ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform EDS VI testing.
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_____ /_____/_____Gender [ ] F [ ] M

Physician __ _____ Physician Phone (_____)_____Practice Specialty_____

Genetic Counselor _____ Counselor Phone (_____)_____

Patient's Ethnicity (check all that apply)

[ ] African American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] Other_____

Does the patient have SYMPTOMS? [ ] No [ ] Yes [ ] Unknown If yes, check all that apply

[ ] Kyphoscoliosis [ ] Thin hyperextensible skin [ ] Arterial rupture
[ ] Hypotonia [ ] Atrophic scarring [ ] Respiratory compromise
[ ] Joint hypermobility [ ] Other _____

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

Does the patient have a FAMILY HISTORY OF EDS VI? [ ] No [ ] Yes

If yes, attach a PEDIGREE or specify the **RELATIONSHIP** of family member(s) to the patient. _____

Is the relative [ ] a healthy carrier or [ ] affected with EDS VI?

What are the *PLOD1* mutations in the family member? _____

Circle 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: A copy of the relative's DNA lab result is REQUIRED for this test.

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

Master Label