ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform molecular genetic testing.
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_____ /_____/_____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 _____

Patient's Clinical Diagnosis [ ] Confirmed [ ] Suspected [ ] Unknown
[ ] Noonan syndrome (NS) [ ] LEOPARD syndrome [ ] Noonan syndrome-like with loose anagen hair
[ ] Cardiofaciocutaneous syndrome (CFCS) [ ] Costello syndrome [ ] Other_____

Does the patient have SYMPTOMS of a Noonan spectrum disorder? [ ] No [ ] Yes; please check all that apply

Perinatal Findings
[ ] Cystic hygroma [ ] Hydronephrosis [ ] Increased nuchal translucency [ ] Polyhydramnios
[ ] Other _____

Postnatal Findings

Dysmorphic Features	**Cardiac**	**Musculoskeletal**
[ ] Broad webbed neck	[ ] Pulmonary valve stenosis	[ ] Short stature
[ ] Characteristic facies	[ ] Hypertrophic cardiomyopathy	[ ] Pectus excavatum/carinatum
[ ] Cryptorchidism	[ ] Other _____	[ ] Other _____
[ ] Curly/sparse/ thin hair		
[ ] Low set nipples	**Cutaneous**	**Neurological**
[ ] Macrocephaly	[ ] Nevi, lentigines, or café au lait macules	[ ] Intellectual disability
[ ] Poor growth/ FTT	[ ] Other _____	[ ] Other _____
Sensory	**[ ] Lymphatic dysplasia _____**	**[ ] GI _____**
[ ] Hearing loss	**[ ] Malignancy _____**	**[ ] Coagulation disorder _____**
[ ] Ocular _____	**[ ] Renal _____**	**[ ] Other _____**

Does the patient have a FAMILY HISTORY of a Noonan spectrum disorder? [ ] No [ ] Yes [ ] Unknown
If yes, attach a PEDIGREE or specify the relatives' RELATIONSHIP to the patient. List relatives' symptoms & age of onset:
_____

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

Has the patient undergone previous genetic testing? [ ] No [ ] Yes [ ] Unknown

Chromosome analysis [ ] normal [ ] abnormal (please attach the report) [ ] not performed
Microarray (aCGH) [ ] normal [ ] abnormal (please attach the report) [ ] not performed
Other (please provide gene name and methodology if DNA testing or attach the report): _____ [ ] normal [ ] abnormal
_____

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

Circle the test you intend to order OR write the test name and number below:

Recommended first tier testing for Noonan spectrum disorders:	
2010772	Noonan Spectrum Disorders Panel, Sequencing, 15 Genes: May be used for prenatal or postnatal testing. If ordering on a fetal sample, please also order maternal cell contamination analysis separately (test code 0050608).
2004189	Noonan Syndrome (*PTPN11)* Sequencing with Reflex to *SOS1* Sequencing
0051805	Noonan Syndrome (*PTPN11*) Sequencing
2004195	Noonan Syndrome (*SOS1*) Sequencing
Targeted testing for known mutation (laboratory report from family member REQUIRED)	
2001961	Familial Mutation, Targeted Sequencing- targeted testing for a known familial sequence variant

Other test not listed: _____

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

Master Label