



## ELN gene

elastin

### Normal Function

The *ELN* gene provides instructions for making a protein called tropoelastin. Multiple copies of the tropoelastin protein attach to one another and are processed to form a mature protein called elastin. Elastin is the major component of elastic fibers, which are slender bundles of proteins that provide strength and flexibility to connective tissue (tissue that supports the body's joints and organs). Elastic fibers are found in the intricate lattice that forms in the spaces between cells (the extracellular matrix), where they give structural support to organs and tissues such as the heart, skin, lungs, ligaments, and blood vessels.

### Health Conditions Related to Genetic Changes

#### Cutis laxa

At least 16 mutations in the *ELN* gene have been identified in people with a skin disorder called cutis laxa. *ELN* gene mutations cause a form of the disorder called autosomal dominant cutis laxa, which is characterized by loose, sagging skin; an increased risk of an abnormal bulging (an aneurysm) in a large blood vessel called the aorta; and a lung disease called emphysema, which can make it difficult to breathe.

The *ELN* mutations that cause autosomal dominant cutis laxa lead to the production of an abnormally long version of the tropoelastin protein. The abnormal protein likely interferes with the formation of mature elastin and the assembly of elastic fibers, which weakens connective tissue in the skin and blood vessels. This defect in connective tissue underlies the major features of cutis laxa.

#### Supravalvular aortic stenosis

More than 60 mutations in the *ELN* gene have been found to cause supravalvular aortic stenosis (SVAS), a heart defect present from birth that is characterized by a narrowing of the large blood vessel that carries blood from the heart to the rest of the body (the aorta). Most of the *ELN* gene mutations that cause SVAS lead to a decrease in the production of tropoelastin. A shortage of tropoelastin reduces the amount of mature elastin protein that is processed and available for forming elastic fibers. As a result, elastic fibers that make up the aorta are thinner than normal. To compensate, the smooth muscle cells that line the aorta increase in number, making the aorta thicker and narrower than usual. A thickened aorta is less flexible and resilient to the stress of constant blood flow and pumping of the heart. Over time, the wall of the aorta can become damaged. Aortic narrowing causes the heart to work

harder to pump blood through the aorta, which can lead to shortness of breath, chest pain, and ultimately heart failure.

#### 7q11.23 duplication syndrome

The *ELN* gene is located in a region of chromosome 7 that is duplicated in people with 7q11.23 duplication syndrome. As a result of this duplication, people with 7q11.23 duplication syndrome have an extra copy of the *ELN* gene and several other genes in each cell. 7q11.23 duplication syndrome can cause a variety of neurological and behavioral problems as well as other abnormalities.

About half of individuals with 7q11.23 duplication syndrome have enlargement (dilatation) of the aorta; this enlargement can get worse over time. Aortic dilatation can lead to life-threatening complications if the wall of the aorta separates into layers (aortic dissection) or breaks open (ruptures). An extra copy of the *ELN* gene in each cell may lead to the production of a greater than normal amount of tropoelastin, and researchers suggest that this excess might be related to the increased risk for aortic dilatation in 7q11.23 duplication syndrome; however, the specific cause of the aortic dilatation remains unclear.

#### Williams syndrome

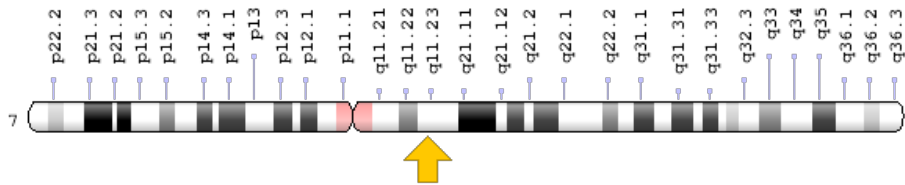
The *ELN* gene is located in a region of chromosome 7 that is deleted in people with Williams syndrome. As a result of this deletion, people with Williams syndrome are missing one copy of the *ELN* gene in each cell. This loss reduces the production of elastin by half, which disrupts the normal structure of elastic fibers in many connective tissues. Large blood vessels with abnormal elastic fibers are often thicker and less resilient than normal. These vessels can narrow, increasing the resistance to normal blood flow and leading to serious medical problems.

In people with Williams syndrome, a loss of the *ELN* gene is associated with connective tissue abnormalities, such as joint problems and loose skin, and cardiovascular disease, particularly SVAS.

## Chromosomal Location

Cytogenetic Location: 7q11.23, which is the long (q) arm of chromosome 7 at position 11.23

Molecular Location: base pairs 74,027,772 to 74,069,907 on chromosome 7 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- elastin (supravalvular aortic stenosis, Williams-Beuren syndrome)
- ELN\_HUMAN
- tropoelastin

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Elastin Gives Tissues Their Elasticity  
<https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3568>

### GeneReviews

- 7q11.23 Duplication Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK327268>
- Williams Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1249>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ELN%5BTIAB%5D%29+OR+%28elastin%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+720+days%22%5Bdp%5D>

## OMIM

- ELASTIN  
<http://omim.org/entry/130160>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ELN.html](http://atlasgeneticsoncology.org/Genes/GC_ELN.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ELN%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=3327](https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3327)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2006>
- UniProt  
<http://www.uniprot.org/uniprot/P15502>

## **Sources for This Summary**

- Brooke BS, Bayes-Genis A, Li DY. New insights into elastin and vascular disease. Trends Cardiovasc Med. 2003 Jul;13(5):176-81. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12837579>
- Callewaert B, Renard M, Huchtagowder V, Albrecht B, Hausser I, Blair E, Dias C, Albino A, Wachi H, Sato F, Mecham RP, Loeys B, Coucke PJ, De Paepe A, Urban Z. New insights into the pathogenesis of autosomal-dominant cutis laxa with report of five ELN mutations. Hum Mutat. 2011 Apr;32(4):445-55. doi: 10.1002/humu.21462. Epub 2011 Mar 1.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21309044>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3383654/>
- Graul-Neumann LM, Hausser I, Essayie M, Rauch A, Kraus C. Highly variable cutis laxa resulting from a dominant splicing mutation of the elastin gene. Am J Med Genet A. 2008 Apr 15;146A(8):977-83. doi: 10.1002/ajmg.a.32242.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18348261>
- Metcalfe K, Rucka AK, Smoot L, Hofstadler G, Tuzler G, McKeown P, Siu V, Rauch A, Dean J, Dennis N, Ellis I, Reardon W, Cytrynbaum C, Osborne L, Yates JR, Read AP, Donnai D, Tassabehji M. Elastin: mutational spectrum in supra-aortic stenosis. Eur J Hum Genet. 2000 Dec;8(12):955-63.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/11175284>
- Milewicz DM, Urbán Z, Boyd C. Genetic disorders of the elastic fiber system. Matrix Biol. 2000 Nov;19(6):471-80. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/11068201>
- Morris CA, Mervis CB, Paciorkowski AP, Abdul-Rahman O, Dugan SL, Rope AF, Bader P, Hendon LG, Velleman SL, Klein-Tasman BP, Osborne LR. 7q11.23 Duplication syndrome: Physical characteristics and natural history. Am J Med Genet A. 2015 Dec;167A(12):2916-35. doi: 10.1002/ajmg.a.37340. Epub 2015 Sep 3.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26333794>

- Morris CA, Mervis CB. Williams syndrome and related disorders. *Annu Rev Genomics Hum Genet.* 2000;1:461-84. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/11701637>
- Morris CA. Williams Syndrome. 1999 Apr 9 [updated 2017 Mar 23]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. *GeneReviews®* [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1249/>  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20301427>
- Park S, Seo EJ, Yoo HW, Kim Y. Novel mutations in the human elastin gene (ELN) causing isolated supravalvular aortic stenosis. *Int J Mol Med.* 2006 Aug;18(2):329-32.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16820942>
- Parrott A, James J, Goldenberg P, Hinton RB, Miller E, Shikany A, Aylsworth AS, Kaiser-Rogers K, Ferns SJ, Lalani SR, Ware SM. Aortopathy in the 7q11.23 microduplication syndrome. *Am J Med Genet A.* 2015 Feb;167A(2):363-70. doi: 10.1002/ajmg.a.36859. Epub 2014 Nov 26.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25428557>
- Rodriguez-Revenga L, Badenas C, Carrió A, Milà M. Elastin mutation screening in a group of patients affected by vascular abnormalities. *Pediatr Cardiol.* 2005 Nov-Dec;26(6):827-31.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15990952>
- Rodriguez-Revenga L, Iranzo P, Badenas C, Puig S, Carrió A, Milà M. A novel elastin gene mutation resulting in an autosomal dominant form of cutis laxa. *Arch Dermatol.* 2004 Sep;140(9):1135-9. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15381555>
- Szabo Z, Crepeau MW, Mitchell AL, Stephan MJ, Puntel RA, Yin Loke K, Kirk RC, Urban Z. Aortic aneurysmal disease and cutis laxa caused by defects in the elastin gene. *J Med Genet.* 2006 Mar;43(3):255-8. Epub 2005 Aug 5.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16085695>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2563239/>
- Tassabehji M. Williams-Beuren syndrome: a challenge for genotype-phenotype correlations. *Hum Mol Genet.* 2003 Oct 15;12 Spec No 2:R229-37. Epub 2003 Sep 2. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12952863>
- Urbán Z, Riazi S, Seidl TL, Katahira J, Smoot LB, Chitayat D, Boyd CD, Hinek A. Connection between elastin haploinsufficiency and increased cell proliferation in patients with supravalvular aortic stenosis and Williams-Beuren syndrome. *Am J Hum Genet.* 2002 Jul;71(1):30-44. Epub 2002 May 6.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12016585>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC384991/>
- Urbán Z, Zhang J, Davis EC, Maeda GK, Kumar A, Stalker H, Belmont JW, Boyd CD, Wallace MR. Supravalvular aortic stenosis: genetic and molecular dissection of a complex mutation in the elastin gene. *Hum Genet.* 2001 Nov;109(5):512-20. Epub 2001 Oct 13.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/11735026>

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Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/gene/ELN>

Reviewed: May 2012

Published: April 17, 2018

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services