



OHSU Clinical Genetics Laboratories
Shipping Address: 2525 SW 3rd Avenue, Suite 350
Portland, OR 97201

Phone: (503) 494-5400
Fax: (503) 494-6922

Biochemical Genetics Laboratory

Cytogenetics Laboratory

Molecular Diagnostic Center

NOONAN SYNDROME

Molecular Diagnostic Center

Noonan syndrome (NS) is an autosomal dominant disorder characterized by cardiac defects (including pulmonary stenosis, hypertrophic cardiomyopathy and septal defects), facial dysmorphism (low-set, posteriorly rotated ears, hypertelorism and downward slanting palpebral fissures), short stature, developmental delay, and bleeding diatheses. NS has been shown to be caused by mutations in *PTPN11* in about 50% of cases. *PTPN11* encodes the non-receptor-type protein tyrosine phosphatase SHP-2 on chromosome 12q24.1. Incidence of NS is estimated to be 1:1000 to 1:2500 live births.

REASONS FOR REFERRAL

- Confirmation of clinical diagnosis in patients with Noonan syndrome
- Prenatal diagnosis

TESTING METHODOLOGY

Mutation scanning: Our laboratory uses PCR followed by denaturing high performance liquid chromatography (dHPLC) to screen the entire coding region of the *PTPN11* gene in two steps for heteroduplex DNA, indicating a DNA sequence change. Samples containing a change are directly sequenced to determine the presence of a disease-causing mutation or a single nucleotide polymorphism.

Test Step 1 analyzes Exons 3, 7, 8, 9, 12, and 13

Test Step 2 analyzes Exons 1, 2, 4, 5, 6, 10, 11, 14, and 15

Mutation Analysis

Step 1 will detect 90% of mutations in *PTPN11*

Step 2 will detect all remaining (10%) mutations in *PTPN11*

Test reporting follows the ACMG guidelines.

SPECIMEN REQUIREMENTS

Blood: EDTA (purple-top) tubes:

Adult: 5 mL

Child: 5 mL

Infant: 2-3 mL

Requisition form must accompany specimen including ethnicity, clinical and family history information. Prior to any genetic testing we recommend genetic counseling. To receive forms and information about prenatal diagnostic testing, please contact our laboratory.

TURNAROUND TIME: 3 weeks (for each step of the test)

CPT CODES

Step 1: 83891, 83898 x 5, 83903 x 5, 83904, 83912

Step 2: 83898 x 9, 83903 x 9