Osteogenesis Imperfecta Case Report

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Introduction

Osteogenesis imperfecta (OI) is a genetic disorder that affects the production of collagen, a protein thatprovides structure and strength to bones, as well as other connective tissues such as tendons, ligaments, teeth, eyes and cardiovascular tissue. There are several types of OI, with varying degrees of severity. It is caused by mutations in one of several genes that are involved in the production of collagen. Inheritance of the mutated gene can be either autosomal dominant or autosomal recessive, depending on the type of OI. In this case, the patient is a child suffering from multiple fractures who was evaluated for child abuse and underwent genetic testing to finally be diagnosed with OI.

Case Report

A 15 month old boy presented to the ER with abdominal pain, body and neck rigidity. Patient was found to have an incidental findings of multiple bone fractures with different stages of healing. Child protective services were consulted for suspected child abuse.

A skeletal survey multiple old and current fractures. Patient was placed in a full body brace. After multiple follow up visits, and continued fractures, patient was referred to genetic testing at CHOP where he was

found positive for Heterozygous COL1A1 gene OI.

Results

XR Bone Survey:

Healing posterior rib fractures of the right ninth, 10th and 11th ribs and left 10th rib.

Healing corner fracture of the proximal left tibia.

Compression fractures of the T8, L1, and L2 vertebral bodies.

Subtle curvilinear sclerosis at distal left radial metaphysis representing a corner fracture.

Mild congenital foreshortening of the proximal radii bilaterally.

Genetic Testing Results:

Positive heterozygous COL1A1 gene

Imaging

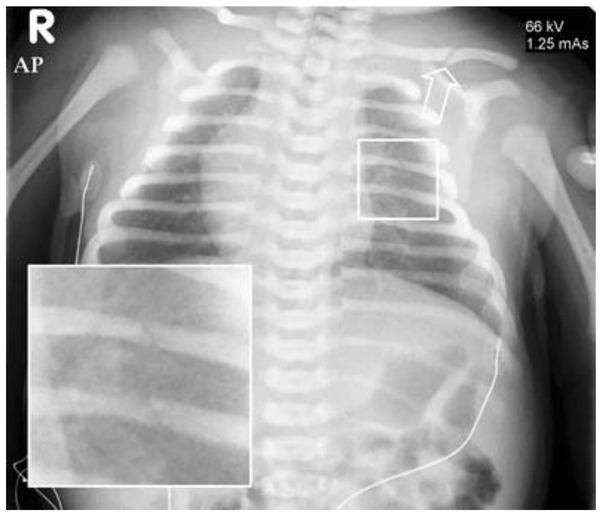

Figure 1: Demonstrates an example of posterior rib fractures of the left 4th and 5th ribs. This image has no reference to patient in case report. This is courtesy of Pediatric Radiology (Kubota)

Discussion

Osteogenesis imperfecta (OI) is a rare genetic disorder that affects the bones, causing them to be brittle and easily broken. It is also known as brittle bone disease. OI is caused by a mutation in one of the genes that make collagen, a protein that provides structure and strength to bones.

There are four types of OI, ranging from mild to severe. People with mild OI may only experience a few fractures in their lifetime, while those with severe OI may have hundreds of fractures and be at risk for respiratory complications and spinal deformities.

The symptoms of OI vary depending on the type and severity of the condition. Common symptoms include frequent bone fractures, bone deformities, joint laxity, short stature, and blue or gray tinted sclerae

There is currently no cure for OI, but treatment can help manage symptoms and improve quality of life. Treatment may include physical therapy, surgery, medication, and assistive devices such as braces or wheelchairs.

Research into OI is ongoing, with a focus on developing new treatments and improving the understanding of the genetic and molecular basis of the condition. Additionally, there are organizations and support groups dedicated to providing resources and support to individuals and families affected by OI.

Conclusion

Osteogenesis Imperfecta is a rare genetic disorder that affects the bones and can lead to recurrent fractures and other complications. Diagnosis is based on clinical features and confirmed by genetic testing. Treatment is aimed at managing symptoms and preventing complications, and ongoing research is focused on developing new treatments and improving understanding of the molecular basis of the condition. With proper management and support, people with OI can lead fulfilling and productive lives.

References

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