

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: +1-507-266-5700 or email MLIINT@mayo.edu**

Patient Information

| | | |
|---|--|-------------------------|
| Patient Name (Last, First Middle) | | Birth Date (mm-dd-yyyy) |
| Sex Assigned at Birth <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Unknown <input type="checkbox"/> Choose not to disclose | Legal/Administrative Sex <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Nonbinary | |

Referring Provider Information

| | | |
|---------------------------------------|-------|------|
| 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.

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 Mental retardation 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).