

**THIS IS NOT A TEST REQUEST FORM.**  
**The information below is required to perform Loeys-Dietz testing.**  
**Please fill out this form and submit it with the test request form or electronic packing list.**

**PATIENT HISTORY FOR LOEYS-DIETZ 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)

- |   |   |  |                                      |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African-American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian           | <input type="checkbox"/> Caucasian   |
| <input type="checkbox"/> Hispanic         | <input type="checkbox"/> Middle Eastern   | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

**Does patient have SYMPTOMS of Loeys-Dietz Syndrome (LDS)?**  No  Yes (check all that apply)

- |  |  |  |   |
|--|--|--|---|
| <input type="checkbox"/> <b>Vascular</b>           | <input type="checkbox"/> <b>Craniofacial</b> | <input type="checkbox"/> <b>Cutaneous</b>    | <input type="checkbox"/> <b>Musculoskeletal</b>     |
| <input type="checkbox"/> Aortic Dilation (____cm)  | <input type="checkbox"/> Bifid uvula         | <input type="checkbox"/> Easy bruising       | <input type="checkbox"/> Arachnodactyly             |
| <input type="checkbox"/> Arterial dissection _____ | <input type="checkbox"/> Cleft palate        | <input type="checkbox"/> Poorly formed scars | <input type="checkbox"/> Club foot                  |
| <input type="checkbox"/> Thoracic aneurysm         | <input type="checkbox"/> Craniosynostosis    | <input type="checkbox"/> Translucent skin    | <input type="checkbox"/> Joint laxity               |
| <input type="checkbox"/> Cerebral aneurysm         | <input type="checkbox"/> Hypertelorism       | <input type="checkbox"/> Velvety skin        | <input type="checkbox"/> Pectus excavatum/carinatum |
| <input type="checkbox"/> Abdominal aneurysm        |  |  | <input type="checkbox"/> Scoliosis                  |

Other \_\_\_\_\_

**Is there any relevant FAMILY HISTORY?**  No  Yes  Unknown

**If yes, attach PEDIGREE, or specify the RELATIONSHIP to the patient and list the symptoms and age of onset:**

\_\_\_\_\_

**Please attach a copy of the relative's DNA laboratory result. (REQUIRED for familial mutation testing)**

**Has the patient undergone previous DNA testing for Loeys-Dietz Syndrome?**  No  Yes  Unknown

If yes, please describe test(s) and results \_\_\_\_\_

**Circle the test you intend to order.**

**2006540 Aortopathy Panel Sequencing and Deletion/Duplication, 17 Genes**

- Confirm diagnosis of an aortopathy in individuals with aortic/vascular aneurysm, dissection, or rupture
- Includes 4 genes associated with Loeys Dietz (*TGFBR1*, *TGFBR2*, *TGFB2*, *SMAD3*) and 13 others associated with various aortopathies (*ACTA2*, *CBS*, *COL3A1*, *COL5A1*, *COL5A2*, *FBN1*, *FBN2*, *MYH11*, *MYLK*, *PLOD1*, *SKI*, *SLC2A10*, *SMAD4*).

**2002705 Loeys-Dietz Syndrome (*TGFBR1* and *TGFBR2*) Sequencing**

Clinical sensitivity is ~95% for Loeys-Dietz.

**2001961 Familial Mutation, Targeted Sequencing**

Tests for a previously identified familial mutation. A copy of a relative's DNA laboratory result is REQUIRED.

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

Master Label