MAYO CLINIC
Mayo Medical Laboratories

Noonan Spectrum Gene Testing Patient Information Sheet

N S G T P I S

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 this paperwork with the specimen.**

Patient Information

Patient Name *(Last, First, Middle)*	Birth Date *(Month DD, YYYY)*	Sex ☐ Male ☐ Female
Referring Physician Name	Phone	Fax
Other Contact	Phone	Fax

Clinical History

Patient 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

☐ 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)* _____

Indicate any additional features present

Ethnic Background and Family History

☐ European Caucasian ☐ African American ☐ Hispanic ☐ Asian ☐ Other *(specify)* _____

Attach pedigree if available

Are other relatives known to be affected? ☐ Yes ☐ No If Yes, indicate their relationship to the patient _____

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

For Known Mutation testing, a familial mutation MUST be provided

Indicate: Gene _____ Exon _____ Amino Acid _____ Nucleotide _____ OR Intron _____ Nucleotide _____

Performing laboratory for relative's testing _____

If relative was tested at Mayo Clinic, name and relationship of relative _____

Attach a copy of the genetic test lab report if available

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