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 Assigned at Birth: ☐Female ☐Male ☐Intersex **Gender Identity (optional):** ☐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 <u>symptoms</u>? ☐ 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 – Parent 1: _____
Parent 2: _____
☐ 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 <u>family history</u> of SHOX deficiency findings? ☐ No ☐ Yes ☐ Unknown

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

_____

_____

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 (<u>REQUIRED for familial variant testing</u>) or specify results/findings:

_____

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

Master Label