual limitations	37%		50%	17%
Had children	23%		22%	25%
Employment				
Employment/ population ratio	57%	63%	50%	67%
Unemployed	32%	6%	40%	20%
Not in labor force	17%	33%	17%	16%
Mobility				
Able to ambulate ^c	37%		17%	67%*
Unable to ambulate	63%		83%	33%*

^a Indicates full completion

^b Includes married, living with significant other, and divorced/separated

^c Includes any person able to ambulate with or without assistive devices

^d Year 1995 from the "Statistical Abstract of the U.S. 1996" (116th edn)

defined as the presence of fractures noted after birth. The tarda group had a significantly higher rate of independent ambulation. Hearing deficit was a significantly greater problem in the congenita group and sexual limitations were reported by 50% of this group.

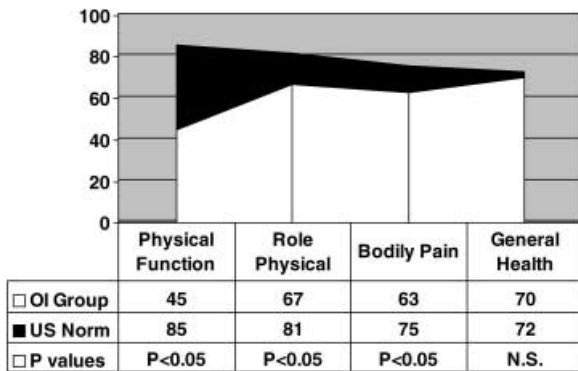


Fig. 1 Physical domain scores of the OI group compared to the U.S. norm. Significant differences are found in Physical Function (PF), Bodily Pain (BP), and General Health (GH). NS nonsignificant

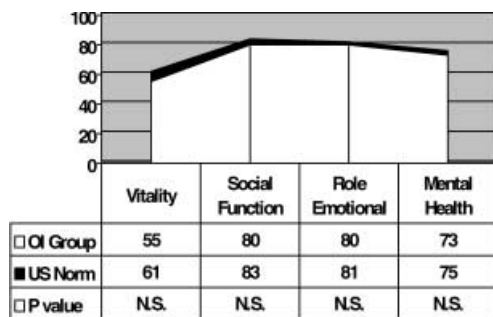


Fig. 2 Mental domain scores of the OI group compared to the U.S. norm. No significant difference could be found in Vitality (V), Social Functioning (SF), Role Emotional (RE), and Mental Health (MH). NS nonsignificant

The study population, as a whole, demonstrated remarkable levels of achievement in terms of higher education compared to the U.S. Census Bureau data (Table 2). All participants graduated from high school and 80% had attended some college. Employment rates were comparable to the U.S. population but the unemployment rate was somewhat higher.

SF-36 results

SF-36 scores were obtained and compared to those of the general U.S. population (Figs. 1, 2, and 3). Our study group of 30 individuals had significantly lower scores than the U.S. adult norm in three of the four domains measuring physical health – PF, RP, and BP. No significant differences between scores were found in those scales measuring mental health.

Functional questionnaire results

Scores were converted to a percent able score to define ability. The FIM described the patients as 97% able (range 60–100%) with only seven of the 23 subjects hav-

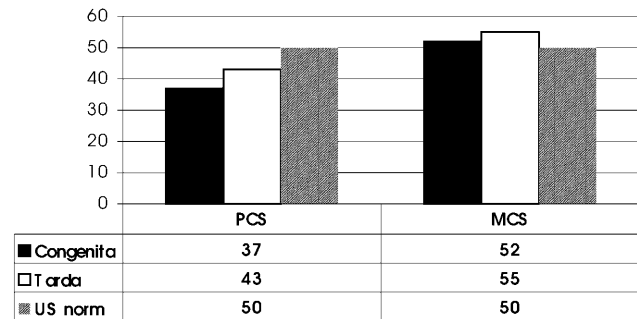


Fig. 3 Physical summary scores were significantly lower than the U.S. norm whereas the mental summary scores were higher than the U.S. norm but not statistically different

ing any deficit according the FIM ADL subset. Results indicate that the study population was very functional despite described musculoskeletal limitations.

Discussion

Previous authors have subjectively evaluated the psychological and social implications of OI [2, 4, 8, 10, 12]. This study represents an initial attempt to quantify quality of life and social and functional outcomes in a small heterogeneous group of skeletally mature patients with OI.

Reite et al. in 1972 [10] performed a psychological evaluation of 12 children with severe OI. They concluded that the profound disturbance of early motor development did not necessarily exert an adverse influence on emotional and cognitive development.

Moorefield and Miller [8] performed a retrospective review of 31 adults with OI who were 17 years and older and at least ten years out from the last major long bone surgery. They performed a clinical evaluation that included evaluation of education and employment. In this study 26% of the patients were nonambulatory and wheelchair bound. Despite motor disabilities these adults were “productive and socially adaptable individuals.” Sixty-five percent of the individuals were employed at the time of the study, and the authors commented on the “remarkable numbers” of individuals who had received education beyond the high school level. Thirty-five percent of the study participants were married.

Bauze [2] noted that the extent of disability in an OI study population of 42 individuals ages 2–65 years corresponded closely with the severity of the underlying disease.

In a previously published study [17] we evaluated the impact of scoliosis and pulmonary compromise on quality of life in OI patients. Scoliosis and pulmonary compromise were correlated with lower physical component scores on the SF-36. The authors are aware of no other studies of OI that address quality of life using validated outcomes instruments. Our current larger study population of 30 adults with OI reported significantly impaired

physical function compared to the adult U.S. norm. However, the mental component scores were comparable to the adult U.S. norm. Individuals with congenita type OI had a significantly lower PCS than those with tarda type OI, but the MCS scores were similar.

The high educational achievements of our study population corroborate the findings of others [8, 10]. All of the individuals completed high school and 57% achieved college or postgraduate degrees. It should be noted that selection bias may favor participation of more educated and literate patients in extensive outcomes studies and surveys. The high employment rate of our study population also parallels the educational achievements of these individuals. Although the ability to ambulate independently was not correlated with higher academic achievement, the inability to ambulate independently was correlated with higher rates of unemployment – 40% of wheelchair users never had a job as opposed to 0% of the ambulators.

Our study population noted that dental problems (60%), back pain (50%), and hearing deficit (30%) were significant health problems. This affirms the findings of Ault and Sillence [1]. In a 20-year follow-up study of 37 patients conducted in Australia, Sillence found that foot pain, hearing deficit, and back pain were the most common physical complaints.

The functional status of patients with OI has often been defined by the ability to ambulate, the presence of contractures, or the occurrence of secondary deformity such as bowing or scoliosis [3, 8, 11]. The literature lacks traditional ADL assessments in the OI population. The ADL section of the FIM was administered to our study population in an attempt to define traditional functional status. A technical adaptation was required because the assessment recommends face-to-face testing and our geographically diverse population required assessments be administered by patient report collected through the mail.

Although the FIM was designed as a comprehensive evaluation, its use as a unidimensional subset scale has been discussed in the literature as an option in assessing segments of function [9]. The FIM described the patients as 97% able (range 60–100%) with only seven of the 23 subjects having any deficit according to the FIM ADL subset. These results indicated that the study population was overall very functional in spite of the described musculoskeletal limitations.

We are aware that the classification of congenita and tarda is not widely used today. There are several reasons for its use in this work: its simplicity, the small number of patients, the difficulty in differentiating between types III and IV of Sillence in certain cases, and for statistical reasons.

The conclusions of this study should be applied with caution since its sample population is not truly a reflection of the total OI population but of a subset of patients receiving ongoing orthopedic follow-up.

Our study helps to quantify the physical, mental, social, and functional limitations imposed by OI. Despite significant measurable physical limitations individuals demonstrated high social achievement including high

levels of academic achievement and employment. Ambulatory status did not correlate with overall health status or academic achievement, but inability to ambulate was correlated with a higher rate of unemployment. Further study with a larger cohort of patients may allow us to assess the impact of surgical intervention on quality of life in individuals with OI.

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