The SHOX gene („Short Stature Homeobox-containing Gene“) was identified during research of genotype-phenotype corelations in patiens with Turner Syndrome. Absence one alele of this gene was the cause of short stature in these girls. Shortly after, mutations in SHOX gene were identified in patients with Léri-Weill and Langer syndrom, thus in patients with growth failure and skeletal deformities.

It is estimated that mutations in SHOX gene or mutations in SHOX regulatory regions affect one in thousand of new born children. Mutations in this gene are one of the most common genetic causation leading to growth failure phenotype.

However, the exact role of SHOX gene in bone growth and development is still unknown, therefore it is importnant to study problems with SHOX gene and try to discover mechanism of SHOX protein activity on molecular levels.