



## Isolated ectopia lentis

Isolated ectopia lentis is a condition that affects the eyes, specifically the positioning of the lens. The lens is a clear structure at the front of the eye that helps focus light. In people with isolated ectopia lentis, the lens in one or both eyes is not centrally positioned as it should be but is off-center (displaced). Isolated ectopia lentis usually becomes apparent in childhood. The lens may drift further off-center over time.

Vision problems are common in isolated ectopia lentis. Affected individuals often have nearsightedness (myopia) and can have an irregular curvature of the lens or a structure that covers the front of the eye (the cornea), which causes blurred vision (astigmatism). They may also develop clouding of the lenses (cataracts) or increased pressure in the eyes (glaucoma) at an earlier age than other adults. In a small number of people with isolated ectopia lentis, tearing of the back lining of the eye (retinal detachment) occurs, which can lead to further vision problems and possible blindness.

In individuals with isolated ectopia lentis, each eye can be affected differently. In addition, the eye problems vary among affected individuals, even those within the same family.

Ectopia lentis is classified as isolated when it occurs alone without signs and symptoms affecting other body systems. Ectopia lentis can also be classified as syndromic, when it is part of a syndrome that affects multiple parts of the body. Ectopia lentis is a common feature of genetic syndromes such as Marfan syndrome and Weill-Marchesani syndrome.

### Frequency

The prevalence of isolated ectopia lentis is unknown. In Denmark, an estimated 6.4 per 100,000 individuals have ectopia lentis, but a large proportion of these cases (about 75 percent) are syndromic.

### Causes

Mutations in the *FBN1* or *ADAMTSL4* gene cause isolated ectopia lentis. These genes provide instructions for making proteins that are necessary for the formation of threadlike filaments called microfibrils. Microfibrils provide support to many tissues, including the lenses of the eyes, which are held in position by these filaments.

Mutations in the *FBN1* or *ADAMTSL4* gene impair protein function and lead to a decrease in microfibril formation or result in the formation of impaired microfibrils. Without functional microfibrils to anchor the lens in its central position at the front of the eye, the lens becomes displaced. The displaced lens cannot focus light correctly,

contributing to the vision problems that are common in people with isolated ectopia lentis.

## **Inheritance Pattern**

When isolated ectopia lentis is caused by mutations in the *FBN1* gene, it is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In some cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

When isolated ectopia lentis is caused by mutations in the *ADAMTSL4* gene, it is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## **Other Names for This Condition**

- congenital ectopia lentis
- ectopia lentis
- lens subluxation
- subluxation of lens

## **Diagnosis & Management**

### Genetic Testing Information

- What is genetic testing?  
[/primer/testing/geneticTesting](#)
- Genetic Testing Registry: Ectopia lentis, isolated autosomal recessive  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3541474/>
- Genetic Testing Registry: Ectopia lentis, isolated, autosomal dominant  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3541518/>

### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22isolated+ectopia+lentis%22+OR+%22Ectopia+Lentis%22+OR+%22Lens+Subluxation%22>

### Other Diagnosis and Management Resources

- GeneReview: ADAMTSL4-Related Eye Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK84111>

## **Additional Information & Resources**

### Health Information from MedlinePlus

- Encyclopedia: Cataract (image)  
<https://medlineplus.gov/ency/imagepages/19587.htm>
- Health Topic: Cataract  
<https://medlineplus.gov/cataract.html>
- Health Topic: Eye Diseases  
<https://medlineplus.gov/eyediseases.html>
- Health Topic: Glaucoma  
<https://medlineplus.gov/glaucoma.html>
- Health Topic: Refractive Errors  
<https://medlineplus.gov/refractiveerrors.html>

### Genetic and Rare Diseases Information Center

- Isolated ectopia lentis  
<https://rarediseases.info.nih.gov/diseases/12251/isolated-ectopia-lentis>

### Additional NIH Resources

- National Eye Institute: Diagram of the Eye  
<https://nei.nih.gov/health/eyediagram>
- National Eye Institute: Facts About Astigmatism  
<https://nei.nih.gov/health/errors/astigmatism>

### Educational Resources

- MalaCards: isolated ectopia lentis  
[https://www.malacards.org/card/isolated\\_ectopia\\_lentis](https://www.malacards.org/card/isolated_ectopia_lentis)
- Merck Manual Consumer Version: Structure and Function of the Eyes  
<https://www.merckmanuals.com/home/eye-disorders/biology-of-the-eyes/structure-and-function-of-the-eyes>
- Orphanet: Isolated ectopia lentis  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=1885](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1885)
- The University of Arizona Health Sciences: Hereditary Ocular Disease: Ectopia Lentis, Isolated AR  
<https://disorders.eyes.arizona.edu/disorders/ectopia-lentis-isolated-ar>
- The University of Arizona: Hereditary Ocular Disease: Ectopia Lentis, Isolated AD  
<https://disorders.eyes.arizona.edu/disorders/ectopia-lentis-isolated-ad>

## Patient Support and Advocacy Resources

- The Marfan Foundation  
<https://www.marfan.org/ectopia-lentis-syndrome>
- The Pediatric Glaucoma & Cataract Family Association  
<http://pgcfa.org/>

## Clinical Information from GeneReviews

- ADAMTSL4-Related Eye Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK84111>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28isolated+ectopia+lentis%5BTIAB%5D%29+OR+%28congenital+ectopia+lentis%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## Catalog of Genes and Diseases from OMIM

- ECTOPIA LENTIS 1, ISOLATED, AUTOSOMAL DOMINANT  
<http://omim.org/entry/129600>
- ECTOPIA LENTIS 2, ISOLATED, AUTOSOMAL RECESSIVE  
<http://omim.org/entry/225100>

## **Sources for This Summary**

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