Complete and print.

MAYO CLINIC LABORATORIES

Noonan Spectrum Gene Testing Patient Information

N S G T P I S

Reset Form

Instructions: The accurate interpretation and reporting of the genetic results is contingent upon the reason for referral, clinical information, ethnic background, and family history. To help provide the best possible service, supply the information requested below and **send paperwork with the specimen, or return by fax to Mayo Clinic Laboratories, Attn: Personalized Genomics Laboratory Genetic Counselors at 507-284-1759. Phone: 507-266-5700 / International clients: +1-507-266-5700 or email mclglobal@mayo.edu**

Patient Information

Patient Name *(Last, First, Middle)*	Birth Date *(mm-dd-yyyy)*	Sex ☐ Male ☐ Female
Referring Provider Name *(Last, First)*	Phone	Fax*
Other Contact Name *(Last, First)*	Phone	Fax*

**Fax number given must be from a fax machine that complies with applicable HIPAA regulations.*

Is this a postmortem specimen? ☐ Yes ☐ No If yes, attach autopsy report if available.

Clinical History

Reason for Testing (check all that apply): ☐ Diagnosis ☐ Carrier testing ☐ Presymptomatic diagnosis ☐ Family history ☐ Sudden death

Note: Genetic testing should always be initiated on an affected family member first, if possible, in order to be most informative for at-risk relatives. See Ethnic Background and Family History section for more information.

Diagnosis/Suspected Diagnosis:

☐ Noonan syndrome ☐ Cardiofaciocutaneous (CFC) syndrome ☐ Costello syndrome ☐ Multiple Lentigines (LEOPARD) syndrome

☐ Other, specify: _____

Indicate whether the following are present.

Cardiovascular:

☐ Pulmonary valve stenosis ☐ Pulmonary artery stenosis ☐ Atrial septal defect ☐ Ventricular septal defect

☐ Hypertrophic cardiomyopathy ☐ Tetralogy of Fallot ☐ EKG abnormality ☐ Aortic coarctation

☐ Other, specify: _____

Skeletal:

☐ Short stature ☐ Pectus abnormality ☐ Scoliosis ☐ Cubitus valgus ☐ Vertebral anomalies

Facial dysmorphism:

☐ Characteristic Noonan facies (hypertelorism, epicanthal folds, ptosis, down-slanting palpebral fissures, triangular facies, low-set, posteriorly rotated ears, light-colored irises)

☐ Characteristic CFC syndrome/Costello facies (macrocephaly, coarse facial features including full lips, large mouth)

Developmental:

☐ Developmental delay ☐ Mental retardation ☐ Attention deficit/hyperactivity disorder

Cutaneous:

☐ Lentigines ☐ Café-au-lait spots ☐ Hyperkeratosis ☐ Ichthyosis ☐ Eczema ☐ Pigmented moles

☐ Hyperkeratosis ☐ Dystrophic nails ☐ Deep palmar and plantar creases

Hair abnormalities:

☐ Sparse ☐ Curly ☐ Fine ☐ Thick ☐ Woolly ☐ Brittle ☐ Absent eyebrows/eyelashes ☐ Loose anagen hair

Additional features:

☐ Hearing loss ☐ Broad or webbed neck with low posterior hairline ☐ Cryptorchidism ☐ Low-set nipples

☐ Feeding difficulties ☐ Postnatally reduced growth ☐ Coagulation defects ☐ Lymphatic dysplasia

☐ Malignancy/Tumor/Leukemia, specify: _____

List any additional features present.

Ethnic Background and Family History

☐ European Caucasian ☐ African American ☐ Hispanic ☐ Asian ☐ Middle Eastern ☐ Other, specify: _____

Are other relatives known to be affected? ☐ Yes ☐ No

If yes, indicate their diagnosis and relationship to the patient: _____

Have other relatives had molecular genetic testing? ☐ Yes ☐ No

For known mutation test requests, order known variant analysis:

KVAR1 / Known Variant Analysis-1 Variant, Varies; KVAR2 / Known Variant Analysis-2 Variants, Varies; KVAR3 / Known Variant Analysis-3+ Variants, Varies

New York State Patients: Informed Consent for Genetic Testing is required. See Informed Consent for Genetic Testing (T576), Informed Consent for Genetic Testing – Spanish (T826), or Informed Consent for Genetic Testing for Deceased Individuals (T782).

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