

THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

PATIENT HISTORY FOR SHOX-RELATED DISORDERS TESTING

Patient Name: _____ **Date of Birth:** _____ **Sex:** Female Male
Ordering Provider: _____ **Provider's Phone:** _____
Practice Specialty: _____ **Provider's Fax:** _____
Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

African American/Black Asian Hispanic White Other: _____

List country of origin (if known): _____

Indication for Testing (REQUIRED): Does the patient have a clinical/suspected diagnosis? No Yes (check all that apply)

Isolated/Idiopathic short stature Langer mesomelic dysplasia (LMD) Turner Syndrome
 Leri-Weill dyschondrosteosis (LWD) Carrier Testing
 Other chromosome abnormality (specify): _____
 Other indication (specify): _____

Does the patient have symptoms? No Yes (check all that apply and describe)

<input type="checkbox"/> Madelung deformity (abnormal alignment of bones at the wrist)	<input type="checkbox"/> Short forearm <input type="checkbox"/> Cubitus Valgus <input type="checkbox"/> Appearance of muscular hypertrophy	<input type="checkbox"/> Reduced arm span/height ratio (<0.965): _____ <input type="checkbox"/> Increased sitting height/height ratio (>0.555): _____ <input type="checkbox"/> Patient's height (percentile): _____ <input type="checkbox"/> Parent's height – Mother: _____ Father: _____
<input type="checkbox"/> Short Stature	<input type="checkbox"/> Dislocation of ulna (at elbow)	<input type="checkbox"/> Body-mass index (BMI): _____
<input type="checkbox"/> Mesomelia		
<input type="checkbox"/> Bowing of the foreman		
<input type="checkbox"/> Other symptom(s): _____		

Has the patient undergone previous genetic testing for short stature or SHOX-related disorders? No Yes Unknown

If yes, describe the test(s) and results: Chromosome analysis (karyotype): _____
 FISH: _____
 Microarray: _____
 DNA testing: _____

Is there any relevant family history of SHOX-related findings? No Yes Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset: _____

Has genetic testing been performed for the family member(s)? No Yes Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing) or specify results/findings: _____

Check the test you intend to order.

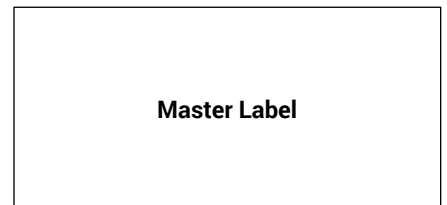
Available testing for SHOX-related disorders:

3001401 SHOX-Related Disorders, Deletion/Duplication Analysis with Reflex to Sequencing: Most comprehensive test for confirmation of a SHOX-related disorder; reflexes to sequencing based on indication for testing and result of deletion/duplication testing.

Targeted testing for known familial variant (laboratory report from family member is REQUIRED):

2001961 Familial Mutation, Targeted Sequencing: Tests for a sequence variant previously identified in a family member; a copy of relative's lab result is REQUIRED.

3003144 Deletion/Duplication Analysis by MLPA: Tests for large deletion/duplication previously identified in a family member; a copy of a relative's lab report is REQUIRED.



For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.