ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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 fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR FREEMAN-SHELDON SYNDROME TESTING

Patient Name _____ **Date of Birth** _____ **Sex** ☐ F ☐ M

Physician _____ **Physician Phone** _____

Practice Specialty _____ **Physician Fax** _____

Genetic Counselor _____ **Counselor Phone** _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American

☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

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

☐ Camptodactyly
☐ Clubfoot
☐ Congenital hearing loss
☐ Cryptorchidism
☐ Dental crowding
☐ Hips/elbow contractures
☐ H-shaped dimpling of the chin
☐ Inguinal hernia
☐ Scoliosis
☐ Small pinched mouth
☐ Strabismus
☐ Ulnar deviation of the wrists and fingers
☐ Underdeveloped/absent flexion creases of the hands
☐ Other symptom(s): _____

Has the patient undergone previous testing for Freeman-Sheldon syndrome? ☐ No ☐ Yes ☐ Unknown

If yes, describe the test(s) and results: _____

_____

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown

If yes, indicate: ☐ Freeman-Sheldon syndrome ☐ Contractures ☐ Neither ☐ Unknown

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

_____

_____

The following testing is available:

2002662 Freeman-Sheldon Syndrome (*MYH3*) Sequencing Exon 17: Offer to patients who have multiple joint and facial contractures. Clinical sensitivity is ~70%.

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

Master Label