

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

PATIENT HISTORY FOR SHOX DEFICIENCY DISORDERS TESTING

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

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

African American/Black Asian Hispanic White Other: _____

List country of origin (if known): _____

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

Has the patient undergone previous genetic testing for short stature or SHOX deficiency 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 deficiency findings?..... No Yes Unknown

If yes, attach a pedigree or specify the relatives' 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 relatives DNA laboratory result (REQUIRED for familial mutation testing) or specify results/findings:

Check the test you intend to order.

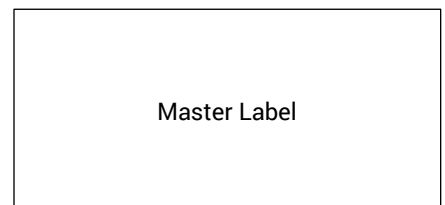
Available testing for SHOX deficiency disorders:

3004603 SHOX Deficiency Disorders, Sequencing and Deletion/Duplication: Detects pathogenic variants in the *SHOX* gene, causative for SHOX deficiency disorders (isolated short stature [ISS], Leri-Weill dyschondrosteosis [LWD] and Langer mesomelic dysplasia [LMD]).

Targeted testing for known familial variant:

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.