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*-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)

☐ Madelung deformity (abnormal alignment of bones at the wrist)
☐ Short Stature
☐ Mesomelia
☐ Bowing of the foreman
☐ 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*-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.

Master Label

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