Global Academic Nursing Journal

EXPERIENCE REPORT

Association between fractures and recurrent falls, symptoms and health orientation: glass bones

Asociación entre fracturas y caídas recurrentes, síntomas y orientación de salud: huesos de vidrio

Associação entre fraturas e quedas recorrentes, sintomatologia e orientação em saúde: ossos de vidro

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How to cite this article:
Teixeira AR, Magalhães BVB, Brito BER, Ferreira DFA, Fortes GS, Domingos IAS, Barbosa MMM, Lopes MO, Rosendo SS, Kataoka FT. Association between fractures and recurrent falls, symptoms and health orientation: glass bones. Glob Acad Nurs. 2022;3(Spe.2):e272. https://dx.doi.org/10.5935/2675-5602.20200272

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Submission: 04-22-2022
Approval: 06-30-2022

Abstract

Osteogenesis imperfecta is a rare hereditary disease characterized by a deficiency in the production of type I collagen that interferes with connective tissue, culminating in recurrent fractures with minimal trauma due to bone fragility. In most cases, the disease results from mutations in the COL1A1 and COL1A2 genes, responsible for encoding the type I collagen chain. It is a descriptive study, with a qualitative approach, of the experience report type, with activities developed during the class practice of the subject of Semiology II, in a hospital in the city of Caratinga-MG, portraying the medical care provided to a patient with osteogenesis imperfecta. Thus, the following steps were taken: data collection during anamnesis, physical examination and consultation of medical records. It refers to a male patient, 65 years old, farmer, diagnosed with the disease, hospitalized for a fracture of the right lower limb in the portion of the diaphysis of the femur, totally displaced. In short, the present study aims to contribute to the dissemination of knowledge about osteogenesis imperfecta, given the scarcity of publications and scientific studies on this disease, exploring the clinical and symptomatological history of a patient with osteogenesis imperfecta, highlighting the necessary conducts and guidelines for a good prognosis.

Descriptors: Osteogenesis Imperfecta; Fractures, Bone; Collagen Type I; Genetics; Bone Tissue.

Resumén

La osteogénesis imperfecta es una rara enfermedad hereditaria caracterizada por una deficiencia en la producción de colágeno tipo I que interfiere con el tejido conectivo, culminando en fracturas recurrentes con mínimo trauma por fragilidad ósea. En la mayoría de los casos, la enfermedad resulta de mutaciones en los genes COL1A1 y COL1A2, responsables de codificar la cadena de colágeno tipo I. Se trata de un estudio descriptivo, con abordaje cualitativo, del tipo relato de experiencia, con actividades desarrolladas durante la práctica de clase de la asignatura de Semiología II, en un hospital de la ciudad de Caratinga-MG, retratando la atención médica brindada a un paciente con osteogénesis imperfecta. Así, se siguieron los siguientes pasos: recolección de datos durante la anamnesis, examen físico y consulta de las historias clínicas. Se trata de un paciente masculino, de 65 años, campesino, diagnosticado de la enfermedad, hospitalizado por fractura del miembro inferior derecho en la porción de la diáfisis del fémur, totalmente desplazada. En definitiva, el presente estudio pretende contribuir a la difusión del conocimiento sobre la osteogénesis imperfecta, dada la escasez de publicaciones y estudios científicos sobre esta enfermedad, explorando la historia clínica y sintomatológica de un paciente con osteogénesis imperfecta, destacando las conductas y pautas necesarias para un buen pronóstico.

Descriptores: Osteogénesis Imperfecta; Fracturas Óseas; Colágeno Tipo I; Genética; Tejido Óseo.

Resumo

A osteogênese imperfeita é uma doença hereditária, rara, caracterizada pela deficiência na produção de colágeno tipo I que interfere no tecido conjuntivo, culminando em fraturas recorrentes a mínimos traumas devido à fragilidade óssea. Na maioria dos casos, a doença resulta de mutações nos genes COL1A1 e COL1A2, responsáveis por codificar a cadeia de colágeno tipo I. Trata-se de um estudo descritivo, de abordagem qualitativa, do tipo relato de experiência, com atividades desenvolvidas durante a aula prática da matéria de Semiologia II, em um hospital da cidade de Caratinga-MG, retratando o atendimento médico prestado a um paciente com osteogênese imperfeita. Atendeu-se assim às etapas: coleta de dados durante anamnese, exame físico e consulta ao prontuário. Refere-se a um paciente sexo masculino, 65 anos, lavrador, com diagnóstico da doença, internado por fratura de membro inferior direito na porção da diáfise do fêmur, totalmente desviada. Em suma, o presente estudo tem como objetivo contribuir na divulgação de conhecimento sobre a osteogênese imperfeita, frente à escassez de publicações e estudos científicos perante tal enfermidade, explorando a história clínica e sintomatológica de um paciente com osteogênese imperfeita, salientando as condutas e orientações necessárias para um bom prognóstico.

Descritores: Osteogênese Imperfeita; Fraturas Ósseas; Colágeno Tipo I; Genética; Tecido Ósseo.

Introduction

Osteogenesis imperfecta (OI), also known as glass bone disease or crystal bone disease, is characterized by a genetic and hereditary disorder that affects the connective tissue matrix, compromising the formation of type I collagen. bone tissue, affecting its structure and causing, ordinarily, the fragility of this tissue, making it susceptible to deformities and repeated fractures[1].

Glass bone disease has been classified into several types (I to VIII) according to the clinical characteristics and the responsible genes, since it presents genotypic and phenotypic heterogeneity. In general, the symptoms of this disorder are associated with osteopenia, dental changes, blue sclera and joint hypermobility generated by the decrease in ligament thickness[1,2].

The OI arises mainly from autosomal dominant genetic alterations. In addition, in the world population it has an estimated incidence of 1:5000/10000, while in the Brazilian population, according to data from the Brazilian Association of Osteogenesis Imperfecta (ABOI), there are approximately 12,000 people[2-4]. There is a significant clinical improvement of patients with OI when adopting the appropriate treatment. However, osteogenesis imperfecta has no cure, its therapeutic approaches are palliative and aim to improve the quality of life of patients, seeking to help them to live in society without great harm[1].

It is observed that OI patients experience a series of obstacles related to their pathology, since it not only causes physical consequences, but also emotional, psychological and social repercussions. In addition, there is a scarcity of publications and scientific studies focused on this disease. Thus, in view of the need to contribute to the dissemination of knowledge about osteogenesis imperfecta among the scientific community, this article aims to describe the clinical history and symptoms of a patient with osteogenesis imperfecta, as well as to highlight the conducts and guidelines necessary for a good prognosis.

Methodology

The present work is a descriptive study, with a qualitative approach, of the experience report type, developed in a hospital in the city of Caratinga, Minas Gerais, Brazil. The activities described were developed in March 2022, during the practical class of the Semiology II subject, of the Bachelor of Medicine course of a Higher Education Institution. The report portrays the medical care provided to a patient with a diagnosis of osteogenesis imperfecta who, for the logistics of care, followed the steps: data collection during anamnesis, physical examination and consultation of medical records. Regarding ethical procedures, the study was limited to discussing the academic experience in the medical consultation of patients with osteogenesis imperfecta, therefore, it did not directly involve the patient, with no need for consideration by the Research Ethics Committee.

Experience Report

During March 29, 2022, at the surgical clinic of a hospital in Caratinga-MG, a medical consultation and semiological evaluation of a patient took place, conducted by students of the fifth period of the Medicine course of a Higher Education Institution. N.G.R., 65 years old, married, farmer, low income, 1.30 m tall and with Osteogenesis Imperfecta, had been hospitalized since March 4, 2022 due to a fall that resulted in a fracture of the right lower limb in the of the femoral diaphysis, which is totally deviated.

During the consultation, it was performed in different periods: anamnesis, physical examination, analysis of vital signs and imaging exams. Regarding the history of the current disease, stage of the anamnesis, the patient inferred that during childhood, he was diagnosed with Dentinogenesis Imperfecta and, in addition, in adolescence, at age 16, his first fracture related to Osteogenesis Imperfecta occurred, located in the upper limb.

Later, N.G.R. he complained of successive fractures in the last 5 years and that, as a result, he found himself in need of frequent medical support. In particular, he highlighted 4 other previous fractures, two of which were also located in the femur, one in the knee and another in the arm.

Furthermore, it is extremely important to emphasize that in the analysis of the patient's family history, the heredity of the disease was identified, since relatives, such as nephews, nieces, four brothers, father and daughter, are also carriers of Osteogenesis Imperfecta. Given these facts, it was emphasized that the daughter presented the disease earlier, since she had three intrauterine fractures.

Regarding the physical examination, in the inspection, short stature, hyperkyphosis, claw hand, short lower and upper limbs and "staff" femur were noted. Furthermore, in the eyes, blue sclera and in the abdomen a brownish hyperchromic stain was observed in the path of the dermatome of the upper limbs. On cardiac and respiratory auscultation, N.G.R. had rhythmic heart sounds with S1 hypophonesis and pulmonary crackles and, in the gastrointestinal tract, the hydro-air sounds were preserved.

Finally, the x-ray analysis confirmed the presence of a fracture in the right lower limb in the femoral diaphysis portion, in addition to the imperfect healing resulting from fractures that had already occurred.

Discussion

Symptomatology

As already mentioned, OI is a hereditary genetic disorder that affects the formation of type 1 collagen. This type of collagen is the most abundant and can be found in bones, ligaments, skin and tendons, in order to generate greater resistance to the fabrics. Thus, in view of the structural changes caused by OI, type 1 collagen is not able to interact with hydroxyapatite, which is one of the fundamental elements in the structural formation of bones, and the combination of these elements is essential for bone strength.[5,6]. Therefore, the genetic alterations of OI cause a variability of clinical manifestations that require greater attention from the health professional for an effective diagnosis and treatment.

The striking features of OI are: bone and physical deformity, repeated fractures, osteopenia and osteoporosis,

frailty, short stature, bluish sclera, deafness, dentinogenesis imperfecta (DI), elastic skin, chronic pain, joint hypermobility and wormian bones in the sutures of the skull in some specific cases[6].

During the semiological evaluation of the patient, some typical characteristics of OI are noticeable, such as his short stature of 1 meter and 30 centimeters, which is a value well below the Brazilian average. In addition, relevant bone deformations were identified, such as hyperkyphosis, "staff" femur and short lower and upper limbs, which is intrinsically related to congenital bone malformation.

Another striking manifestation in the patient is his history of repeated fractures, and the reason for hospitalization was a fall that culminated in a fracture of the right lower limb in the diaphysis portion of the femur. Furthermore, the patient reported that the first fracture he remembers was when he was 16 years old, when he fractured one of his upper limbs. In addition, also in the last 5 years he highlighted 4 other fractures, which made him dependent on medical support and affected one of the knee joints. This situation is related to bone fragility in the OI, living up to the popular name "Glass Bones", which reflects a bone formation without resistance and brittleness, similar to the characteristics of glass.

Another symptom reported by the patient during his childhood was dentinogenesis imperfecta, whose condition may or may not be related to OI. Type 1 ID, the most common in these cases, is a genetic alteration that affects the formation of dentin and is linked to OI. In this condition, the teeth become opalescent, with shades varying between yellowish and brown, in which the dentin is exposed and with a softened appearance[7].

Thus, it was observed that the patient had bluish sclerotitis, caused by the thinning of type 1 collagen in the most superficial layer of the eyeball. Furthermore, in the inspection of the abdomen, a brownish hyperchromic stain was observed in the path of the upper limbs dermatome, which, after further medical investigation, led to the diagnosis of herpes zoster already in the healing phase. Regarding deafness, during the medical investigation there was no hearing impairment of the patient N.G.R.

Finally, it is important to highlight the patient's family history in relation to the heredity of osteogenesis imperfecta which, according to the literature[8], in most cases, it is characterized by mutations in the COL1A1 and COL1A2 genes. Therefore, it was reported by the patient that his daughter had the disease early and had 3 intrauterine fractures. Such a situation is common in OI, especially in more severe cases of the disease such as type 2 OI, characterized by great fragility of the bones. It is important to note that N.G.R has type 3 OI, characterized by short stature and varied fractures with the presence of deformities. In addition, other relatives of N.G.R, such as his father and 4 more brothers, had the disease.

Diagnosis and treatment

Early diagnosis is essential in these cases and can be clinical, identifying the common signs and symptoms presented and relating them to each type of OI, laboratory, through bone density and imaging tests and also through family history. In this report, the individual presented had the initial diagnosis in childhood, through the clinic, when he presented dentinogenesis imperfecta and his first fracture at 16 years old[9].

The treatment of OI goes far beyond preventing fractures, it also has the objective of promoting growth, mobility, functional independence and pain reduction, that is, ensuring an improvement in quality of life. In addition, this must occur in a multidisciplinary way, as the pathology has several clinical repercussions, requiring follow-up with a physical therapist, dentist, nutritionist and surgical interventions when necessary[10]. Thus, the sooner the disease is diagnosed and treatment is started, the better the results will be. However, this was not the patient's reality, as he had his first fracture at age 16 and many others throughout his life.

Thus, the treatment described in the literature has flaws in practice, mainly when analyzing the difficulty of diagnosing OI, whether because it is a rare disease, lack of knowledge of professionals in action, lack of adherence to treatment by the patient, which needs to work and does not attend follow-ups correctly, resulting in multiple lifetime fractures and worsening clinical status.

There are several pharmacological therapies available, such as growth hormone therapy, antiresorptive treatments and monoclonal antibody treatments that aim to reduce symptoms and treat the disease at the cellular and genetic level[11]. However, most of these drugs are still in animal testing phases or have pediatric use, not serving the elderly, as is the case. Another point is that they are expensive and unfeasible therapies at the level of the Unified Health System (SUS), as it is a rare disease; SUS prefers to invest in cheaper medicines that serve a greater number of users, which distances even more patients, as reported, from these alternatives.

Health guidance

As it is a disease, whose evolution disables the carrier, health guidance is a way to alleviate the symptoms. In addition, the orientation aims to prevent the patient from becoming prostrate, depressed and anxious with their physical condition. A proof of this is that, in the mid-2000s, it was believed that the person with OI should make as little effort as possible to maintain bone integrity and avoid injuries, currently, it is known the physical and psychological importance for these individuals to move, work and have their leisure[6]. Therefore, it is important to correctly guide the patient on the need to carry out a multidisciplinary follow-up (psychologist, nutritionist, dentist and physiotherapist) to improve the quality of life.

It is also necessary for the individual to understand his pathology so that he does not exaggerate in day-to-day activities, such as carrying a lot of weight, jumping from high places and performing dangerous activities, since he has bone fragility and exaggeration can lead to serious fractures. , with difficult and extensive treatments, that is, the patient does need to move, but not in a mistaken and exaggerated way[6,12]. In the case reported, the patient is a farmer, a heavy

task and not suitable for his condition, however, it is his only form of livelihood, it would be an alternative to inform about the option of retirement or even to warn that this work is carried out in a least harmful possible.

Finally, when the diagnosis of OI is confirmed, genetic screening is guided in order to diagnose close relatives who may be carriers of the disease-causing gene and, consequently, may in the future have children with OI, in addition to identifying if the offspring of the patient has the pathology[13]. In this case, screening was extremely important, identifying several carriers in the family and a daughter of the patient with OI, who could start early treatment, since she had an early diagnosis.

Final Considerations

In view of the exposed by the experience report added to the available literature, there is a need for comprehensive care for patients with Osteogenesis Imperfecta, given the various clinical manifestations that reflect on physical and psychological states. Therefore, it is necessary a multidisciplinary follow-up with doctors, dentists, nutritionists, physiotherapists and psychologists, which aims to alleviate the consequences caused by different symptoms and offer better expectations regarding mobility and autonomy.

As already mentioned, health guidance, both with regard to professionals and the patient himself, is of paramount importance for improving the quality of life, since the scarcity of knowledge and the absence of professional follow-up have consequences in terms of regarding symptom improvement, functional independence, performing daily activities and pain reduction.

In summary, patients with OI treated correctly and with the proper information are more likely to have positive prognoses, less likely to have repeated fractures due to inappropriate activities, better lifestyle habits, greater adherence to treatments, decent work and leisure, constituting an integral action to compose the health of individuals.

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