

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.

Other Names for This Gene

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ELANE ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1991\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1991[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%29>)

Catalog of Genes and Diseases from OMIM

- ELASTASE, NEUTROPHIL-EXPRESSED; ELANE (<https://omim.org/entry/130130>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1991>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ELANE\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ELANE[gene]))

References

- Bellanne-Chantelot C, Clauin S, Leblanc T, Cassinat B, Rodrigues-Lima F, Beaufils S, Vaury C, Barkaoui M, Fenneteau O, Maier-Redelsperger M, Chomienne C, Donadieu J. Mutations in the ELA2 gene correlate with more severe expression of neutropenia: a study of 81 patients from the French Neutropenia Register. *Blood*. 2004 Jun 1;103(11):4119-25. doi: 10.1182/blood-2003-10-3518. Epub 2004 Feb 12. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14962902>)
- Boztug K, Klein C. Novel genetic etiologies of severe congenital neutropenia. *Curr*

Opin Immunol. 2009 Oct;21(5):472-80. doi: 10.1016/j.coi.2009.09.003. Epub 2009 Sep 24. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19782549>)

- Grenda DS, Murakami M, Ghatak J, Xia J, Boxer LA, Dale D, Dinanuer MC, Link DC. Mutations of the ELA2 gene found in patients with severe congenital neutropenia induce the unfolded protein response and cellular apoptosis. *Blood*. 2007 Dec 15;110(13):4179-87. doi: 10.1182/blood-2006-11-057299. Epub 2007 Aug 30. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17761833>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2234798/>)
- Horwitz MS, Duan Z, Korkmaz B, Lee HH, Mealiffe ME, Salipante SJ. Neutrophil elastase in cyclic and severe congenital neutropenia. *Blood*. 2007 Mar 1;109(5):1817-24. doi: 10.1182/blood-2006-08-019166. Epub 2006 Oct 19. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17053055>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1801070/>)
- Rosenberg PS, Alter BP, Link DC, Stein S, Rodger E, Bolyard AA, Aprikyan AA, Bonilla MA, Dror Y, Kannourakis G, Newburger PE, Boxer LA, Dale DC. Neutrophil elastase mutations and risk of leukaemia in severe congenital neutropenia. *Br J Haematol*. 2008 Jan;140(2):210-3. doi: 10.1111/j.1365-2141.2007.06897.x. Epub 2007 Nov 20. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18028488>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3143022/>)
- Schaffer AA, Klein C. Genetic heterogeneity in severe congenital neutropenia: how many aberrant pathways can kill a neutrophil? *Curr Opin Allergy Clin Immunol*. 2007 Dec;7(6):481-94. doi: 10.1097/ACI.0b013e3282f1d690. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17989524>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2720578/>)
- Xia J, Bolyard AA, Rodger E, Stein S, Aprikyan AA, Dale DC, Link DC. Prevalence of mutations in ELANE, GFI1, HAX1, SBDS, WAS and G6PC3 in patients with severe congenital neutropenia. *Br J Haematol*. 2009 Nov;147(4):535-42. doi: 10.1111/j.1365-2141.2009.07888.x. Epub 2009 Sep 22. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19775295>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2783282/>)
- Zeidler C, Germeshausen M, Klein C, Welte K. Clinical implications of ELA2-, HAX1-, and G-CSF-receptor (CSF3R) mutations in severe congenital neutropenia. *Br J Haematol*. 2009 Feb;144(4):459-67. doi: 10.1111/j.1365-2141.2008.07425.x. Epub 2008 Dec 10. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19120359>)

Genomic Location

The *ELANE* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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