

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** NIJMEGEN SYNDROME
- **Test Code:** 2425

Related Diseases & Genes:

- Nijmegen breakage syndrome (OMIM #: 251260) || Gene: *NBS1* (OMIM #: 602667)

Synonyms:

- "NIJMEGEN BREAKAGE SYNDROME, NBS"
- 657del5 Mutation
- SEEMANOVA SYNDROME II
- "NONSYNDROMAL MICROCEPHALY, AUTOSOMAL RECESSIVE, WITH NORMAL INTELLIGENCE"
- "IMMUNODEFICIENCY, MICROCEPHALY, AND CHROMOSOMAL INSTABILITY"
- ATAXIA-TELANGIECTASIA VARIANT V1; AT-V1

Test Details:

- **Methodology:** PCR
- **Performed:** Everyday
- **Reported:** 2 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Dsymorphology

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