

**New Test**      **3004603**      **SHOX Deficiency Disorders, Sequencing and Deletion/Duplication**      **SHOX NGS**



Patient History for SHOX Deficiency Disorders Testing



Additional Technical Information

**Methodology:** Massively Parallel Sequencing/ Multiplex Ligation-dependent Probe Amplification  
**Performed:** Varies  
**Reported:** 3-6 weeks

**Specimen Required:** Collect: Lavender or pink (EDTA) or yellow (ACD solution A or B).  
Specimen Preparation: Transport 3 mL whole blood. (Min: 2 mL)  
Storage/Transport Temperature: Refrigerated  
Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens; saliva, buccal brush, or swab; FFPE tissue; DNA.  
Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

**Reference Interval:** By report

**Interpretive Data:**  
Refer to report

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

**Note:** Gene Tested: *SHOX* (NM\_000451), exon 6b (NM\_006883) is not sequenced.

**CPT Code(s):** 81405; 81479

New York DOH approval pending. Call for status update.

**HOTLINE NOTE:** Refer to the Test Mix Addendum for interface build information.