Clinical Application
• The short stature homeobox-containing (SHOX) gene is located in the pseudoautosomal 1 (PAR 1) region of the X and Y chromosomes.1,2
  • Haploinsufficiency due to deficiency of 1 copy of the SHOX gene can result in a range of phenotypes from short stature to Leri-Weill dychondrosteosis (LWD).3-10
  • Haploinsufficiency of the SHOX gene is indicated as the cause of short stature in Turner syndrome.1,2,11,12
  • Deficiency of both copies of the SHOX gene results in the severe growth retardation condition Langer mesomelic dysplasia (LMD).3,4,13,14
• Studies indicate that 2% to 15% of children with idiopathic short stature have mutations in the SHOX gene.7-10 Those children who have SHOX gene mutations are responsive to growth hormone therapy.15,16
• Identification of a SHOX gene mutation in a patient may be useful for:
  • Establishing a genetic basis for idiopathic short stature.
  • Family studies.
  • Confirming a diagnosis of LWD.
  • Guiding therapeutic decisions.
• Comprehensive testing is performed by PCR amplification and DHPLC screening for mutations in and complete deletion of the SHOX gene.
  • Specific mutations in the SHOX gene are identified by DNA sequence analysis.
  • Single nucleotide polymorphism (SNP) analysis is performed across the SHOX gene to detect whole gene deletion.

Scientific Expertise
• Industry leading endocrine sciences laboratory with a history exceeding 30 years
• Extensive pituitary and androgen disorder test menu and expertise
• Assay method developed, validated, and maintained on site by PhD-level scientists
• Complete normative data for children and adults
• PhD and MD consultation available

Superior Service
• Comprehensive services for the endocrinology specialist
• Broad network of managed care health plans
• Flexible connectivity options for test ordering and result reporting
• Patient service centers available nationwide
• Courier and logistics services
• Local sales representation

SHOX Gene Analysis
A Diagnostic Tool for Children With Short Stature

Whole blood (EDTA)
Genomic DNA
PCR amplification of exons and intragenic SNPs using custom primers
Heteroduplex formation
DHPLC
Exon variant(s)
No exon variants
DNA sequencing to identify exon variant(s)
Evaluate intragenic SNPs for absence of heterozygosity
SHOX Gene Analysis
A Diagnostic Tool for Children With Short Stature

SHOX, DHPLC

Specimen 3.0 mL whole blood
Minimum Volume 1.0 mL whole blood
Container Lavender-top (EDTA) tube
Storage Instructions Maintain specimen at room temperature. Stable refrigerated for up to seven days.
Method Mutation analysis by PCR, DHPLC, and sequencing as needed
Schedule/Turnaround Time Performed M-F, TAT 14-21 days

References

Please contact your local account representative for more information.
For the most current information regarding test options, including specimen requirements and CPT codes, please consult the online Test Menu at www.LabCorp.com.

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