

## Fuchs endothelial dystrophy

### Description

Fuchs endothelial dystrophy is a condition that causes vision problems. The first symptom of this condition is typically blurred vision in the morning that usually clears during the day. Over time, affected individuals lose the ability to see details (visual acuity). People with Fuchs endothelial dystrophy also become sensitive to bright lights.

Fuchs endothelial dystrophy specifically affects the front surface of the eye called the cornea. Deposits called guttae, which are detectable during an eye exam, form in the middle of the cornea and eventually spread throughout the cornea. These guttae contribute to the ongoing cell death within the cornea, leading to worsening vision problems. Tiny blisters may develop on the cornea, which can burst and cause eye pain.

The signs and symptoms of Fuchs endothelial dystrophy usually begin in a person's forties or fifties. A very rare early-onset variant of this condition starts to affect vision in a person's twenties.

### Frequency

The late-onset form of Fuchs endothelial dystrophy is a common condition, affecting approximately 4 percent of people over the age of 40 in the United States. The early-onset variant of Fuchs endothelial dystrophy is rare, although the exact prevalence is unknown.

For reasons that are unclear, Fuchs endothelial dystrophy affects women two to four times more frequently than men.

### Causes

The genetics of Fuchs endothelial dystrophy are unclear. Researchers have identified several genes and regions within a few chromosomes that they think may play a role in the development of Fuchs endothelial dystrophy. However, many of these genetic associations have been found in only a few affected individuals or families and it is unclear what role they have in the development of the condition.

Fuchs endothelial dystrophy affects a thin layer of cells that line the back of the cornea, called corneal endothelial cells. These cells regulate the amount of fluid inside the

cornea. An appropriate fluid balance in the cornea is necessary for clear vision. Fuchs endothelial dystrophy occurs when the endothelial cells die and the cornea becomes swollen with too much fluid. Corneal endothelial cells continue to die over time, resulting in worsening vision problems. It is thought that mutations in genes that are active (expressed) primarily in corneal endothelial cells or surrounding tissue contribute to the death of corneal endothelial cells, resulting in Fuchs endothelial dystrophy.

Some cases of the early-onset variant of Fuchs endothelial dystrophy are caused by mutations in a gene called *COL8A2*. This gene provides instructions for making a protein that is part of type VIII collagen. Type VIII collagen is largely found within the cornea, surrounding the endothelial cells. Specifically, type VIII collagen is a major component of a tissue at the back of the cornea, called Descemet's membrane. This membrane is a thin, sheet-like structure that separates and supports corneal endothelial cells. *COL8A2* gene mutations that cause the early-onset variant of Fuchs endothelial dystrophy lead to an abnormal Descemet's membrane, which causes corneal endothelial cells to die and leads to the vision problems in people with this condition.

Mutations in additional, unidentified genes are also thought to be involved in the development of both the early-onset variant of Fuchs endothelial dystrophy and the later onset form of the disorder.

[Learn more about the genes associated with Fuchs endothelial dystrophy](#)

- COL8A2
- TCF4

#### **Additional Information from NCBI Gene:**

- AGBL1
- ATP1B1
- KANK4
- LAMC1
- SLC4A11
- ZEB1

#### **Inheritance**

In many cases, the inheritance pattern of Fuchs endothelial dystrophy is unknown.

In some families, Fuchs endothelial dystrophy appears to be inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. When this condition is caused by a mutation in the *COL8A2* gene, it is inherited in an autosomal dominant pattern. In addition, an autosomal dominant inheritance pattern is apparent in some situations in which the condition is caused by alterations in an unknown gene.

Some cases result from new mutations in a gene and occur in people with no history of the disorder in their family.

## **Other Names for This Condition**

- Fuchs atrophy
- Fuchs corneal dystrophy
- Fuchs dystrophy
- Fuchs endothelial corneal dystrophy
- Fuchs' endothelial dystrophy

## **Additional Information & Resources**

### Genetic Testing Information

- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 3 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750451/>)
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 4 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750450/>)
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 6 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750448/>)
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 8 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3809798/>)
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 1 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850959/>)

### Genetic and Rare Diseases Information Center

- Fuchs endothelial corneal dystrophy (<https://rarediseases.info.nih.gov/diseases/10018/index>)

### Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

### Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Fuchs endothelial dystrophy%22](https://clinicaltrials.gov/search?cond=%22Fuchs%20endothelial%20dystrophy%22))

### Catalog of Genes and Diseases from OMIM

- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 1; FECD1 (<https://omim.org/entry/136800>)
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 2; FECD2 (<https://omim.org/entry/610158>)
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 3; FECD3 (<https://omim.org/entry/613267>)
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 4; FECD4 (<https://omim.org/entry/613268>)
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 5; FECD5 (<https://omim.org/entry/613269>)
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 6; FECD6 (<https://omim.org/entry/613270>)
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 7; FECD7 (<https://omim.org/entry/613271>)
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 8; FECD8 (<https://omim.org/entry/615523>)

#### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Fuchs&#x27;+Endothelial+Dystrophy%5BMAJR%5D%29+AND+%28%28Fuchs+endothelial+dystrophy%5BTIAB%5D%29+OR+%28Fuchs+corneal+dystrophy%5BTIAB%5D%29+OR+%28Fuchs+endothelial+corneal+dystrophy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

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