

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** Congenital Neutropenia (*ELANE* Gene)
- **Test Code:** 3634

Related Diseases & Genes:

- “Neutropenia, severe congenital 1, autosomal dominant” (OMIM #: 202700) || Gene: *ELANE* (OMIM #: 130130)
- “Neutropenia, cyclic” (OMIM #: 162800)

Synonyms:

- Elastase, Neutrophil-Expressed
- Elastase 2 (ELA2)

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 3 weeks

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):

- Hematogenetics

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For yesterday, today and *tomorrows...*

