Mothers' Role Regarding Care of their Children Suffering from Osteogenesis Imperfecta

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Abstract

Background: Osteogenesis Imperfecta (OI) known as brittle bone disease, it is a genetic disorder of connective tissue which is characterized by bones that fracture easily from little or no apparent trauma. However, the multiple fractures will lead to progressive bone deformities, short stature and restricted mobility. This study **aimed** to assess mothers' role regarding care of their children suffering from OI. **Research design:** A descriptive design was utilized. **Setting:** The study was conducted at Genetic Department in Children Hospital affiliated to Ain Shams University Hospitals. **Subjects** consisted of 80 mothers accompanying their children suffering from OI. **Tools:** Data were collected through using. **Tool (1):** Interviewing questionnaire tool it was consisted of: **Part 1:** Characteristics of the study subjects. **Part 2:** Knowledge of mothers regarding to OI. **Tool (2):** mothers' reported practices regarding to care of their children suffering from OI. **The results** of the study revealed that, the mean age of studied mothers was 32.82±5.3 years; the mean age of the studied children was 8.14±5.71years. Majority of studied mothers had negative family history of OI. Also, less than two thirds of them had consanguinity relation. **Conclusion:** This study concluded that less than half of the studied mothers had satisfactory knowledge and less than half of them had adequate reported practices regarding to care of their children suffering from OI. **Recommendation:** Educational program intervention should be conducted for mothers of children with OI to raise their level of knowledge and practices regarding caring of their children with OI.

Keywords: Mothers role, Children, Osteogenesis Imperfecta.

Introduction:

Osteogenesis Imperfecta (OI) is a chronic, genetic condition frequently described as "brittle bones." This condition is expressed by low bone density and characterized by frequent fractures with or without trauma, in addition to pain, altered growth, challenges with mobility, deformity and disability **(Wiggins & Kreikemeier, 2017).**

The worldwide incidence of OI is approximately 1/15,000 to 1/20,000, including children diagnosed after birth. OI occurs with equal frequency among males and females and among all racial and ethnic groups. In Egypt, the prevalence of the disease in children at genetic department at Children's Hospitals affiliated to Ain Shams University in the year 2017 were about 100 children **(Statistical Office of Genetic department of Children's Hospitals of Ain Shams University, 2018).**

All family members take an active role in keeping the child safe from fractures. They face worries that even basic handling and care could result in a fracture. Parents may doubt in their ability to care for their fragile child and wanted additional support. However, the diagnoses of OI come as a shock and the parents will be unaware of the implications for their child **(Santos et al., 2018).**

Nurses play a key role in guidance and support the families and ensuring that the multidisciplinary team is involved and helping to instigate a policy of continuity of care for children and their families. So, the earliest nursing intervention should be at the time of diagnosis **(McDowell et al., 2019).**

The nurse has very important role to educate the parents regarding the likelihood of survival and what to expect regarding deformity, disability, and ambulatory capacity. Genetic counseling and prenatal screening including ultrasonography may be necessary during future pregnancies **(Zhytnik et al., 2020).**

Nurses should be able to provide general information about the child condition and parents should be encouraged to participate in child's care to encourage confidence and increase understanding of handling and management. It is also important to help families find support for coping with their feelings and referral to family social service **(Bishop, 2018).**

Significance of the study:

Osteogenesis Imperfecta (OI) is a rare bone disorder characterized by bone fragility (fracturing with little-to-no trauma), short stature, long bone deformities, bone pain, low muscle mass. The prevelance of OI in children at Genetic Out- Patient Clinic in Children's Hospitals affiliated to Ain Shams University in the last year 2017 were about 100 child.

Children with OI are considered to affect the whole family, requiring new modes of organization and structure for the family. So, this study was conducted to reduce fracture rates and bone fragility while maximizing mobility and improving knowledge and practices of mothers having children suffering from OI and encourage parents to seek professional help if they seem to be overwhelmed or excessively sad or angry.

Aim of the study:

The aim of the study was to assess mothers' role regarding care of their children suffering from OI.

Subjects and Methods:

Research Design

A descriptive design was utilized for this study.

Research Setting:

This study was conducted at Genetic Department in Children Hospital affiliated to Ain Shams University Hospitals.

Research Subjects:

A purposive sample of mothers accompanying their children suffering from OI who was attended to the previously mentioned setting. The sample size was determined statistically by power analysis

$$\frac{N \times P(1-p)}{[N-1 \times (d^2/Z^2)] + p(1-p)}$$

Considering the total number of children during the year 2016-2017. Accordingly, the sample size was 80 mothers accompanying their children. Inclusion criteria of children were:

- Both gender regardless level of education
- The mainly diagnosis is OI
- Children aged 5:15 years.

Tools of data collection:

- Data were collected by using the following tools:

1- An interviewing questionnaire tool:

It was developed in a simple Arabic language by the researcher after reviewing the related literatures and reviewed by supervisors; it was consisted of three parts:

Part 1: This part deal with the following:

- Characteristics of the studied children which included age, gender, ranking, and educational level.
- Characteristics of the studied mothers, which included age, educational level, occupation, residence, marital status.
- Characteristics of the studied family which included type of family, number of children, adequacy of family income, presence of family history of OI.

- **Part 2:** This part concerned with mothers' knowledge regarding to OI which included:

- Meaning, types, causes, clinical manifestations, types of conventional treatment of OI, medication used for treatment of OI its precautions and its side effects. Also, the tool included mothers' knowledge about the compliance with medical treatment and knowledge about preventive measures for the expected complications for OI.

❖ **Scoring system:**

Mothers' answers were checked with the model key answer and scored one degree for the "correct answer" and zero for "incorrect answer".

Scores of questionnaire were summed up and accordingly total mothers, knowledge were divided into:

- Satisfactory knowledge, if score ≥ 60%.
- Unsatisfactory knowledge, if score < 60%.

2- Mothers reported practices assessment sheet:

Reported practices of mothers regarding to care of their children suffering from OI which included measures to prevent injury and avoid fracture during clothing. Also, mothers' reported practices related to their role in dental care of their children, first aid of fracture, management of pain result from fracture and care of cast.

❖ **Scoring system:**

Mothers' reported practices were checked and scored as one degree to the step which was done correctly" and zero to the step which was not done or done incorrectly. The total score of mothers reported practices was 43 point. All scores were summed up and accordingly, total mothers' reported practices were classified into:

- Adequate level of mothers reported practices, if score ≥ 60%.
- In adequate level of mothers reported practices, if score<60%.

Validity & Reliability

Tools were validated by five experts in the pediatric medical and nursing fields and the final form of tools was obtained.

Alpha cronbach reliability analysis of the used tool

Items	No. of cases	No. of variables	Alpha Cronbach
knowledge	8	36	0.826
Practices	8	41	0.790

Tools reliability:

The reliability was scaled as follows: <0-0.25 weak reliability, 0.25-0.75 moderate reliability, 0.75-<1strong reliability and 1 is optimum.

Preparatory Phase:

The researcher reviewed the past, current local and international related literature covering various aspects of the problem using books, articles, periodicals and magazines to get acquainted with the research to develop the study tools.

Pilot Study:

A pilot study was carried out on 10% (8 mothers and their children) of the studied sample at the previously mentioned setting to test the clarity and applicability of the study in terms of its setting, tools, time needed based on the results of the pilot study, the necessary modifications were done. Where certain items were added as time of first fracture occurrence, more fractured bone, and times of fractures through last 6 month and previous history of orthopedic surgery. The pilot sample was excluded from study sample.

Operational design:

Field Work:

The actual field work was carried out over a period of 14 months from beginning of January 2019 up to the end of February 2020. The researcher attended the study settings according to attendance of the study subjects in the previously mention setting. .The researcher first introduced herself to the studied mothers and their children in the previously mentioned setting and explained

the aim of the study. The researcher used the constructed tools for collecting the data about mothers' knowledge and their reported practices regarding to care of their children suffering from OI. Each mother was interviewed individually in the waiting area at genetic department that children can receive their treatment (bisphosphonate). The study tools were filled within 30-45 minutes.

Ethical Consideration:

The ethical considerations in the study included the following:

All the gathered data was used for research purpose only. The study sample was informed about the purpose and expected outcomes of the study and they was assured that the study is harmless and their participation is voluntary and they have the right to withdraw from the study at any time and without given any reason. They were assured also that anonymity and confidentiality were guaranteed.

Administrative Design:

Written approval was obtained through an issued letter from Dean of Faculty of Nursing, Ain Shams University to the Hospital director of the previously mentioned setting and the researcher was explained the aim of the study and its expected outcomes.

Statistical Design:

After data were collected, data were coded and transferred into specially designed formats (Excel program) to be suitable for computer feeding. Frequency analysis and manual revision were used to detect possible error. The data was statistically analyzed using the Statistical Package for the Social Science (SPSS) version 10.0. Means and standard deviations were determined for quantitative data and frequency determined for categorical variables.

Results:

Table (1): shows that, the mean age of studied mothers was 32.82±5.3 years and less than half (47.5%) of them had secondary level of education. Meanwhile, 88.7% of mothers were not working and more than half (57.5%) of them were living in urban areas.

Table (2): shows that, the mean age of the studied children was 8.14±5.71years and half(50%) of them were boys . Regarding to their birth order, more than half (52.5%) of them were ranking as first child and less than half (45%) of them were not yet enrolled.

Figure (1): reveals that, majority (91.3%) of studied mothers had negative family history of OI.

Table (3): As observed from that, the majority (96.3%) of studied mothers had incorrect knowledge about meaning of OI. Additionally, the cause of OI was incorrect among less than half (46.3%) of the studied mothers. Moreover, less than two thirds (62.5%) of them had correct knowledge regarding preventive measures for complications of OI.

Figure (2): shows that less than half (42.5%) of the studied mothers had satisfactory knowledge regarding to care of their children suffering from OI

Table (4): according to the studied mothers' reported practice regarding care of cast of broken bones of their children. this table illustrates that, (56.3% & 71.3%) of them keep the cast clean dry not enter the water or waste and If the cast covers the foot or leg, avoid walking and placing the weight on the affected leg unless the doctor allows.

Figure (3): clarifies that, 48.8% of the studied mothers had adequate reported practices regarding to care of their children suffering from OI.

Table (1): Distribution of the studied mothers according to their characteristics.

Items	Total number (80)	
	N.	%
Age (years)		
20: < 30	39	48.75
30 : < 40	33	41.25
40 years and more	8	10
Mean±SD	32.82±5.3	
Educational level		
Illiterate	9	11.3
Read & write	6	7.5
Elementary education	5	6.2
Secondary education	38	47.5
Highly educated	22	27.5
Marital status		
Married	73	91.3
Divorced	6	7.5
Widowed	1	1.2
Occupation		
Working	9	11.3
Not working	71	88.7
Residence		
Rural	34	42.5
Urban	46	57.5

Table (2): Distribution of the studied children according to their characteristics.

Items	Total number (80)	
	N.	%
Children' age (years)		
< 3	19	23.7
3 : < 6	25	31.3
6: < 9	16	20
9 : < 12	12	15
12 : ≤ 15	8	10
Mean±SD	8.14±5.71	
Gender		
Boys	40	50
Girls	40	50
Birth order		
First	42	52.5
Second	20	25
Third and more	18	22.5
Educational level		
Not yet enrolled	36	45
Educational evader	10	12.5
Nursery school	11	13.7
Elementary school	21	26.3
Secondary school	2	2.5

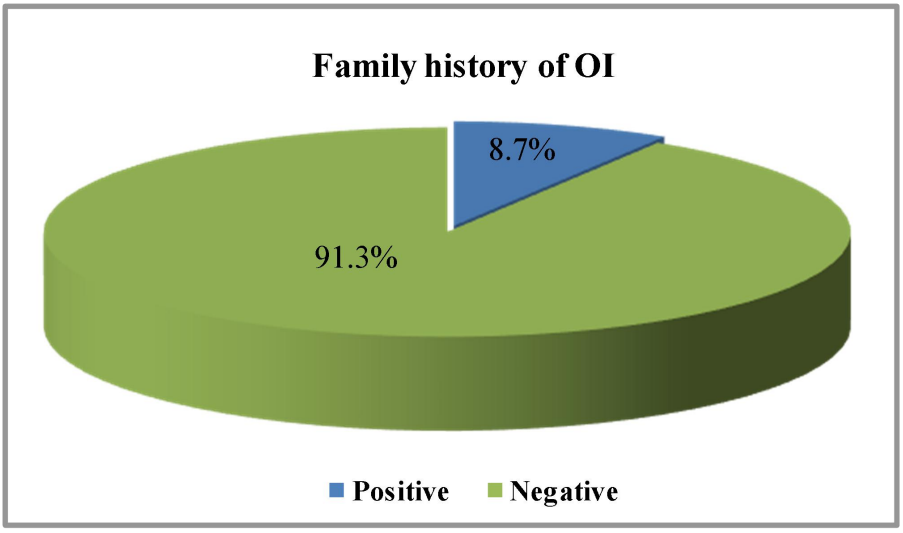

Figure (1): Distribution of the studied mothers according to their family history of osteogenesis imperfecta.

Table (3): Distribution of the studied mothers according to their knowledge regarding the meaning, causes, types, signs & symptoms and preventive measures for complications of OI.

Items	Correct		Incorrect	
	N.	%	N.	%
Meaning of osteogenesis imperfecta	3	3.8	77	96.3
Causes of osteogenesis imperfecta	43	53.8	37	46.3
Types of osteogenesis imperfecta	10	12.5	70	87.5
Sign and symptoms of osteogenesis imperfecta	35	43.8	45	56.3
Preventive measures for the complications of OI	50	62.5	30	37.5

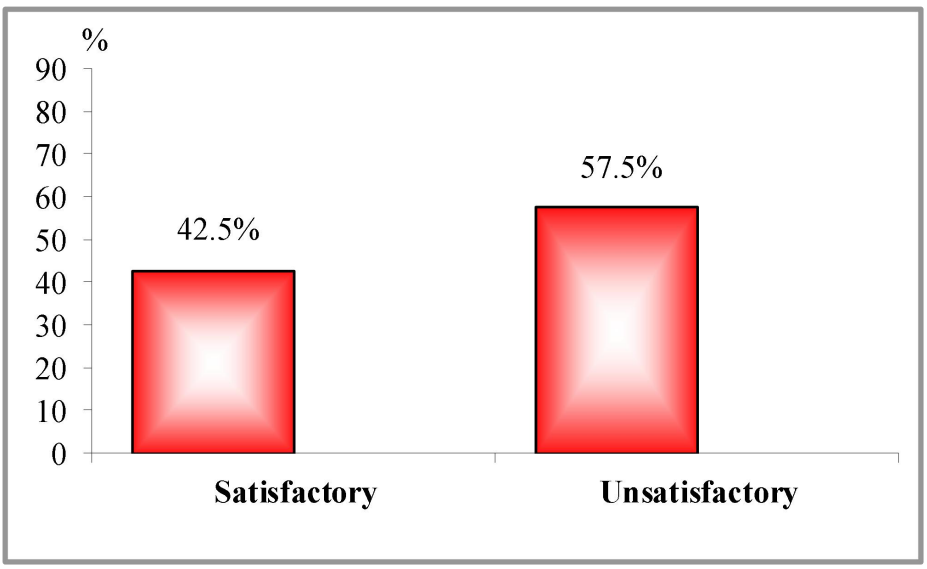

Figure (2): Distribution of the studied mothers according to their total knowledge regarding OI.

Table (4): Distribution of the studied mothers according to their reported practice regarding care of cast of broken bones of their children.

Items	Done		Not done		Chi-square	
	N	%	N	%	X^2	P-value
Keep the cast clean and dry not enter the water or waste	45	56.25	35	43.75	2.500	0.114
Observe signs of swelling, blue disclouration and numbness of the limbs	42	52.5	38	47.5	0.400	0.527
To prevent the swelling, the limb should be lifted on a pillow that is higher than the heart level	34	42.5	46	57.5	3.600	0.058
Do not lean or press the cast, it may lead to breaking it	43	53.75	37	46.25	0.900	0.343
Do not put anything inside the cast, do not try to rub the skin under cast by sharp tool and do not put lushin or powder inside cast.	48	60	32	40	6.400	0.011*
Do not trim the cast or cut sharp edges of it	58	72.5	22	27.5	32.400	<0.001**
If the cast covers the foot or leg, avoid walking and placing the weight on the affected leg unless the doctor allows.	57	71.25	23	28.75	28.900	<0.001**
Use armrest to support cast if it covers the hand, wrist, arm or elbow.	39	48.75	41	51.25	0.100	0.752
Use crutches to walk if the cast on the foot or ankle or leg.	33	41.25	47	58.75	4.900	0.027*

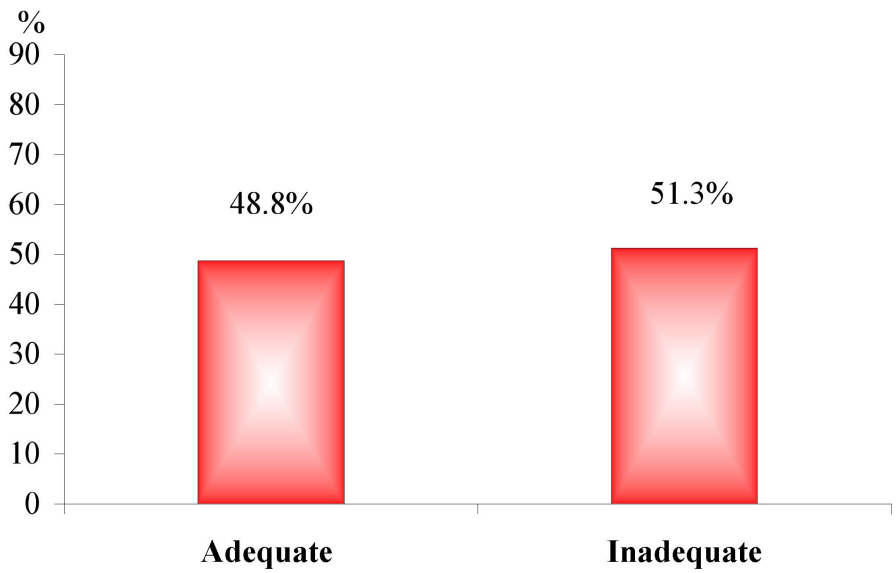

Figure (3): Distribution of the studied mothers according to their total reported practices regarding to care of their children suffering from OI.

Discussion

Osteogenesis Imperfecta (OI) is a rare genetic condition characterized by increased bone fragility leading to frequent fractures and can also cause teeth and soft tissue abnormalities in addition to discolored sclera, hearing loss, shortened stature and skeletal deformities. Family caregivers, such as parents, play a critical role in helping children having OI with unique needs thrive at home and in the community. Understanding the day-to-day experiences of caregivers of children with rare conditions, including caregivers of children with OI, is imperative to inform the development of policies and services tailored to their needs **(Etich et al., 2020).**

As regards to the studied mothers' characteristics, the finding of the study results showed that, less than half of the studied mothers, their age ranged between 30 to less than 40 years and have secondary education. Also, the results showed that, more than half of them were living in urban areas. These results were disagree with **Jovanovic, Guterman-Ram & Marini, (2021)** who carried out a study entitled "Osteogenesis Imperfecta: Mechanisms and signaling pathways connecting classical and rare OI types. Endocrine Reviews" and mentioned that, more than one third of the studied mothers were in the age group 31 - 40 years, and more than half of them were highly educated and were from rural area.

Regarding characteristics of the studied children, the results of the present study illustrated that, about one third of the studied children were in the age group 3 to less than 6years and more than half of them were ranked as first child. These results were disagree with results of **Castro et al., (2020)** who studied the day to day experiences of caring for children with OI: A qualitative descriptive study in Canada and found that less than half of the studied children were in the age group6–12 years old and about half of them were ranked as second child.

The results of the current study revealed that, half of the studied children were boys and less than half of them weren't yet enrolled in education. These results were disagreed with **Vanz et al., (2018)** who studied, health-related quality of life for children and adolescents with osteogenesis imperfecta: A cross-sectional study using Peds QL in Brazil and mentioned that, slightly more than half of the studied children were male and less than half of them weren't yet enrolled in education.

In relation to the family history of OI of the studied children, the results of present study showed that, the majority of the studied children had negative family history of OI. This result was agreed with result of **Pepin & Byers, (2015)** who carried out a study entitled "What every clinical geneticist should know about testing for osteogenesis imperfecta in suspected child abuse cases. In America" and mentioned that, approximately 85% of children with Osteogenesis Imperfecta are born into a family with no family history of Osteogenesis Imperfecta. Most often this is due to a new mutation to a gene and not by anything the parents did before or during pregnancy.

The present result similar with **Wiggins& Kreikemeier, (2017)** who conducted a study entitled "Bisphosphonate therapy and osteogenesis imperfecta: the lived experience of children and their mothers in USA" and mentioned that, the majority of the studied children had negative family history of OI.

As regards studies mothers' knowledge regarding the meaning and causes of OI, the results of present study showed that, the minority of the studied mothers had a correct knowledge regarding meaning of OI, a cause of OI was answered correctly by less than half of mothers'. These results were congruent

with **Dogba et al., (2016)** who carried out a study entitled "Involving families with OI in Health Service Research: Joint Development of the OI/ECE Questionnaire in Canada" and mentioned that, most families had a generally poor knowledge regarding definition and causes of OI.

Also, the results showed that less than half of the studied mothers had a correct knowledge regarding sign and symptoms of OI. Additionally, less than two thirds of the studied mothers had a correct knowledge regarding preventive measures for the complications of OI. This result agreed with **Thomas & DiMeglio, (2016)** who carried out a study entitled "Advances in the classification and treatment of OI. Current osteoporosis reports" and mentioned that about half of the studied mothers had a correct knowledge regarding sign and symptoms of OI and preventive measures for the complications of OI.

Concerning to the total knowledge of the studied mothers regarding care of their children suffering from OI, the result of present study showed that, less than half of the studied mothers had satisfactory knowledge regarding OI. From the researcher point of view, unsatisfactory level of the studied mothers' knowledge might be due to inadequate information given by healthcare team and the nature of the disease which is considered a rare disease. This result was congruent with **Lim et al., (2017)** who carried out a study entitled "Genetic causes and mechanisms of osteogenesis imperfecta" and stated that, most of the studied mothers had satisfactory knowledge regarding OI.

As regard the studied mothers reported practices regarding care of cast for their children, the present study showed that more than one half of the studied mother keep the cast clean, dry not enter the water or waste. This result was disagreed with ***Hill et al.,(2019)*** who conduct a study entitled "Exploring the impact of Osteogenesis Imperfecta on families in UK" and mentioned that, three quarters of the studied mothers keep the cast clean, dry not enter the water or waste.

As regard of the studied mothers' total reported practices regarding to care of their children suffering from OI, the present study showed that, less than half of the studied mothers had adequate reported practices regarding to care of their children suffering from OI. From the researcher point of view, this result might be due to lack of their knowledge about the child disease and fear from dealing with their child due to their fragile bone and increase occurrence of fractures. This result was in the same line with **Castro et al., (2020)** who mentioned that less than half of the studied mothers had adequate reported practices regarding to care of their children suffering from OI.

Conclusion

Based on the finding of this study it can be concluded that less than half of the studied mothers had satisfactory knowledge about OI and less than half of them had adequate reported practices regarding to care of their children suffering from OI.

Recommendations

In the light of the findings of the current study the following recommendations are suggested:

- Educational program intervention should be conducted for mothers of children with OI to raise their knowledge and practices regarding dealing with their children suffering from OI.
- Availability and distribution of pamphlets and booklet containing the basic knowledge and practices for mothers about the disease of their children.
- Further study on large sample and in various places in Egypt to generalization of results.
- Further researches should be conducted to determine the barriers/ challenges that

affect mothers' knowledge and practices in caring of their children with OI.

References:

Bishop, N. (2018): Osteogenesis Imperfecta: Pathophysiology and Treatment in Childhood. Rheumatology, 57(suppl_3), key075-149.

Castro, R., Marinello, J., Chougui, K., Morand, M., Bilodeau, C., & Tsimicalis, A. (2020): The Day to Day Experiences of Caring for Children with Osteogenesis Imperfecta: A qualitative Descriptive Study. Journal of Clinical Nursing, 29(15-16), 2999-3011.

Dogba, M., Dahan-Oliel, N., Snider, L., Glorieux, F., Durigova, M., Palomo, T., & Rauch, F. (2016): Involving Families with Osteogenesis Imperfecta in Health Service Research: Joint Development of the OI/ECE Questionnaire. PloS one, 11(1), e0147654.

Etich, J., Leßmeier, L., Rehberg, M., Sill, H., Zaucke, F., Netzer, C., & Semler, O. (2020): Osteogenesis Imperfect Pathophysiology and Therapeutic Options. Molecular and Cellular Pediatrics, 7(1), 1-9.

Hill, M., Lewis, C., Riddington, C., Crowe, B., DeVile, C, & Chitty, L. (2019): Exploring the Impact of Osteogenesis Imperfecta on Families: A mixed-Methods Systematic Review. Disability and Health Journal, 12(3), 340-349.

Jovanovic, M., Guterman-Ram, G., & Marini, J. (2021): Osteogenesis Imperfecta: Mechanisms and Signaling Pathways Connecting Classical and Rare OI Types. Endocrine Reviews.

Lim, J., Grafe, I., Alexander, S., & Lee, B. (2017): Genetic Causes and Mechanisms of Osteogenesis Imperfecta. Bone, 102, 40-49.

McDowell, R., McKendry, A., Smyth, G., & Cardwell, P. (2019): Reflection on the Assessment and Care of A child with Osteogenesis Imperfecta. Nursing Children and Young People, 31(2),35-40.

Pepin, G., & Byers, P.H. (2015): What Every Clinical Geneticist Should Know about Testing for Osteogenesis Imperfecta in Suspected Child Abuse Cases. In American Journal of Medical Genetics Part C: Seminars in Medical Genetics (Vol. 169, No. 4, pp. 307-313).

Santos, D., Pires, F., Soares, K., & Barros, L. (2018): Family Experience with Osteogenesis Imperfecta Type 1: The Most Distressing Situations. Disability and Rehabilitation, 40(19), 2281-2287.

Statistical Office of Genetic Department of Children Hospital of Ain Shams University, 2018.

Thomas, I., & DiMeglio, L. (2016): Advances in the Classification and Treatment of Osteogenesis Imperfecta. Current Osteoporosis Reports, 14(1), 1-9.

Vanz, A., Lee, S., Pinheiro, B., Zambrano, M., Brizola, E., & Rocha, D. (2018): Health-Related Quality of Life of Children and Adolescents with Osteogenesis Imperfecta: A cross-Sectional Study using PedsQL. BMC Pediatrics 3:6.

Wiggins, S., & Kreikemeier, R. (2017): Bisphosphonate Therapy and Osteogenesis Imperfecta: The lived Experience of Children and Their Mothers. Journal for Specialists in Pediatric Nursing, 22(4), e12192.

Zhytnik, L., Simm, K., Salumets, A., Peters, M., Märtson, A., & Maasalu, K. (2020): Reproductive Options for Families at Risk of Osteogenesis Imperfecta: A review. Orphanet Journal of Rare Diseases, 15, 1-20.