Fish-eye disease

Fish-eye disease, also called partial LCAT deficiency, is a disorder that causes the clear front surface of the eyes (the corneas) to gradually become cloudy. The cloudiness, which generally first appears in adolescence or early adulthood, consists of small grayish dots of cholesterol (opacities) distributed across the corneas. Cholesterol is a waxy, fat-like substance that is produced in the body and obtained from foods that come from animals; it aids in many functions of the body but can become harmful in excessive amounts. As fish-eye disease progresses, the corneal cloudiness worsens and can lead to severely impaired vision.

Frequency

Fish-eye disease is a rare disorder. Approximately 30 cases have been reported in the medical literature.

Causes

Fish-eye disease is caused by mutations in the *LCAT* gene. This gene provides instructions for making an enzyme called lecithin-cholesterol acyltransferase (LCAT).

The LCAT enzyme plays a role in removing cholesterol from the blood and tissues by helping it attach to molecules called lipoproteins, which carry it to the liver. Once in the liver, the cholesterol is redistributed to other tissues or removed from the body. The enzyme has two major functions, called alpha- and beta-LCAT activity. Alpha-LCAT activity helps attach cholesterol to a lipoprotein called high-density lipoprotein (HDL). Beta-LCAT activity helps attach cholesterol to other lipoproteins called very low-density lipoprotein (VLDL) and low-density lipoprotein (LDL).

LCAT gene mutations that cause fish-eye disease impair alpha-LCAT activity, reducing the enzyme's ability to attach cholesterol to HDL. Impairment of this mechanism for reducing cholesterol in the body leads to cholesterol-containing opacities in the corneas. It is not known why the cholesterol deposits affect only the corneas in this disorder. Mutations that affect both alpha-LCAT activity and beta-LCAT activity lead to a related disorder called complete LCAT deficiency, which involves corneal opacities in combination with features affecting other parts of the body.

Inheritance Pattern

This condition 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

- alpha-LCAT deficiency
- alpha-lecithin:cholesterol acyltransferase deficiency
- dyslipoproteinemic corneal dystrophy
- FED
- LCATA deficiency
- partial LCAT deficiency

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Fish-eye disease
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342895/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22fish-eye+disease%22+OR+%22LCATA+deficiency%22+OR+%22alpha-LCAT+deficiency%22+OR+%22alpha-lecithin%3Acholesterol+acyltransferase+deficiency%22+OR+%22dyslipoproteinemic+corneal+dystrophy%22

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Corneal Transplant
  https://medlineplus.gov/ency/article/003008.htm
- Oregon Health and Science University: Corneal Dystrophy
  https://www.ohsu.edu/xd/health/services/casey-eye/your-eyes/eye-disorders/cornea-disorders/corneal-dystrophy.cfm

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Corneal Transplant
  https://medlineplus.gov/ency/article/003008.htm
- Health Topic: Corneal Disorders
  https://medlineplus.gov/cornealdisorders.html

Genetic and Rare Diseases Information Center

- Fish-eye disease
  https://rarediseases.info.nih.gov/diseases/6450/fish-eye-disease

Additional NIH Resources

- National Eye Institute: Facts About the Cornea and Corneal Disease
  https://nei.nih.gov/health/cornealdisease/

Educational Resources

- MalaCards: fish-eye disease
  https://www.malacards.org/card/fish_eye_disease
- Orphanet: Fish-eye disease
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=79292
- The University of Arizona Health Sciences
  https://disorders.eyes.arizona.edu/category/alternate-names/fish-eye-disease

Patient Support and Advocacy Resources

- American Foundation for the Blind
  https://www.afb.org/
- Royal National Institute of Blind People: Corneal Dystrophies
  https://www.rnib.org.uk/eye-health/eye-conditions/corneal-dystrophies

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28fish-eye+disease%5BTIAB%5D%29+OR+%28alpha-lcat+deficiency%5BTIAB%5D%29+OR+%28cholesterol+acyltransferase+deficiency%5BTIAB%5D%29+OR+%28partial+lcat+deficiency%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

- FISH-EYE DISEASE
  http://omim.org/entry/136120

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