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CASE REPORT: OSTEOGENESIS IMPERFECTA IN DAUGHTER PATIENTS AGED 6 YEARS 9 MONTHS

Kirnia Tri Wulandari, Tikto
General practitioner at Gantrung Field Hospital, Pediatrician at Dolopo Hospital, Madiun Regency, East Java
E-mail: kirnia.wulandari@gmail.com, puskgantrung@gmail.com

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ABSTRACT

Osteogenesis imperfecta (OI) is a rare disease with a wide spectrum of clinical and genetic variability; It is characterized by very brittle bones, blue sclera, dentinogenesis imperfecta, scoliosis and hearing loss. This study aims to identify and report cases of osteogenesis imperfecta in child patients aged 6 years 9 months. This study uses a qualitative method with the type of case report. The sampling technique used in this study is random sampling technique by Slovin formula in Husein Umar. In this study, each population has same opportunity to be selected as a sample. Based on the results of the analysis and discussion, it can be concluded that the case of osteogenesis imperfecta is a complex congenital disorder and must be distinguished from other differential diagnoses. Furthermore, with a careful examination, it is hoped that the diagnosis of osteogenesis imperfecta cases can be better.

KEYWORDS
Osteogenesis imperfecta, Daughter, Aged 6 Years 9 Months

INTRODUCTION

This study aims to identify and report cases of osteogenesis imperfecta in child patients aged 6 years 9 months. Osteogenesis imperfecta (OI) is a rare disease with a wide

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spectrum of clinical and genetic variability; characterized by very brittle bones, blue sclera, dentinogenesis imperfecta, scoliosis and hearing loss (Ginting & Sitanggang, 2015).

Osteogenesis imperfecta (OI) is a genetic disease that causes bone fragility caused by mutations in the gene encoding the type I collagen chain. Collagen is the most abundant protein in bones, teeth, sclera and ligaments. The known incidence in under-fives is about 1 in 20,000 births. It is estimated that 25,000 to 50,000 people suffer from OI in the United States (Hasanah, 2014).

The majority of OIs occur due to mutations in genes responsible for the production of intracellular type I procollagen, which plays an important role in the formation of tissues such as bone, tooth enamel, eye sclera, skin, tendons, and ligaments (Suadiatmika, 2018). However, the actual incidence is estimated to be higher considering that there are pediatric patients who are not diagnosed because they have mild signs. OI occurs in all racial and ethnic groups.

Osteogenesis imperfecta, also known as 'brittle bone disease', is primarily caused by mutations in the genes COL1A1 and COL1A2.3,4 These genes provide the instructions for making type I collagen, which is the most abundant protein in bone, skin, and connective tissue. others to ensure the structure and strength of the body (Suadiatmika, 2018). Changes in the COL1A1 and COL1A2 genes cause a pro-alpha 1 or pro-alpha 2 chain defect, so that type I collagen production is reduced and results in brittle bones. Of the 250 mutations, the two most common types are null mutations and negative dominant mutations (Febriani, 2013).

Although OI is not found in daily practice, this disorder is a common disease for which the provision of appropriate management must be considered (Norlela & Muflihatin, 2015). Dental, oral and craniofacial manifestations are often observed and can be a very important diagnostic tool if physical signs and symptoms are uncertain (Fauziah, 2012). Thus dentists must know the dental abnormalities that occur in patients with OI, because it involves poor aesthetics, causing most sufferers to feel inferior (Muliyawan, 2013).

Osteogenesis imperfecta (OI) is a serious genetic disorder affecting the connective tissue, characterized by easy fracture of the bone, often due to very minor or no visible trauma. Synonyms of this disease are: imperfect osteogenesis, Van der Hoeve syndrome, Eddowe syndrome, Lobstein disease, fragile bone disease, Vrolik disease.

Although fractures can often occur in pediatric patients with OI, the number of fractures can also decrease in adults due to the influence of sex and growth hormones (SENJA, 2018). On the other hand, the reduced amount of hormones present at menopause may exacerbate the clinical manifestations of OI. The prognosis varies from very good (autosomal dominant form) to very poor (autosomal recessive form) because the variation in clinical manifestations is very large (Dewi, 2019).

RESEARCH METHOD

This study uses a qualitative method with the type of case report. The sampling technique used in this study the author uses the Random Sampling technique or by using the Slovin formula in Husein Umar. Where each population has the same opportunity to be selected as a sample in this study. Tthe sampling technique is done by random sampling technique and collected the data using observation, interview and documentation.

RESULT AND DISCUSSION

A. Pathophysiology, Symptoms and Diagnosis of Osteogenesis Imperfecta (OI)

1) Pathophysiology

Osteogenesis imperfecta, also known as 'brittle bone disease', is primarily caused by mutations in the genes COL1A1 and COL1A2.3,4 These genes provide the instructions for making type I collagen, which is the most abundant protein in bone, skin, and connective tissue. others to ensure the structure and strength of the body. Changes in the COL1A1 and COL1A2 genes cause a pro-alpha 1 or pro-alpha 2 chain effect, so that type I collagen production is reduced and results in brittle bones. Of the 250 mutations, the two most common types are null mutations and negative dominant mutations.

2) Symptoms

There are several types of OI, with clinical symptoms ranging from mild to severe, and each patient may have a different combination of symptoms. The bones of all patients with OI are generally more brittle. Common symptoms of OI include: short stature, triangular face, difficulty breathing, hearing loss, weak teeth, deformities, such as arched legs or scoliosis.

Osteogenesis imperfecta can range from mild to severe. Most people with osteogenesis imperfecta suffer from weak bones and hearing loss. Babies with severe osteogenesis imperfecta usually have multiple fractures at birth, and the skull may be so soft that the brain cannot be immune to the pressure of the head at birth.

For simple osteogenesis imperfecta, the bone often breaks after a very minor injury, usually when the child begins to walk. Children with mild osteogenesis imperfecta may experience multiple fractures during childhood and even after adolescence when the bones are stronger. Sometimes, children with osteogenesis imperfecta develop heart or lung disease.

Common health problems in children and adults with OI include: short stature, weak tissues, fragile skin, muscle weakness and loose joints, bleeding, easy bruising, frequent nosebleeds, and a small number of people suffer from severe bleeding from injuries and hearing loss. . Starting in childhood, it affects about 50% of adults, breathing problems, a higher risk of asthma and other lung problems, and curvature of the spine.

In addition to fractures, OI patients often experience muscle weakness, hearing loss, fatigue, joint weakness, crooked bones, scoliosis, blue sclera, dentinal hypoplasia (tooth decay), and short stature. Restrictive lung disease occurs in people who are more severely affected. OI is caused by an error called a genetic mutation that affects the production of collagen found in bones and other tissues in the body. It is not caused by too little calcium or a nutritional deficiency. OI is a variable of the type described in the medical literature. Its severity ranges from fatal to mild forms, with almost no obvious symptoms. A person's specific medical problem will depend on its severity. People with mild OI may experience several fractures, while patients with severe OI may experience hundreds of fractures in their lifetime. The number of Americans affected by OI is estimated at 25,000-50,000. This range is very wide, as mild OI often goes undiagnosed.

3) Diagnosis

The clinical diagnosis of osteogenesis imperfecta is based mainly on the signs and symptoms outlined above. Traditionally, much emphasis has been laid on the presence or absence of blue sclera and dentinogenesis imperfecta as diagnostic signs of osteogenesis imperfecta. This practice still holds true, but some limitations should be recognised. Dark or bluish sclerae are very typical in healthy infants, and therefore this finding is not of much diagnostic use in this age-group. Dentinogenesis imperfecta is

more frequently clinically evident in primary than in permanent teeth of patients with osteogenesis imperfecta. Radiological or histological examinations frequently show abnormalities, even in individuals whose teeth look normal on inspection.

Clinically evident hearing loss is rare in the first two decades of life, even though subtle audiometric abnormalities can be recorded in a large proportion of children and adolescents with osteogenesis imperfecta. About half of patients older than age 50 years report hearing loss, and an even higher proportion of adults have clearly pathological audiometric findings.

Diagnosis of osteogenesis imperfecta is straightforward in individuals with a positive family history or in whom several typical features are present, but can be difficult in the absence of affected family members and when bone fragility is not associated with obvious extraskeletal abnormalities.

The uncertainty in such cases is compounded by the fact that there are no agreed minimum criteria that establish a clinical diagnosis of the disorder. In this situation, analysis of the collagen type 1 genes can provide helpful information, which can be done by investigating the amount and structure of type 1 procollagen molecules that are derived from the patient's cultured skin fibroblasts.

Alternatively, genomic DNA can be extracted from white blood cells and the coding region of the *COL1A1* and *COL1A2* genes can then be screened for mutations. Both of these approaches are thought to detect almost 90% of all collagen type 1 mutations. A positive collagen type 1 study thus confirms the diagnosis of osteogenesis imperfecta. However, a negative result leaves open the possibility that either a collagen type 1 mutation is present but was not detected or the patient has a form of the disorder that is not associated with collagen type 1 mutations (see below). Therefore, a negative collagen type 1 study does not rule out osteogenesis imperfecta.

B. Case and Discussion

The case that we present is a patient, initials F, female, aged 6 years 9 months, came to the pediatric polyclinic for control and asked for a referral to RSUP Soetomo. His medical history, the patient has experienced 32 different fractures since birth, physical examination revealed curved arms and legs, laterally curved spine, blue sclera, carious teeth honey brown cororation, barrel chest, radiological examination showed severe osteoporosis without abnormalities in laboratory findings. There is no history of similar disease in the patient's family.

The patient underwent routine treatment since the age of 1 year 9 months. In the last 2 years the patient has not experienced a fracture. The patient was treated with zoledronic acid, vitamin D and oral calcium.

Osteogenesis Imperfecta is a bone disorder in the form of easily broken bones caused by abnormalities in the formation of bone collagen, this disorder is a genetic disorder caused by mutations in genes. In children with OI, BMD is very low so it is easy to fracture even though the impact is not hard. Cases were established based on anamnesis, physical and radiological examinations. This patient has not made a differential diagnosis because it is considered quite typical for an OI. The administration of bisphosphonates in this case is in accordance with the literature, which is to increase bone mass and reduce the incidence of fractures.

CONCLUSION

Based on the results of the analysis and discussion, it can be concluded that the case of osteogenesis imperfecta is a complex congenital disorder and must be

distinguished from other differential diagnoses. With a careful examination, it is hoped that the diagnosis of osteogenesis imperfecta cases can be better.

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