Cystic fibrosis

Cystic fibrosis is an inherited disease characterized by the buildup of thick, sticky mucus that can damage many of the body's organs. The disorder's most common signs and symptoms include progressive damage to the respiratory system and chronic digestive system problems. The features of the disorder and their severity varies among affected individuals.

Mucus is a slippery substance that lubricates and protects the linings of the airways, digestive system, reproductive system, and other organs and tissues. In people with cystic fibrosis, the body produces mucus that is abnormally thick and sticky. This abnormal mucus can clog the airways, leading to severe problems with breathing and bacterial infections in the lungs. These infections cause chronic coughing, wheezing, and inflammation. Over time, mucus buildup and infections result in permanent lung damage, including the formation of scar tissue (fibrosis) and cysts in the lungs.

Most people with cystic fibrosis also have digestive problems. Some affected babies have meconium ileus, a blockage of the intestine that occurs shortly after birth. Other digestive problems result from a buildup of thick, sticky mucus in the pancreas. The pancreas is an organ that produces insulin (a hormone that helps control blood sugar levels). It also makes enzymes that help digest food. In people with cystic fibrosis, mucus often damages the pancreas, impairing its ability to produce insulin and digestive enzymes. Problems with digestion can lead to diarrhea, malnutrition, poor growth, and weight loss. In adolescence or adulthood, a shortage of insulin can cause a form of diabetes known as cystic fibrosis-related diabetes mellitus (CFRDM).

Cystic fibrosis used to be considered a fatal disease of childhood. With improved treatments and better ways to manage the disease, many people with cystic fibrosis now live well into adulthood. Adults with cystic fibrosis experience health problems affecting the respiratory, digestive, and reproductive systems. Most men with cystic fibrosis have congenital bilateral absence of the vas deferens (CBAVD), a condition in which the tubes that carry sperm (the vas deferens) are blocked by mucus and do not develop properly. Men with CBAVD are unable to father children (infertile) unless they undergo fertility treatment. Women with cystic fibrosis may experience complications in pregnancy.

Frequency

Cystic fibrosis is a common genetic disease within the white population in the United States. The disease occurs in 1 in 2,500 to 3,500 white newborns. Cystic fibrosis is less common in other ethnic groups, affecting about 1 in 17,000 African Americans and 1 in 31,000 Asian Americans.
Causes

Mutations in the *CFTR* gene cause cystic fibrosis. The *CFTR* gene provides instructions for making a channel that transports negatively charged particles called chloride ions into and out of cells. Chloride is a component of sodium chloride, a common salt found in sweat. Chloride also has important functions in cells; for example, the flow of chloride ions helps control the movement of water in tissues, which is necessary for the production of thin, freely flowing mucus.

Mutations in the *CFTR* gene disrupt the function of the chloride channels, preventing them from regulating the flow of chloride ions and water across cell membranes. As a result, cells that line the passageways of the lungs, pancreas, and other organs produce mucus that is unusually thick and sticky. This mucus clogs the airways and various ducts, causing the characteristic signs and symptoms of cystic fibrosis.

Other genetic and environmental factors likely influence the severity of the condition. For example, mutations in genes other than *CFTR* might help explain why some people with cystic fibrosis are more severely affected than others. Most of these genetic changes have not been identified, however.

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

- CF
- cystic fibrosis of pancreas
- fibrocystic disease of pancreas
- mucoviscidosis

Diagnosis & Management

Formal Diagnostic Criteria

- ACT Sheet: Carrier Screening ACT Sheet: CFTR Mutations except R117H Cystic Fibrosis (CF)
- ACT Sheet: Carrier Screening ACT Sheet: No Mutations Detected by Carrier Screening Cystic Fibrosis (CF)
- ACT Sheet: Elevated IRT +/- DNA
  https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/CF.pdf
• American College of Obstetricians and Gynecologists Committee on Genetics.  
ACOG Committee Opinion No. 486: Update on carrier screening for cystic fibrosis.  
Review.  
_Citation on PubMed:_ https://www.ncbi.nlm.nih.gov/pubmed/21422883

• Grody WW, Thompson BH, Gregg AR, Bean LH, Monaghan KG, Schneider A,  
Lebo RV. ACMG position statement on prenatal/preconception expanded carrier  
_Citation on PubMed:_ https://www.ncbi.nlm.nih.gov/pubmed/23619275

**Genetic Testing Information**

• What is genetic testing?  
/ primer/testing/genetictesting

• Genetic Testing Registry: Cystic fibrosis  

**Research Studies from ClinicalTrials.gov**

• ClinicalTrials.gov  
https://clinicaltrials.gov/ct2/results?cond=%22cystic+fibrosis%22

**Other Diagnosis and Management Resources**

• American Society for Reproductive Medicine: Male Infertility  
https://www.reproductivefacts.org/topics/topics-index/male-infertility/

• Baby's First Test  
https://www.babysfirsttest.org/newborn-screening/conditions/cystic-fibrosis-cf

• GeneReview: Cystic Fibrosis and Congenital Absence of the Vas Deferens  
https://www.ncbi.nlm.nih.gov/books/NBK1250

• Genomics Education Programme (UK)  
https://www.genomicseducation.hee.nhs.uk/documents/cystic-fibrosis/

• MedlinePlus Encyclopedia: Cystic Fibrosis  
https://medlineplus.gov/ency/article/000107.htm

• MedlinePlus Medical Tests: Sweat Test for Cystic Fibrosis  
https://medlineplus.gov/lab-tests/sweat-test-for-cystic-fibrosis/

**Additional Information & Resources**

**Health Information from MedlinePlus**

• Encyclopedia: Cystic Fibrosis  
https://medlineplus.gov/ency/article/000107.htm

• Health Topic: Cystic Fibrosis  
https://medlineplus.gov/cysticfibrosis.html
• Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

• Medical Tests: Sweat Test for Cystic Fibrosis
  https://medlineplus.gov/lab-tests/sweat-test-for-cystic-fibrosis/

**Genetic and Rare Diseases Information Center**

• Cystic fibrosis
  https://rarediseases.info.nih.gov/diseases/6233/cystic-fibrosis

**Additional NIH Resources**

• National Heart, Lung, and Blood Institute
  https://www.nhlbi.nih.gov/health-topics/cystic-fibrosis

• National Human Genome Research Institute
  https://www.genome.gov/Genetic-Disorders/Cystic-Fibrosis

**Educational Resources**

• Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/c/cystic-fibrosis

• Centre for Genetics Education

• CFTR2: Clinical and Functional Translation of CFTR
  https://cftr2.org/

• Cochrane Cystic Fibrosis and Genetic Disorders
  https://cfgd.cochrane.org/

• Emory University School of Medicine: Cystic Fibrosis Carrier Testing
  http://genetics.emory.edu/documents/resources/Emory_Human_Genetics_Cystic_Fibrosis_Carrier.PDF

• Genetic Science Learning Center, University of Utah
  https://learn.genetics.utah.edu/content/disorders/singlegene/

• KidsHealth from the Nemours Foundation

• MalaCards: cystic fibrosis
  https://www.malacards.org/card/cystic_fibrosis

• March of Dimes
• Merck Manual Consumer Version
• Orphanet: Cystic fibrosis
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=586
• Virginia Department of Health
• Your Genes Your Health from Cold Spring Harbor Laboratory
  http://www.ygyh.org/cf/whatisit.htm
• Your Genome from Wellcome Genome Campus
  https://www.yourgenome.org/facts/what-is-cystic-fibrosis

Patient Support and Advocacy Resources
• American Lung Association
• Cystic Fibrosis Canada
  https://www.cysticfibrosis.ca/
• Cystic Fibrosis Foundation
  https://www.cff.org/
• Cystic Fibrosis Research, Inc.
  http://cfri.org/
• Cystic Fibrosis Trust (UK)
  https://www.cysticfibrosis.org.uk/
• Medical Home Portal
  https://www.medicalhomeportal.org/newborn/cystic-fibrosis
• Medical Home Portal
  https://www.medicalhomeportal.org/diagnoses-and-conditions/cystic-fibrosis
• National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/cystic-fibrosis/

Clinical Information from GeneReviews
• Cystic Fibrosis and Congenital Absence of the Vas Deferens
  https://www.ncbi.nlm.nih.gov/books/NBK12500
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Cystic+Fibrosis%5BMAJR%29+AND+%28cystic+fibrosis%5BTI%29+AND+review%5Bpt%5D+AND+english%5BLa%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- CYSTIC FIBROSIS
  http://omim.org/entry/219700

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12606185

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