Léri-Weill Dyschondrosteosis

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Léri-Weill dyschondrosteosis is a disorder of bone growth.

Keywords: genetic conditions

1. Introduction

Affected individuals typically have shortening of the long bones in the arms and legs (mesomelia). As a result of the shortened leg bones, people with Leri-Weill dyschondrosteosis typically have short stature. Most people with the condition also have an abnormality of the wrist and forearm bones called Madelung deformity, which may cause pain and limit wrist movement. This abnormality usually appears in childhood or early adolescence. Other features of Léri-Weill dyschondrosteosis can include increased muscle mass (muscle hypertrophy); bowing of a bone in the lower leg called the tibia; a greater-than-normal angling of the elbow away from the body (increased carrying angle); and a high arched palate.

Léri-Weill dyschondrosteosis occurs in both males and females, although its signs and symptoms tend to be more severe in females. Researchers believe that the more severe features may result from hormonal differences.

2. Frequency

The prevalence of Léri-Weill dyschondrosteosis is unknown. It is diagnosed more often in females than in males.

3. Causes

Most cases of Léri-Weill dyschondrosteosis result from changes involving the *SHOX* gene. The protein produced from this gene plays a role in bone development and is particularly important for the growth and maturation of bones in the arms and legs. The most common cause of Léri-Weill dyschondrosteosis is a deletion of the entire *SHOX* gene. Other genetic changes that can cause the disorder include mutations in the *SHOX* gene or deletions of nearby genetic material that normally helps regulate the gene's activity. These changes reduce the amount of SHOX protein that is produced. A shortage of this protein disrupts normal bone development and growth, which underlies the major features of Léri-Weill dyschondrosteosis.

In affected people who do not have a genetic change involving the *SHOX* gene, the cause of the condition is unknown.

3.1. The gene associated with Léri-Weill dyschondrosteosis

- SHOX

4. Inheritance

Léri-Weill dyschondrosteosis has a pseudoautosomal dominant pattern of inheritance. The *SHOX* gene is located on both the X and Y chromosomes (sex chromosomes) in an area known as the pseudoautosomal region. Although many genes are unique to either the X or Y chromosome, genes in the pseudoautosomal region are present on both sex chromosomes. As a result, both females (who have two X chromosomes) and males (who have one X and one Y chromosome) normally have two functional copies of the *SHOX* gene in each cell. The inheritance pattern of Léri-Weill dyschondrosteosis is described as dominant because one missing or altered copy of the *SHOX* gene in each cell is sufficient to cause the disorder. In females, the condition results when the gene is missing or altered on one of the two copies of the X chromosome; in males, it results when the gene is missing or altered on either the X chromosome or the Y chromosome.

A related skeletal disorder called Langer mesomelic dysplasia occurs when both copies of the *SHOX* gene are mutated in each cell. This disorder has signs and symptoms that are similar to, but typically more severe than, those of Léri-Weill dyschondrosteosis.

5. Other Names for This Condition

- DCO
- dyschondrosteosis
- Leri-Weill dyschondrosteosis
- LWD

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