Léri-Weill dyschondrosteosis

Léri-Weill dyschondrosteosis is a disorder of bone growth. 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 Léri-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.

Frequency

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

Causes

Most cases of Léri-Weill dyschondrosteosis result from changes involving the \textit{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 \textit{SHOX} gene. Other genetic changes that can cause the disorder include mutations in the \textit{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 \textit{SHOX} gene, the cause of the condition is unknown.

Inheritance Pattern

Léri-Weill dyschondrosteosis has a pseudoautosomal dominant pattern of inheritance. The \textit{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 \textit{SHOX} gene in each cell. The inheritance pattern of Léri-Weill dyschondrosteosis is described as dominant because one missing or altered copy of the \textit{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 \textit{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.

\textbf{Other Names for This Condition}

- DCO
- dyschondrosteosis
- Leri-Weill dyschondrosteosis
- LWD

\textbf{Diagnosis & Management}

\textbf{Genetic Testing Information}

- What is genetic testing? [primer/testing/genetictesting](https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265309/)

\textbf{Other Diagnosis and Management Resources}


\textbf{Additional Information & Resources}

\textbf{Health Information from MedlinePlus}

- Health Topic: Bone Diseases [https://medlineplus.gov/bonediseases.html](https://medlineplus.gov/bonediseases.html)

\textbf{Genetic and Rare Diseases Information Center}

Educational Resources

• Boston Children's Hospital: Madelung’s Deformity  
http://www.childrenshospital.org/conditions-and-treatments/conditions/m/madelungs-deformity

• MalaCards: leri-weill dyschondrosteosis  
https://www.malacards.org/card/leri_weill_dyschondrosteosis

• Orphanet: Léri-Weill dyschondrosteosis  
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=240

Patient Support and Advocacy Resources

• Human Growth Foundation  
https://www.hgfound.org/

• International Skeletal Dysplasia Registry, UCLA  
https://www.uclahealth.org/ortho/isdr

• Little People of America  
https://www.lpaonline.org/

• Little People UK  
https://littlepeopleuk.org/

• Resource list from the University of Kansas Medical Center: Dwarfism / Short Stature  
http://www.kumc.edu/gec/support/dwarfism.html

• The MAGIC Foundation  
https://www.magicfoundation.org/

Clinical Information from GeneReviews

• SHOX Deficiency Disorders  
https://www.ncbi.nlm.nih.gov/books/NBK1215

Scientific Articles on PubMed

• PubMed  
https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28leri-weill+dyschondrosteosis%5BTIAB%5D%29+OR+%28dyschondrosteosis%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

• LERI-WEILL DYSCHONDROSTEOSIS  
http://omim.org/entry/127300
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9590292

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