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[1-3]

The phenotypic spectrum of SHOX deficiency includes conditions of short stature and skeletal abnormalities[1,3]

SHOX Gene[1,4]

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

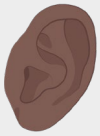
Prevalence of *SHOX* Variants in Certain Conditions[2]

Short stature	2 to 15%
Léri-Weill dyschondrosteosis	50 to 90%
Turner syndrome	Nearly 100%

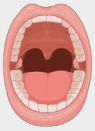
Clinical Presentation[3,5]

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

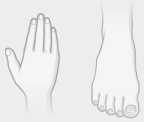
Short stature



Ear infections



High-arched palate



Short fingers and toes

Diagnosis and Management

SHOX deficiency can be diagnosed through genetic testing or supported by clinical findings[3]

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

Abbreviations: SHOX, short stature homeobox.

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