# Familial candidiasis

## Description

Familial candidiasis is an inherited tendency to develop infections caused by a type of fungus called *Candida*. Affected individuals typically have infections of the skin, the nails, and the moist lining of body cavities (mucous membranes). These infections are recurrent and persistent, which means they come back repeatedly and can last a long time. This pattern of infection is called chronic mucocutaneous candidiasis.

*Candida* is commonly present on the skin and on the mucous membranes, and in most people usually causes no health problems. However, certain medications (such as antibiotics and corticosteroids) and other factors can lead to occasional overgrowth of *Candida* (candidiasis) in the mouth (where it is known as thrush) or in the vagina. These episodes, commonly called yeast infections, usually last only a short time before being cleared by a healthy immune system.

Most people with familial candidiasis have chronic or recurrent yeast infections that begin in early childhood. Skin infections lead to a rash with crusty, thickened patches; when these patches occur on the scalp, they can cause loss of hair in the affected area ( scarring alopecia). Candidiasis of the nails can result in thick, cracked, and discolored nails and swelling and redness of the surrounding skin. Thrush and gastrointestinal symptoms such as bloating, constipation, or diarrhea are common in affected individuals. Women with familial candidiasis can develop frequent vaginal yeast infections, and infants can have yeast infections on the skin that cause persistent diaper rash.

Depending on the genetic change involved in this condition, some affected individuals are at risk for developing systemic candidiasis, a more severe condition in which the infection spreads through the bloodstream to various organs including the brain and the meninges, which are the membranes covering the brain and spinal cord. Systemic candidiasis can be life-threatening.

Chronic or recurrent yeast infections can occur in people without familial candidiasis. Some individuals experience recurrent candidiasis as part of a general susceptibility to infections because their immune systems are impaired by a disease such as acquired immune deficiency syndrome (AIDS) or severe combined immunodeficiency (SCID), medications, or other factors. Other individuals have syndromes such as autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED) or autosomal dominant hyper-IgE syndrome (AD-HIES) that include a tendency to develop

candidiasis along with other signs and symptoms affecting various organs and systems of the body.

## Frequency

*Candida* is present on the skin and mucous membranes of up to half the population at any given time, normally without creating health problems. The prevalence of the inherited susceptibility to *Candida* infections that characterizes familial candidiasis is unknown, but the condition is thought to be rare.

## Causes

Mutations in any of several genes have been identified in people with familial candidiasis. These genes include *CARD9*, *IL17RC*, *STAT1*, and others. The genes associated with familial candidiasis provide instructions for making proteins that are involved in immune system function.

When the immune system recognizes *Candida*, it generates cells called Th17 cells. These cells produce signaling molecules (cytokines) called the interleukin-17 (IL-17) family as part of an immune process called the IL-17 pathway. The IL-17 pathway creates inflammation, sending other cytokines and white blood cells that fight foreign invaders and promote tissue repair. In addition, the IL-17 pathway promotes the production of certain antimicrobial protein segments (peptides) that control growth of *Candida* on the surface of mucous membranes.

The gene mutations associated with familial candidiasis interfere with the IL-17 pathway in various ways. Mutations in several genes, including *IL17RC*, impair signaling in the IL-17 pathway. Mutations in other genes, including *STAT1* and *CARD9*, are thought to block (inhibit) the activity of the pathway. Impairment of the IL-17 pathway diminishes the body's immune response to *Candida*, leading to the chronic or recurrent yeast infections that occur in people with familial candidiasis. Mutations in most of the genes associated with familial candidiasis cause chronic mucocutaneous candidiasis; only *CARD9* gene mutations have also been known to lead to systemic candidiasis in some affected individuals.

Learn more about the genes associated with Familial candidiasis

- CARD9
- IL17RC
- STAT1

**Additional Information from NCBI Gene:**

- CLEC7A
- IL17F
- IL17RA

- RORC
- TRAF3IP2

## Inheritance

Familial candidiasis can be inherited in different patterns. People with this disorder inherit a tendency to develop recurrent or chronic *Candida* infections, not the infections themselves. Familial candidiasis caused by mutations in some genes, including *STAT1*, is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Familial candidiasis caused by mutations other genes, such as *CARD9* or *IL17RC*, 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

- Familial chronic mucocutaneous candidiasis

## Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Familial chronic mucocutaneous candidiasis (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0341024/)

Genetic and Rare Diseases Information Center

- Familiar chronic mucocutaneous candidiasis (https://rarediseases.info.nih.gov/diseases/12313/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CANDIDIASIS, FAMILIAL, 1; CANDF1 (https://omim.org/entry/114580)
- IMMUNODEFICIENCY 103, SUSCEPTIBILITY TO FUNGAL INFECTIONS; IMD103 (https://omim.org/entry/212050)
- CANDIDIASIS, FAMILIAL, 3; CANDF3 (https://omim.org/entry/607644)

- IMMUNODEFICIENCY 42; IMD42 (https://omim.org/entry/616622)
- IMMUNODEFICIENCY 31C; IMD31C (https://omim.org/entry/614162)
- CANDIDIASIS, FAMILIAL, 8; CANDF8 (https://omim.org/entry/615527)
- CANDIDIASIS, FAMILIAL, 4; CANDF4 (https://omim.org/entry/613108)
- IMMUNODEFICIENCY 51; IMD51 (https://omim.org/entry/613953)
- CANDIDIASIS, FAMILIAL, 6; CANDF6 (https://omim.org/entry/613956)
- CANDIDIASIS, FAMILIAL, 9; CANDF9 (https://omim.org/entry/616445)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28familial+candidiasis%5BTIAB%5D%29+OR+%28chronic+mucocutaneous+candidiasis%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

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