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 EHLERS-DANLOS KYPHOSCOLIOTIC FORM (TYPE VI) 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)

☐ Arterial rupture
☐ Atrophic scarring
☐ Hypotonia
☐ Joint hypermobility
☐ Kyphoscoliosis
☐ Respiratory compromise
☐ Thin hyperextensible skin
☐ Other symptom(s): _____

Laboratory Findings

Deoxypyridinoline: Pyridinoline crosslinks in urine:
☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown List ratio: _____

Lysyl hydroxylase activity in fibroblasts:
☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown List percent activity: _____

Collagen screening in fibroblasts:
☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown
If yes, attach a pedigree or specify the relative's relationship to the patient. _____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown
If yes, list the *PLOD1* mutations in the family member: _____
The relative is: ☐ a healthy carrier ☐ affected with EDS VI

Check the test you intend to order.

☐ **0080351 Ehlers-Danlos Syndrome Type VI Screen:** Urine screen, ordered as first line test before DNA.

☐ **2005559 Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (*PLOD1*) Sequencing and Deletion/Duplication:** Sequencing and deletion/duplication analysis of *PLOD1* coding regions and intron/exon boundaries. Clinical sensitivity of this test is unknown.

☐ **2005555 Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (*PLOD1*) Deletion/Duplication:** Deletion/duplication analysis of *PLOD1* coding regions. Clinical sensitivity of this test is approximately 20%.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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