

# SHOX Deficiency

SHOX deficiency is the most common monogenic cause of short stature, with a highly variable phenotype that is often accompanied by shortened extremities<sup>1-3</sup>

The phenotypic spectrum of SHOX deficiency includes conditions of short stature and skeletal abnormalities<sup>1,3</sup>

## SHOX Gene<sup>1,4</sup>

The *short stature homeobox (SHOX)* gene, which is located on the X and Y chromosomes, encodes a transcription factor that is expressed in developing skeletal tissue, and plays an important role in the differentiation and proliferation of chondrocytes

## Prevalence of SHOX Variants in Certain Conditions<sup>2</sup>

Short stature

2 to 15%

Léri-Weill  
dyschondrosteosis

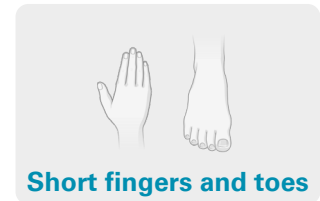
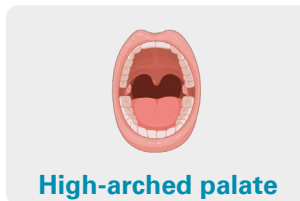
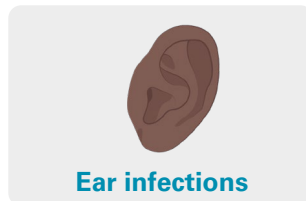
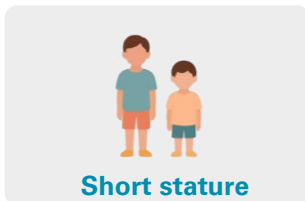
50 to 90%

Turner syndrome

Nearly 100%

## Clinical Presentation<sup>3,5</sup>

Individuals with SHOX deficiency may experience skeletal abnormalities and malformations with resulting manifestations including:



## Diagnosis and Management

SHOX deficiency can be diagnosed through genetic testing or supported by clinical findings<sup>3</sup>

Growth hormone treatment is approved for use in individuals with SHOX deficiency and has shown effectiveness in improving growth outcomes<sup>2,6-8</sup>

**Abbreviations:** SHOX, short stature homeobox.

**References:** 1. Marchini A, et al. *Endocr Rev.* 2016;37(4):417-448. 2. Binder G. *Horm Res Paediatr.* 2011;75(2):81-89. 3. Binder G, Rappold GA. SHOX Deficiency Disorders (Updated 2005). In: Adam MP, Bick S, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, eds. GeneReviews®. 1993. 4. Hoyer-Kuhn H, et al. *J Pediatr Endocrinol Metab.* 2018;31(1):25-31. 5. Oliveira CS, Alves C. *Endocrinol Nutr.* 2011;58(8):433-442. 6. Genoni G, et al. *Pediatr Res.* 2018;83(2):438-444. 7. Blum WF, et al. *J Clin Endocrinol Metab.* 2007;92(1):219-228. 8. Blum WF, et al. *J Clin Endocrinol Metab.* 2013;98(8):E1383-E1392.