

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** PURINE NUCLEOSIDE PHOSPHORYLASE DEFICIENCY
- **Test Code:** 2174

Related Diseases & Genes:

- Immunodeficiency due to purine nucleoside phosphorylase deficiency (OMIM #: 613179) || Gene: PNP (OMIM #: 164050)

Synonyms:

- PNP Sequence Analysis
- NUCLEOSIDE PHOSPHORYLASE; NP
- PURINE-NUCLEOSIDE:ORTHOPHOSPHATE RIBOSYLTRANSFERASE

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):

- Dsymorphology

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