The role of SHOX gene in Idiopathic Short Stature: An Italian multicenter study

The short stature homeobox-containing (SHOX) gene, located in the telomeric pseudoautosomal region 1 (PAR1) on the short arm of both sex chromosomes, is important for linear growth.

**AIM:** to evaluate the presence of SHOX gene deletions/point mutations in children with short stature in order to understand the role of SHOX gene in Idiopathic Short Stature (ISS) and estimate its frequency.

**BACKGROUND**

This study, supported by the Eli Lilly Italia and approved by the Italian Society for Pediatric Endocrinology and Diabetes (ISPED), is a multicenter study involving several Italian Pediatric Endocrinology Units. Out of a total number of 152 blood samples received, 68 were from patients with ISS.

**RESULTS**

Genomic DNA was extracted and used for Multiplex Ligation-dependent Probe Amplification (MLPA) and sequencing analysis. MLPA was performed in the Pediatric Laboratory of Parma, using the SALSA MLPA P018-F1 SHOX probemix kit, analyzing both the coding region and the enhancer of the SHOX gene. Deletions and duplications were confirmed with Copy Number Assay (Applied Biosystems) in Real Time PCR.

**CONCLUSIONS**

If we exclude the patients with Leri-Weill syndrome who presented a SHOX gene deletion in 50%, in our cohort of patients with ISS the incidence of SHOX gene alterations was very low (4.4%), suggesting that the presence of mesomelia, minor skeletal abnormalities, and eventually subtle radiographic signs are essential for requiring genetic analysis properly.