



ELANE gene

elastase, neutrophil expressed

Normal Function

The *ELANE* gene provides instructions for making a protein called neutrophil elastase. This protein is found in neutrophils, a type of white blood cell that plays a role in inflammation and in fighting infection. When the body starts an immune response to fight an infection, neutrophils release neutrophil elastase. This protein then modifies the function of certain cells and proteins to fight the infection.

Health Conditions Related to Genetic Changes

Cyclic neutropenia

More than 15 mutations in the *ELANE* gene have been found to cause cyclic neutropenia, a condition characterized by episodes of neutrophil shortages (neutropenia) and increased risk of infection. *ELANE* gene mutations that cause cyclic neutropenia change single protein building blocks (amino acids) in neutrophil elastase. These mutations are thought to create an abnormal protein that retains some function. However, neutrophils that produce abnormal neutrophil elastase proteins appear to have a shorter lifespan than normal. The shorter neutrophil lifespan is thought to be responsible for the cyclic nature of this condition. When the affected neutrophils die early, there is a period in which there is a shortage of neutrophils because it takes time for the body to replenish its supply. For most affected individuals, neutropenia recurs every 21 days.

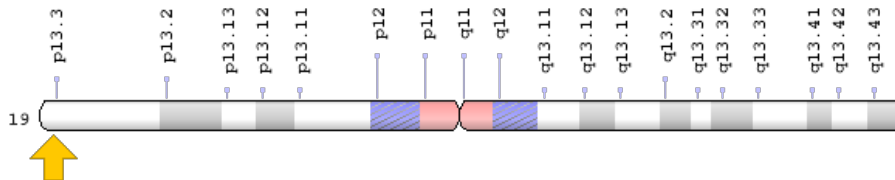
Severe congenital neutropenia

More than 70 mutations in the *ELANE* gene have been found to cause severe congenital neutropenia, a condition characterized by neutropenia beginning in infancy. Most of these mutations alter the structure of neutrophil elastase, causing it to fold into an incorrect 3-dimensional shape. Research findings indicate that misfolded neutrophil elastase protein accumulates in neutrophils. This accumulation likely damages and kills these infection-fighting cells. A deficiency of neutrophils results in recurrent infections, episodes of inflammation, and other immune problems in people with severe congenital neutropenia.

Chromosomal Location

Cytogenetic Location: 19p13.3, which is the short (p) arm of chromosome 19 at position 13.3

Molecular Location: base pairs 850,997 to 856,250 on chromosome 19 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- bone marrow serine protease
- ELA2
- elastase-2
- ELNE_HUMAN
- granulocyte-derived elastase
- NE
- neutrophil elastase

Additional Information & Resources

Educational Resources

- Cancer Medicine (sixth edition, 2003): Neutropenia
<https://www.ncbi.nlm.nih.gov/books/NBK13000/#A43604>
- Immunobiology (fifth edition, 2001): Neutrophils make up the first wave of cells that cross the blood vessel wall to enter inflammatory sites
<https://www.ncbi.nlm.nih.gov/books/NBK27122/#A213>

Clinical Information from GeneReviews

- ELANE-Related Neutropenia
<https://www.ncbi.nlm.nih.gov/books/NBK1533>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ELANE%5BTIAB%5D%29+OR+%28%28ELA2%5BTIAB%5D%29+OR+%28neutrophil+elastase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- ELASTASE, NEUTROPHIL-EXPRESSED
<http://omim.org/entry/130130>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ELANE.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ELANE%5Bgene%5D>
- ELA2base: Mutation Registry for Cyclic Neutropenia; Congenital Neutropenia
<http://structure.bmc.lu.se/idbase/ELA2base/index.php>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3309
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:1991>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1991>
- UniProt
<https://www.uniprot.org/uniprot/P08246>

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