

Familial cylindromatosis

Description

Familial cylindromatosis is a condition involving multiple skin tumors that develop from structures associated with the skin (skin appendages), such as hair follicles and sweat glands. People with familial cylindromatosis typically develop large numbers of tumors called cylindromas. While previously thought to derive from sweat glands, cylindromas are now generally believed to begin in hair follicles.

Individuals with familial cylindromatosis occasionally develop other types of tumors, including growths called spiradenomas and trichoepitheliomas. Spiradenomas begin in sweat glands. Trichoepitheliomas arise from hair follicles. The tumors associated with familial cylindromatosis are generally noncancerous (benign), but occasionally they may become cancerous (malignant). Affected individuals are also at increased risk of developing tumors in tissues other than skin appendages, particularly benign or malignant tumors of the salivary glands.

People with familial cylindromatosis typically begin developing tumors in adolescence or early adulthood. The tumors are most often found in hairy regions of the body, with approximately 90 percent occurring on the head and neck. They grow larger and increase in number over time.

In severely affected individuals, multiple tumors on the scalp may combine into a large, turban-like growth. Large growths frequently develop open sores (ulcers) and are prone to infections. The tumors may also get in the way of the eyes, ears, nose, or mouth and affect vision, hearing, or other functions. The growths can be disfiguring and may contribute to depression or other psychological problems. For reasons that are unclear, females with familial cylindromatosis are often more severely affected than males.

Frequency

Familial cylindromatosis is a rare disorder; its prevalence is unknown.

Causes

Familial cylindromatosis is caused by mutations in the *CYLD* gene. This gene provides instructions for making a protein that helps regulate nuclear factor-kappa-B. Nuclear factor-kappa-B is a group of related proteins that help protect cells from self-destruction (apoptosis) in response to certain signals. In regulating the action of nuclear factor-

kappa-B, the CYLD protein allows cells to respond properly to signals to self-destruct when appropriate, such as when the cells become abnormal. By this mechanism, the CYLD protein acts as a tumor suppressor, which means that it helps prevent cells from growing and dividing too fast or in an uncontrolled way.

People with familial cylindromatosis are born with a mutation in one of the two copies of the *CYLD* gene in each cell. This mutation prevents the cell from making functional CYLD protein from the altered copy of the gene. However, enough protein is usually produced from the other, normal copy of the gene to regulate cell growth effectively. For tumors to develop, a second mutation or deletion of genetic material involving the other copy of the *CYLD* gene must occur in certain cells during a person's lifetime.

When both copies of the *CYLD* gene are mutated in a particular cell, that cell cannot produce any functional CYLD protein. The loss of this protein allows the cell to grow and divide in an uncontrolled way to form a tumor. In people with familial cylindromatosis, a second *CYLD* mutation typically occurs in multiple cells over an affected person's lifetime. The loss of CYLD protein in these cells leads to the growth of skin appendage tumors.

Some researchers consider familial cylindromatosis and two related conditions called multiple familial trichoepithelioma and Brooke-Spiegler syndrome, which are also caused by *CYLD* gene mutations, to be different forms of the same disorder. It is unclear why mutations in the *CYLD* gene cause different patterns of skin appendage tumors in each of these conditions, or why the tumors are generally confined to the skin in these disorders.

[Learn more about the gene associated with Familial cylindromatosis](#)

- CYLD

Inheritance

Susceptibility to familial cylindromatosis has an autosomal dominant pattern of inheritance, which means one copy of the altered gene in each cell increases the risk of developing this condition. However, a second, non-inherited mutation is required for development of skin appendage tumors in this disorder.

Other Names for This Condition

- Anzell-Spiegler cylindromas
- cylindromatosis, familial
- dermal eccrine cylindroma
- turban tumor syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Cyldromatosis, familial (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1851526/>)

Genetic and Rare Diseases Information Center

- Dermal eccrine cylindroma (<https://rarediseases.info.nih.gov/diseases/10345/dermal-eccrine-cylindroma>)

Patient Support and Advocacy Resources

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

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov (<https://clinicaltrials.gov/ct2/results?cond=%22dermal+eccrine+cylindroma%22+OR+%22Sweat+Gland+Neoplasms%22>)

Catalog of Genes and Diseases from OMIM

- CYLINDROMATOSIS, FAMILIAL (<https://omim.org/entry/132700>)

Scientific Articles on PubMed

- PubMed (<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28familial+cylindromatosis%5BTIAB%5D%29+OR+%28ancell-spiegler+cylindromas%5BTIAB%5D%29+OR+%28turban+tumor+syndrome%5BTIAB%5D%29+OR+%28dermal+eccrine+cylindroma%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D>)

References

- Almeida S, Maillard C, Itin P, Hohl D, Huber M. Five new CYLD mutations in skin appendage tumors and evidence that aspartic acid 681 in CYLD is essential for deubiquitinase activity. *J Invest Dermatol.* 2008 Mar;128(3):587-93. Epub 2007 Sep 13. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/17851586>)
- Bignell GR, Warren W, Seal S, Takahashi M, Rapley E, Barfoot R, Green H, Brown C, Biggs PJ, Lakhani SR, Jones C, Hansen J, Blair E, Hofmann B, Siebert R, Turner G, Evans DG, Schrandt-Stumpel C, Beemer FA, van Den Ouweland A,

Halley D, Delpech B, Cleveland MG, Leigh I, Leisti J, Rasmussen S. Identification of the familial cylindromatosis tumour-suppressor gene. *Nat Genet.* 2000 Jun;25(2):160-5. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/10835629>)

- Bowen S, Gill M, Lee DA, Fisher G, Geronemus RG, Vazquez ME, Celebi JT. Mutations in the CYLD gene in Brooke-Spiegler syndrome, familial cylindromatosis, and multiple familial trichoepithelioma: lack of genotype-phenotype correlation. *J Invest Dermatol.* 2005 May;124(5):919-20. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/15854031>)
- Lee DA, Grossman ME, Schneiderman P, Celebi JT. Genetics of skin appendage neoplasms and related syndromes. *J Med Genet.* 2005 Nov;42(11):811-9. Review. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/16272260>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735949/>)
- Massoumi R, Paus R. Cylindromatosis and the CYLD gene: new lessons on the molecular principles of epithelial growth control. *Bioessays.* 2007 Dec;29(12):1203-14. Review. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/18008375>)
- Saggar S, Chernoff KA, Lodha S, Horev L, Kohl S, Honjo RS, Brandt HR, Hartmann K, Celebi JT. CYLD mutations in familial skin appendage tumours. *J Med Genet.* 2008 May;45(5):298-302. doi: 10.1136/jmg.2007.056127. Epub 2008 Jan 30. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/18234730>)
- Young AL, Kellermayer R, Szigeti R, Tészás A, Azmi S, Celebi JT. CYLD mutations underlie Brooke-Spiegler, familial cylindromatosis, and multiple familial trichoepithelioma syndromes. *Clin Genet.* 2006 Sep;70(3):246-9. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/16922728>)

Page last updated on 18 August 2020

Page last reviewed: 1 June 2012