Complete and print.

MAYO CLINIC LABORATORIES

Noonan Spectrum Genetic Testing Patient Information

N S G T P I S

Reset Form

Instructions: Accurate interpretation and reporting of genetic results is contingent upon the reason for testing, clinical information, ethnic background/ancestry, 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: Molecular Technologies Laboratory Genetic Counselors at 507-284-1759. Phone: 800-533-1710 / International clients: 855-379-3115 or +1-507-284-9273, or email MLIINT@mayo.edu**

Patient Information

Patient Name (Last, First Middle)	Birth Date (mm-dd-yyyy)
Sex Assigned at Birth ☐ Male ☐ Female ☐ Unknown ☐ Choose not to disclose	Legal/Administrative Sex ☐ Male ☐ Female ☐ Nonbinary

Referring Healthcare Professional Information

Referring Healthcare Professional 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.*

Reason for Testing Check all that apply.

☐ Diagnosis ☐ Family history** ☐ Prenatal diagnosis

**Genetic testing should be performed on an affected family member first, when possible. FMTT / Familial Variant, Targeted Testing should be ordered when there is a previous positive genetic test result in the family.

Clinical History

Diagnosis/Suspected Diagnosis

☐ Baraitser-Winter syndrome ☐ Cardiofaciocutaneous (CFC) syndrome ☐ Costello syndrome ☐ Legius syndrome
☐ Multiple Lentigines (LEOPARD) syndrome ☐ Noonan 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 ☐ Intellectual disability ☐ Attention deficit/hyperactivity disorder

Cutaneous: ☐ Lentigines ☐ Café-au-lait spots ☐ Hyperkeratosis ☐ Ichthyosis ☐ Eczema
☐ Hyperkeratosis ☐ Dystrophic nails ☐ Deep palmar and plantar creases ☐ Pigmented moles

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

Additional features: ☐ Hearing loss ☐ Postnatally reduced growth ☐ Low-set nipples
☐ Feeding difficulties ☐ Cryptorchidism ☐ Lymphatic dysplasia
☐ Broad or webbed neck with low posterior hairline ☐ Coagulation defects
☐ Malignancy/Tumor/Leukemia; specify:

Note: Skin biopsy is the preferred specimen type to detect germline variants in patients with active hematological malignancy.

Family History

Are there similarly affected relatives? ☐ Yes ☐ No
If "Yes," indicate relationship and symptoms:

Have any family member had genetic testing? ☐ Yes*** ☐ No ☐ Unknown

***FMTT / Familial Variant, Targeted Testing should be ordered when there is a previous positive genetic test result in the family. Contact the lab for ordering assistance.**

History of consanguinity: ☐ No ☐ Yes; relationship details:

Ancestry

☐ African/African American ☐ East Asian ☐ Latinx/Latine ☐ South Asian ☐ Unknown
☐ Ashkenazi Jewish ☐ European ☐ Middle Eastern ☐ None of the above ☐ Choose not to disclose

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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