

THIS IS NOT A TEST REQUEST FORM. Please complete and submit 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:** 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): _____

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

Deoxyypyridinoline: 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.
- 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.