

## 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 a major component of the tissue that supports the body's joints and organs (connective tissue). Elastic fibers are found in the intricate lattice that forms in the spaces between cells (the extracellular matrix). They can be stretched and then snap back into place, which is how they provide resilience and flexibility to organs and tissues such as the heart, skin, lungs, ligaments, and blood vessels.

### Health Conditions Related to Genetic Changes

#### Cutis laxa

At least 18 variants (also known as mutations) in the *ELN* gene have been identified in people with a skin disorder called cutis laxa. *ELN* gene variants cause a form of the condition called autosomal dominant cutis laxa type 1 (ADCL1), 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* gene variants that cause ADCL1 lead to the production of an abnormal version of the tropoelastin protein. The abnormal protein is less able to be incorporated into mature elastin, which impairs the formation of strong elastic fibers. This defect in elastic fibers weakens connective tissue in the skin and blood vessels and makes it difficult for tissue to snap back into shape when stretched. The problems with connective tissue underlie the major features of cutis laxa.

#### Supravalvular aortic stenosis

More than 70 variants in the *ELN* gene have been found to cause supravalvular aortic stenosis (SVAS), a blood vessel defect most commonly identified in infancy or early childhood 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 variants 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 delivers blood too forcefully, which can damage organs. 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.

Some individuals with 7q11.23 duplication syndrome have mild to moderate enlargement (dilatation) of the aorta; this enlargement can get worse over time. Aortic dilatation in people with this condition can very rarely lead to life-threatening complications if the wall of the aorta separates into layers (aortic dissection) or breaks open (ruptures). Individuals with 7q11.23 duplication may have other heart or blood vessel abnormalities. 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, which is a developmental disorder that is characterized by mild to moderate intellectual disability or learning problems, unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems. As a result of the deletion, people with Williams syndrome are missing one copy of the *ELN* gene in each cell. This loss reduces the production of elastin by approximately half, which disrupts the normal structure of elastic fibers in many connective tissues. As a result, large blood vessels such as the aorta are often thicker and less resilient than normal. These vessels can narrow (as in SVAS, described above), increasing the resistance to normal blood flow and leading to serious medical problems.

In addition to cardiovascular problems like SVAS, a loss of the *ELN* gene is associated with other connective tissue abnormalities, such as joint problems, soft skin, and mild lung problems. Loss of the *ELN* gene may also play a role in other features of the condition, such as premature skin wrinkling and abnormalities of the digestive tract.

### **Other Names for This Gene**

- elastin

- ELN\_HUMAN
- tropoelastin

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of ELN ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2006\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2006[geneid]))

### Scientific Articles on PubMed

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

### Catalog of Genes and Diseases from OMIM

- ELASTIN; ELN (<https://omim.org/entry/130160>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2006>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ELN\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ELN[gene]))

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## Genomic Location

The *ELN* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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