ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

500 Chipeta Way
Salt Lake City, Utah 84108-1221
phone: (801) 583-2787 | toll free: (800) 242-2787
fax: (801) 584-5249 | www.aruplab.com

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 NOONAN SPECTRUM DISORDERS 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: _____

Patient's diagnosis: ☐ Confirmed ☐ Suspected ☐ Unknown

☐ Cardiofaciocutaneous syndrome (CFCS) ☐ LEOPARD syndrome ☐ Noonan syndrome-like with loose anagen hair
☐ Costello syndrome ☐ Noonan syndrome (NS)
☐ Other: _____

Does the patient have symptoms of a Noonan Spectrum disorder? ☐ No ☐ Yes (check all that apply and describe)

Postnatal Findings

☐ Nevi, lentigines, or café au lait macules
☐ Intellectual disability
☐ Hearing loss
☐ Lymphatic dysplasia: _____
☐ GI: _____
☐ Malignancy: _____
☐ Coagulation disorder: _____
☐ Renal: _____
☐ Other symptom(s): _____

Postnatal Findings (continued)

Dysmorphic Features
☐ Broad webbed neck
☐ Characteristic facies
☐ Cryptorchidism
☐ Curly/sparse/ thin hair
☐ Low set nipples
☐ Macrocephaly
☐ Poor growth/ FTT

Postnatal Findings (continued)

Musculoskeletal
☐ Short stature
☐ Pectus excavatum/carinatum

Cardiac
☐ Pulmonary valve stenosis
☐ Hypertrophic cardiomyopathy

Perinatal Findings

☐ Cystic hygroma
☐ Hydronephrosis
☐ Increased nuchal translucency
☐ Polyhydramnios

Has the patient undergone previous testing for this condition? ☐ No ☐ Yes ☐ Unknown

☐ Chromosome analysis ☐ Normal ☐ Abnormal (please attach the report) ☐ Not Performed
☐ Microarray (aCGH) ☐ Normal ☐ Abnormal (please attach the report) ☐ Not Performed
☐ Other (describe gene, method, result)_____

Does this patient have genetic variant(s) previously identified in tumor/bone marrow? ☐ No ☐ Yes ☐ Unknown
If yes, attach result or describe: _____

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown
If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:

_____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown
If yes, attach a copy of the relative's DNA laboratory result. (REQUIRED for familial mutation testing.)

Check the test you intend to order.

☐ **2010772 Noonan Spectrum Disorders Panel, Sequencing**- recommended first tier test for Noonan Spectrum Disorders
☐ **2010769 Noonan Spectrum Disorders Panel, Sequencing, Fetal-** order maternal cell contamination analysis separately (0050608).
☐ **0051805 Noonan Syndrome (*PTPN11*) Sequencing**
☐ **2004195 Noonan Syndrome (*SOS1*) Sequencing**
☐ **2001961 Familial Mutation Targeted Sequencing**- targeted testing for a familial variant; a copy of the family member's laboratory report is REQUIRED.

Master Label

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