

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** NOONAN Syndrome
- **Test Code:** 4825

Related Diseases & Genes:

- Noonan syndrome 1 (OMIM #: 163950) || Gene: *PTPN11* (OMIM #: 176876)

Synonyms:

- NOONAN SYNDROME 1; NS1
- MALE TURNER SYNDROME
- FEMALE PSEUDO-TURNER SYNDROME
- TURNER PHENOTYPE WITH NORMAL KARYOTYPE
- PTPN11 Mutations

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 7-10 days

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Dysmorphology

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