

SHOX Gene Analysis

A Diagnostic Tool for Children With Short Stature

Clinical Application

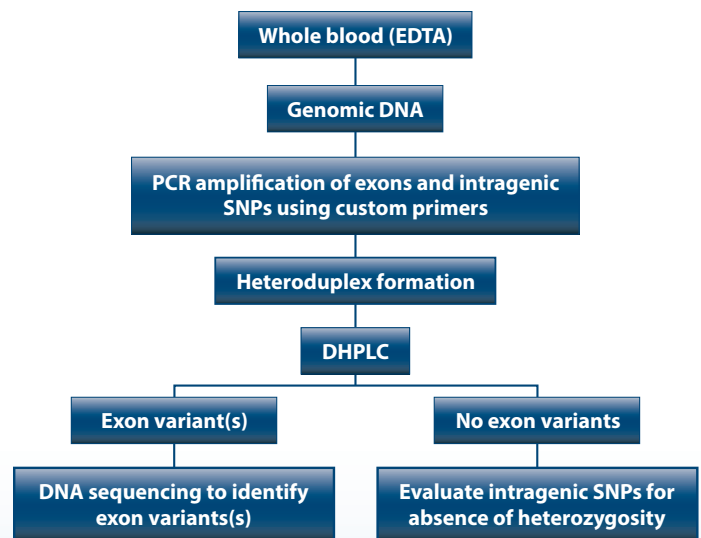
- The short stature homeobox-containing (*SHOX*) gene is located in the pseudoautosomal 1 (PAR 1) region of the X and Y chromosomes.^{1,2}
 - Haploinsufficiency due to deficiency of 1 copy of the *SHOX* gene can result in a range of phenotypes from short stature to Leri-Weill dyschondrosteosis (LWD).³⁻¹⁰
 - 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



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SHOX, DHPLC.....500110

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

Esoterix Direct Accounts: Please use Esoterix test code **504005**

References

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