

SAMPLE REQUIREMENTS:

The preferred blood specimen is a 7 ml blood sample (3-5ml for infants) collected in a lavender top (K3EDTA) blood tube. Smaller blood samples or other tissue specimens may also be acceptable for certain tests. Please contact the laboratory for more details.

SHIPPING SAMPLES:

Each sample must be accompanied by a requisition form (available at <http://www.hpcgg.org/LMM/forms.html>). Please ensure that all samples are labeled with the patient's name and date of birth. All genetic tests require informed consent. Please have the patient (or parent) read and sign the consent form below.

The blood sample (with forms) should be shipped overnight at room temperature to:

Laboratory for Molecular Medicine
65 Landsdowne Street
Cambridge, MA 02139

For more detailed information about shipping procedures, see our website <http://www.hpcgg.org/lmm/>.

INFORMED CONSENT:

I understand that:

1. The purpose of this test is to determine if I/my child may have a mutation in the gene(s) being tested, which has been found to be associated with Noonan syndrome and related disorders. My healthcare provider should have provided me with information about the test(s) ordered and the condition(s) that may be identified. This information is also available on our website at <http://pcpgm.partners.org/LMM>
2. Genetic counseling is available to me and I have been provided with written information identifying genetic counselors, should I desire further information about this condition.
3. I have discussed with my genetic counselor that, except in the case of a known mutation test, a negative genetic test result does not rule out a diagnosis of, a predisposition towards, or the ability to pass on this condition, but does diminish the likelihood that this gene is involved.
4. I have discussed with my genetic counselor that this genetic test is specific for the indication for testing and does not test for other conditions. Therefore, a negative result does not guarantee my/my child's health.
5. In some families, genetic testing might discover non-paternity, or some other previously unknown information about family relationships, such as adoption.
6. The testing process includes highly skilled technicians and advanced technology. Although the method is extremely reliable, as in any laboratory, there is a small possibility that the test will not work properly, or an error may occur.
7. The lab will make every attempt to report results in the indicated turn-around time but cannot accept responsibility for delays.
8. If this test requisition form is incomplete, and my health care provider cannot provide the information, I understand that it may be necessary for lab staff to contact me directly to obtain or verify the information needed to complete the form.
9. I have been informed and agree that my ordering provider will receive the results of my genetic tests and that the ordering provider will disclose the results to me.
10. I understand that my test results as well as any updates to those results may become part of my permanent medical record electronically or otherwise or be made available, electronically or otherwise, to the ordering healthcare institution and its healthcare team.

I have carefully read and understand the above, have had any questions explained to my satisfaction, and do hereby consent to provide a specimen for testing.

Name of patient (please print)

(If applicable) Name of patient's legal representative (please print)

Signature (Patient or patient's legal representative) Date

Signature (Physician) Date

Please note: A physician may sign this form in lieu of the patient if prior consent has been obtained from the patient and if testing is for diagnostic purposes only.

Name: _____ DOB: ____ / ____ / ____ (MM/DD/YYYY)

GENE TEST TO BE PERFORMED:

NOONAN SPECTRUM DISORDERS (NOONAN, LEOPARD, CARDIO-FACIO-CUTANEOUS, and COSTELLO SYNDROMES)

 Noonan Spectrum Gene Chip (PTPN11, SOS1, RAF1, KRAS, NRAS, SHOC2, BRAF, MEK1, MEK2, HRAS)

Contact the laboratory for individual gene sequencing tests

FAMILIAL KNOWN MUTATION TEST

 Familial Mutation Test (Indicate gene, mutation, and information on proband (1st person tested) below)

Gene _____ Mutation _____ LMM Accession #: PM- _____

Proband Name _____ Relationship To Proband _____

PURPOSE OF STUDY

Clinical status: Symptomatic AsymptomaticPurpose of study: Diagnostic Carrier Screen Presymptomatic Prenatal Other _____Has another family member already had genetic testing for this disease? Yes No

If yes, please describe in the comments section and attach a copy of the genetic test lab report and pedigree.

Clinical diagnosis: Noonan LEOPARD **ICD-9 Codes:** 759.89 (Noonan/LEOPARD)

(check all known/suspected clinical diagnoses)

 CFC Costello 759.8 (Other specified abnormalities)Age at diagnosis: _____ Other _____Ultrasound Finding: Cystic hygroma Increased NT - Size: _____ None
 Heart defect - Type: _____ Other _____Congenital heart defect: Pulmonic valve stenosis Hypertrophic cardiomyopathy None
 Septal defect Other _____Facial dysmorphism: Epicanthal folds Ptosis of the eyelids Low nasal bridge
 Hypertelorism Downward eye slant Low set ears and posteriorly rotated
 Papillomatas Coarseness NoneShort stature: Yes - Height(%): _____ Parental Heights: _____ NoCognitive development: Learning disabilities Developmental delay Mental retardation NormalSkeletal: Pectus excavatum Pectus carinatum Scoliosis NormalGenitourinary: Cryptorchidism (undescended testes) Normal
 Kidney malformation If yes, please describe: _____Bleeding diathesis: Yes No If yes, please describe: _____Hair/Skin findings: Yes No If yes, please describe: _____Malignancy: Yes No If yes, please describe: _____

Other: _____

Previous Genetic Testing: No Yes Gene(s) _____

If variants detected, please elaborate: _____

Family history : Yes No (Sketch below or attach pedigree if appropriate)

Comments/Special Instructions:

Pedigree:

Standard Symbols

○ = Female = Male
◇ = Gender unknown ⊙ = Carrier
■ ● ◆ = Affected individualEthnicity Paternal side: _____
Maternal side: _____Consanguinity? Yes No