ARUP LABORATORIES

A nonprofit enterprise of the University of Utah and its Department of Pathology

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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)

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

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.

Master Label

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