Fuchs endothelial dystrophy

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. These guttae contribute to the loss of cells in the cornea, leading to 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. The early-onset variant of Fuchs endothelial dystrophy is rare, although the exact prevalence is unknown.

For reasons that are unclear, women are affected with Fuchs endothelial dystrophy somewhat more frequently than men.

Genetic Changes

The genetics of Fuchs endothelial dystrophy are unclear. Researchers have identified regions of a few chromosomes and several genes that they think may play a role in the development of Fuchs endothelial dystrophy, but many of these associations need to be further tested.

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 further vision problems. It is thought that mutations in genes that are active (expressed) primarily in corneal endothelial cells or surrounding tissue may lead 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 the COL8A2 gene. 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 the cells to die and leads to the vision problems in people with this condition.

Mutations in unidentified genes are also likely to cause the early-onset variant of Fuchs endothelial dystrophy. The genetic causes of the late-onset form of the disorder are unknown.

Inheritance Pattern

In some cases, 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.

In many families, the inheritance pattern is unknown.

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

Diagnosis & Management

Genetic Testing

• Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 5
• Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 6
• Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 7

Other Diagnosis and Management Resources
• Duke Health: Corneal Disease
  https://www.dukehealth.org/treatments/eye-care/cornea-disease
• MedlinePlus Encyclopedia: Fuchs Dystrophy
  https://medlineplus.gov/ency/article/007295.htm

General Information from MedlinePlus
• Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
• Drug Therapy
  https://medlineplus.gov/drugtherapy.html
• Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
• Palliative Care
  https://medlineplus.gov/palliativecare.html
• Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources
  MedlinePlus
• Encyclopedia: Fuchs Dystrophy
  https://medlineplus.gov/ency/article/007295.htm
• Health Topic: Corneal Disorders
  https://medlineplus.gov/cornealdisorders.html
• Health Topic: Vision Impairment and Blindness
  https://medlineplus.gov/visionimpairmentandblindness.html

  Genetic and Rare Diseases Information Center
• Fuchs endothelial corneal dystrophy
  https://rarediseases.info.nih.gov/diseases/10018/fuchs-endothelial-corneal-dystrophy
Additional NIH Resources

• National Eye Institute: Cornea and Corneal Disease
  https://nei.nih.gov/health/cornealdisease/

• National Eye Institute: Low Vision
  https://nei.nih.gov/health/lownvision/

Educational Resources

• Digital Reference of Ophthalmology
  http://dro.hs.columbia.edu/fuchs2.htm

• Disease InfoSearch: Corneal Dystrophy Fuchs Endothelial 1
  http://www.diseaseinfosearch.org/Corneal+Dystrophy+Fuchs+Endothelial+1/1908

• MalaCards: fuchs' endothelial dystrophy
  http://www.malacards.org/card/fuchs_endothelial_dystrophy

• Orphanet: Fuchs endothelial corneal dystrophy
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98974

• Wilmer Eye Institute at Johns Hopkins: What is Fuchs Endothelial Corneal Dystrophy?
  https://www.hopkinsmedicine.org/wilmer/conditions/Fuchs/about/

Patient Support and Advocacy Resources

• American Foundation for the Blind
  https://www.afb.org/default.aspx

• Cornea Research Foundation of America
  http://www.cornea.org/

• Corneal Dystrophy Foundation
  https://www.cornealdystrophyfoundation.org/

• National Organization for Rare Disorders (NORD): Corneal Dystrophies
  https://rarediseases.org/rare-diseases/corneal-dystrophies/

• Prevent Blindness America
  https://www.preventblindness.org/

• The Foundation Fighting Blindness
  http://ffb.ca/

ClinicalTrials.gov

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Fuchs+endothelial+dystrophy%22+OR+%22Corneal+Granular+Dystrophies%22+OR+%22Hereditary+Corneal+Dystrophies%22+OR+%22Corneal+Macular+Dystrophy%22
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Fuchs'+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+AND+english%5BLa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 1
  http://omim.org/entry/136800
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 2
  http://omim.org/entry/610158
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 3
  http://omim.org/entry/613267
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 4
  http://omim.org/entry/613268
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 5
  http://omim.org/entry/613269
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 6
  http://omim.org/entry/613270
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 7
  http://omim.org/entry/613271

Sources for This Summary

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2897712/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15914606

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2970683/

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2801746/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18024964

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