

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

PATIENT HISTORY FOR NOONAN OR LEOPARD SYNDROME 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 the patient have SYMPTOMS? No Yes (check all that apply)

- | | | |
|--|--|---|
| <input type="checkbox"/> Broad webbed neck | <input type="checkbox"/> Low set nipples | <input type="checkbox"/> Coagulation disorder _____ |
| <input type="checkbox"/> Characteristic facies | <input type="checkbox"/> Nevi, lentiginos, or café au lait patches | <input type="checkbox"/> Lymphatic dysplasia _____ |
| <input type="checkbox"/> Cryptorchidism | <input type="checkbox"/> Pectus excavatum/carinatum | <input type="checkbox"/> Ocular findings _____ |
| <input type="checkbox"/> Deafness | <input type="checkbox"/> Pulmonary valve stenosis | <input type="checkbox"/> Other cardiac defect _____ |
| <input type="checkbox"/> Developmental delay | <input type="checkbox"/> Short stature | <input type="checkbox"/> Other _____ |
| <input type="checkbox"/> Hypertrophic cardiomyopathy | | |

FAMILY MEMBER(S) with a confirmed diagnosis of Noonan/LEOPARD syndrome? No Yes Unknown

If yes, attach a PEDIGREE or specify the RELATIONSHIP of the family member(s) to the patient and detail the symptoms in each affected relative.

Has DNA testing been performed for these family member(s)? No Yes Unknown

If yes, attach copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

FAMILY MEMBER(S) with symptoms suggestive of Noonan or LEOPARD syndrome? No Yes Unknown

Describe symptoms and relationship(s) to patient _____

Has the patient undergone previous DNA testing for Noonan/LEOPARD syndrome? No Yes Unknown

If yes, please describe test(s) and results _____

Circle the Noonan/LEOPARD test you intend to order.

2004189 Noonan Syndrome, *PTPN11* Sequencing with Reflex to *SOS1* Sequencing: To confirm a clinical diagnosis of Noonan Syndrome; clinical sensitivity is approximately 70% for Noonan syndrome.

0051805 Noonan Syndrome (*PTPN11*) Sequencing: To confirm a clinical diagnosis of Noonan or LEOPARD Syndrome; clinical sensitivity is approximately 50-60% for Noonan syndrome and 90% for LEOPARD.

2004195 Noonan Syndrome (*SOS1*) Sequencing: To confirm a clinical diagnosis of Noonan Syndrome; clinical sensitivity is approximately 10% for Noonan syndrome.

2001961 Familial Mutation, Targeted Sequencing: Tests for a previously identified *PTPN11* familial mutation; copy of a relative's lab result is REQUIRED.

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

Master Label