Juvenile Primary Osteoporosis

Subjects: Genetics & Heredity
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Juvenile primary osteoporosis is a skeletal disorder characterized by thinning of the bones (osteoporosis) that begins in childhood. Osteoporosis is caused by a shortage of calcium and other minerals in bones (decreased bone mineral density), which makes the bones brittle and prone to fracture.

Keywords: genetic conditions

1. Introduction

Affected individuals often have multiple fractures in the long bones of the arms and legs, especially in the regions where new bone forms (metaphyses). They also have fractures in the bones that form the spine (vertebrae), which can cause collapse of the affected vertebrae (compressed vertebrae). Multiple fractures can cause bone pain and lead to movement problems.

2. Frequency

The prevalence of juvenile primary osteoporosis is unknown. Nearly 1 in 10 adults over age 50 have osteoporosis, but the condition is uncommon in children. Osteoporosis can occur at a young age as a feature of other conditions but rarely occurs without other signs and symptoms (primary osteoporosis).

3. Causes

Mutations in the *LRP5* gene can cause juvenile primary osteoporosis. This gene provides instructions for making a protein that participates in a chemical signaling pathway that affects the way cells and tissues develop. In particular, the LRP5 protein is involved in the regulation of bone mineral density.

LRP5 gene mutations that cause juvenile primary osteoporosis result in an LRP5 protein that cannot transmit signals along the pathway. The resulting reduction in signaling impairs proper bone development, causing decreased bone mineral density and osteoporosis at a young age.

Many people with childhood-onset osteoporosis do not have a mutation in the *LRP5* gene. (When its cause is unknown, the condition is often called idiopathic juvenile osteoporosis). It is likely that mutations in other genes that have not been identified are involved in this condition.

3.1. The gene associated with Juvenile primary osteoporosis

- LRP5

4. Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

5. Other Names for This Condition

- childhood-onset primary osteoporosis
- idiopathic juvenile osteoporosis

References

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