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PEDSQL 4.0 in Children with Osteogenesis Imperfecta After the Covid-19 Pandemic

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ABSTRACT

Background: Osteogenesis Imperfecta (OI) is an autosomal dominant genetic disorder caused by mutations in the COL1A1 and COL1A2 genes, resulting in brittle bones and an increased risk of fractures. Patients with OI often experience acute and chronic pain, which significantly reduces their quality of life (QoL). During the COVID-19 pandemic, social distancing policies limited patient access to healthcare services. Consequently, therapy for OI patients during the first year of the pandemic decreased, further negatively impacting their QoL. ***Methods:*** This study is a qualitative descriptive research with a cross-sectional approach. The sample consisted of OI patients aged 2–18 years. A total of 26 patients were included based on the following inclusion criteria: OI patients receiving care at the Pediatric Endocrinology Clinic of Dr. Soetomo Hospital, and parents who were able to communicate with the researchers and complete the questionnaire. Patients treated in the PICU were excluded. The variables studied included parents' age, parents' educational background, children's gender, bisphosphonate or vitamin D therapy, and COVID-19 history. ***Results:*** Most patients were aged 8–12 years (34.61%) and were male (57.7%). The majority of parents were aged 15–40 years (57.7%). OI patients who received bisphosphonate or vitamin D therapy had better overall quality of life (QoL) scores compared to those who did not receive treatment. However, the average overall QoL scores were particularly low in the domain of physical functioning. ***Conclusion:*** The quality of life of Osteogenesis Imperfecta patients after the COVID-19 pandemic at Dr. Soetomo Hospital, Surabaya, remains low.

Keywords: osteogenesis imperfecta; pediatric quality of life 4.0 (PedsQL 4.0); covid-19.

INTRODUCTION

Osteogenesis Imperfecta (OI) is a genetic disorder inherited in an autosomal dominant manner caused by mutations in the COL1A1 and COL1A2 genes. Approximately 90% of the 3,000 OI patients have mutations in COL1A1 or COL1A2 [1]. The COL1A1 and COL1A2 genes form the molecular structure of type I collagen and extracellular components for bones and skin. These mutations affect the formation of bone connective tissue, making bones brittle and fragile, and are associated with osteopenia, hearing loss, skin disorders, dentinogenesis imperfecta, and blue sclera [2].

According to a study conducted in Utah, United States, the incidence of Osteogenesis Imperfecta (OI) is 0.79 per 10,000 live births [3]. Based on the Clinical Practice Guidelines of the Indonesian Pediatric Society, the detected incidence of OI ranges from 1:20,000 to 1:50,000 live births. This incidence is not influenced by the patient's gender or ethnicity [4]. OI patients are characterized by increased susceptibility to fractures and heightened bone fragility. This is supported by a study conducted at Cipto Mangunkusumo Hospital in Jakarta, where all 41 OI patients reported a history of recurrent fractures [5].

Osteogenesis Imperfecta (OI) requires regular medical check-ups to evaluate the clinical condition and manage treatment effectively. Calcium carbonate (50 mg/kg/day) is administered to prevent hypocalcemia, with the dosage adjusted based on regular laboratory assessments of the patient's serum calcium levels [6]. A study conducted at Dr. Soetomo Hospital, Surabaya, involving 20 OI patients undergoing zoledronate therapy every six months for two years, showed improvements in the quality of life of OI patients [7].

COVID-19 was first reported in Wuhan, China, in 2019 and subsequently spread globally. Social restriction policies implemented by governments significantly reduced patient visits to hospitals and other healthcare services [8]. Patients undergoing routine treatments reported delays in receiving their care due to the impact of the COVID-19 pandemic [9].

The Pediatric Quality of Life Inventory 4.0 (PedsQL 4.0) is designed for pediatric patients aged 2–18 years and can be applied to children with both acute and chronic illnesses [10]. Quality of life is studied to understand the impact of disease or treatment on an individual's life [11]. Calcium and/or vitamin D supplementation in Osteogenesis Imperfecta patients decreased during the first year of the COVID-19 pandemic, which consequently affected the patients' Quality of Life (QoL) [12].

METHODS

Children aged 2–18 years with Osteogenesis Imperfecta, both male and female, were included in this study. The patients were registered at the Pediatric Endocrinology Clinic of Dr. Soetomo Hospital, Surabaya. This study used the PedsQL 4.0 questionnaire, which had been translated into Indonesian. The questionnaire consists of 23 questions covering physical, emotional, social, and school functioning.

The questionnaire results were collected using purposive sampling, with inclusion criteria being Osteogenesis Imperfecta patients registered at the Pediatric Endocrinology Clinic of Dr. Soetomo Hospital, Surabaya, and parents and patients who were willing and able to communicate with the researchers. OI patients in the PICU were excluded. This study was approved by the Ethics Committee of Dr. Soetomo Hospital, Surabaya (3179/105/3/IX/2024).

RESULTS

The characteristics of the patients in this study included parental age, parental education level, child's age, child's gender, bisphosphonate/vitamin D therapy, and the COVID-19 history of the Osteogenesis Imperfecta (OI) patients. These characteristics were assessed using the Pediatric Quality of Life Inventory 4.0 (PedsQL 4.0) and are described in TABLE 1.

A total of 26 patients were included in the study. The majority of parents were in the age range of 25–40 years, with 15 individuals (57.7%). Most parents had a high school education or equivalent, comprising 12 individuals (46.2%). Among the child patients, the largest age group was 8–12 years, with 9 individuals (34.62%). Male patients constituted the majority of OI participants, with 16 individuals (61.54%).

Out of 26 OI patients, 5 individuals (19.23%) were not undergoing bisphosphonate or vitamin D therapy. In this study, none of the OI patient respondents had a positive history of the Coronavirus. All 26 individuals (100%) had no history of COVID-19, as confirmed through antigen swab tests, PCR swab tests, or rapid antibody tests.

OI patients with parents aged 25–40 years had a higher total QoL score, averaging 66.83. Patients with parents in this age range also showed higher physical functioning scores. The highest quality of life, based on total QoL scores, was observed in the group of parents with a college-level education, with a score of 71.4. The highest physical functioning score was found in the group of parents with an elementary school-level education (72.92), while the lowest score was recorded in the group with a middle school-level education (61.46).

OI patients aged 2–4 years had the highest total QoL score among all age groups, with a score of 77.9. This age group also achieved the highest scores in physical, emotional, and school functioning domains. The lowest physical functioning score was observed in the 13–18 years age group, with a score of only 37.71. The total Quality of Life (QoL) score for male pediatric respondents was 69.03, which was higher than the total QoL score for female respondents at 59.47. However, the physical functioning domain score was higher among female respondents compared to males, with a score of 54.26.

Patients undergoing therapy had superior scores across all QoL domains: physical functioning (57.29), emotional functioning (66.9), social functioning (75), school functioning (68.8), and total QoL (67), compared to those who were not receiving therapy. To evaluate the Quality of Life (QoL) of patients with Osteogenesis Imperfecta (OI) following the COVID-19 pandemic, respondents were categorized into two groups:

TABLE 1: Characteristics of Research Subjects.

Characteristics	Category	Frequency (n)	Percentage (%)
Parental age (years)	25 - 40	15	57.7%
	41 - 60	11	42.3%
Parental education level	Elementary School	3	11.24%
	Junior High School	3	11.24%
	Senior High School	12	46.2%
	Higher Education	8	31.32%

Characteristics	Category	Frequency (n)	Percentage (%)
Patient's age (years)	2 - 4	5	19.23%
	5 - 7	8	30.85%
	8 - 12	9	34.62%
	13 - 18	4	15.3%
Patient's gender	Female	10	38.46%
	Male	16	61.54%
Bisphosphonate/ vitamin D therapy	No	5	19.23%
	Yes	21	80.77%
COVID-19 History	Yes	0	0%
	No	26	100%

Those with a history of COVID-19 and those without. All respondents reported no history of COVID-19 within the past year. The overall QoL score among OI patients were 62.41.

The highest mean QoL domain score was observed in social functioning (74.79), whereas the lowest was in physical functioning (50.52).

TABLE 2: Quality of Life of Osteogenesis Imperfecta Patients.

Characteristics	n (%)	Physical Functioning	Emotional Functioning	Social Functioning	School Functioning	Total QoL
Parental age (years)						
25 - 40	15 (57.7%)	70	62.67	71	63.67	66.83
41 - 60	11 (42.3%)	64.9	57.72	63.32	64.54	63.28
Parental education level						
Elementary School	3 (11.24%)	72.92	55	60	43.33	57.81
Junior High School	3 (11.24%)	61.46	56.67	63.33	83.33	66.2
Senior High School	12 (46.2%)	66.41	57.5	69.37	55.65	62.22
Higher Education	8 (31.32%)	71.88	66.87	75	71.87	71.4
Patient's age (years)						
2 - 4	5 (19.23%)	70.63	75	66	100	77.9
5 - 7	8 (30.85%)	59.4	59.35	83.75	58.1	65.15
8 - 12	9 (34.61%)	48.33	69.53	72.11	61.93	62.97
13 - 18	4 (15.4%)	37.71	57.05	62.9	65	55.66
Patient's gender						
Female	11 (38.46%)	54.26	57.72	62.27	63.63	59.47
Male	15 (61.54%)	53.13	70.67	80.67	71.67	69.03
Bisphosphonate/vitamin D therapy						
No	5 (19.23%)	38.13	58	64	66	56.53
Yes	21 (80.77%)	57.29	66.9	75	68.8	67
COVID-19 History						
Yes	0 (0%)	0	0	0	0	0
No	26 (100%)	53.6	65.19	72.88	68.26	64.98

TABLE 3: Average Quality of Life of Osteogenesis Imperfecta Patients.

Characteristics	Average QoL
Physical functioning	50.52
Emotional functioning	63.27
Social functioning	74.79
School functioning	61.05
Total QoL	62.41

DISCUSSIONS

Parental Age

In this study, parents aged 25-40 years had a total QoL score higher than the parents aged 41-60 years group. Parents aged 25-40 years (57.7%) in this study indicate that OI patients have higher physical, emotional, and social functioning.

The role of parents or guardians as caregivers is associated with the quality of life (QoL) of children [13]. A study by Utami et al. (2023) explained that parents aged >45 years tend to have children with better QoL. However, this study found that parents aged 25–40 years had higher total QoL scores compared to those in the 41–60 years age group. Parental age reflects their ability to provide care and support, particularly for children with chronic illnesses [14].

Parental Education Level

Based on TABLE 2, most of the parental education levels of the OI patients were in Senior High School as many as 12 (46.2%). However, the QoL score is higher in parents in the Higher Education group with a score of 71.4, while the parents Senior High School group have a total score of 62.22. Parents with a history of education in Elementary School have the lowest total QoL score with a score of 57.81.

The highest total QoL score was observed in the group of parents with a higher education level, scoring 71.4. Higher parental education levels were associated with better physical functioning. Parents with higher education are more likely to seek professional assistance for their child's care, which can significantly improve the child's quality of life [15].

Patient's Age

According to the PedsQL 4.0 questionnaire, pediatric patients are divided into four groups, namely ages 2-4 years old, 5-7 years old, 8-12 years old, and 13-18 years old. In this study, OI patients aged 2-4 years have the highest total QoL score, while OI patients aged 13-18 years have the lowest total QoL score. In addition, physical functions in OI patients aged 13-18 years are the lowest among the patient's age group with a score of 37.71. The school function in OI patients aged 2-4 years old is considered to be no obstacle since they have not started school.

The age of children with Osteogenesis Imperfecta (OI) can reflect the severity of symptoms experienced. Earlier onset of symptoms is associated with greater impairment in physical functioning [16].

A study by Vanz et al. reported a positive correlation between age and social functioning. However, differing from those findings, this study observed that the highest social functioning score (83.75) was found in the 5–7 years age group, while the lowest score (62.9) was recorded in the 13–18 years age group [17].

Patient's Gender

The total QoL score for male pediatric patients was higher than that of female patients, with scores of 69.03 and 59.47, respectively. A study by Song et al. reported that QoL scores did not differ significantly between male and female patients [18]. However, genetically, male OI patients are more likely to experience mutations in the COL1A2 gene compared to females, which may affect the severity of symptoms reported by patients [19].

Bisphosphonate/Vitamin D Therapy

The total QoL score for OI patients undergoing bisphosphonate/vitamin D therapy was higher than those not receiving therapy, with a score of 67. According to Vanz et al., the use of alendronate improves the well-being of OI patients by reducing pain [17]. The primary goal of OI treatment is to increase bone mineral density (BMD), which is why osteoporosis medications are commonly used. Therapies such as pamidronate and alendronate have been shown to enhance BMD, thereby improving the mobility of OI patients [20].

COVID-19 History

In this study, no history of COVID-19 was reported among Osteogenesis Imperfecta (OI) patients within the past year. OI patients without a history of COVID-19 had a total QoL score of 64.98. In cases of pulmonary diseases, clinical signs and symptoms of OI might be affected based on its type. OI type III patients show a decrease in tidal volume compared to OI type I and IV due to the inadequacy of rib cartilage expansion. [21]

Average Quality of Life in Osteogenesis Imperfecta Patients

The average total QoL score for Osteogenesis Imperfecta (OI) patients at Dr. Soetomo Hospital after the COVID-19 pandemic was 62.41. Since the total QoL score does not reach 100, it indicates that the patient's quality of life is significantly impaired. The lowest QoL domain score was physical functioning, with an average score of 50.52. A history of fractures can result in physical inactivity, leading to decreased muscle function. This is a contributing factor to the tendency for lower physical functioning scores in OI patients [22].

CONCLUSIONS

The quality of life (QoL) of Osteogenesis Imperfecta (OI) patients after the COVID-19 pandemic remains low. The QoL of OI patients, based on parental age, parental education level, child's age, child's gender, bisphosphonate/vitamin D therapy, and COVID-19 history, is still low, with physical functioning being the lowest-scoring QoL domain.

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