



## Overview of Pachyonychia Congenita

Pachyonychia congenita is a very rare genetic disorder that mostly affects the skin and nails. The disorder is usually seen starting at birth or early in life, and it affects people of both sexes and all racial and ethnic groups.

### What happens in pachyonychia congenita?

Pachyonychia congenita causes overgrowth of the nails and thick, painful calluses on the bottoms of the feet.

### Who Gets Pachyonychia Congenita?

Pachyonychia congenita affects people of both sexes and all racial and ethnic groups. In about half of the cases, the disorder is inherited from a parent. In the other half, there is no family history of the disorder.

### Types of Pachyonychia Congenita

There are five types of pachyonychia congenita, based on the gene that is altered. These genes include: *KRT6A*, *KRT6B*, *KRT6C*, *KRT16*, and *KRT17*. Thickened nails and calluses occur in all forms of the disorder. Severity of symptoms depends largely on the specific genetic mutation.

### Symptoms of Pachyonychia Congenita

Common symptoms of pachyonychia congenita include:

- Thickened nails on the fingers and toes.
- Painful blisters and thick calluses on the soles of the feet when a child begins to walk. In some cases, blisters and calluses can form on the palms of the hands.

Other symptoms depend on the specific type of pachyonychia congenita and can include:

- Thick white patches on the tongue and inside the cheeks.

- Bumps around the hair follicles on the elbows, knees, and waistline.
- Cysts all over the body.
- Babies born with teeth that crumble and fall out soon after birth.

Some symptoms tend to persist over time. Others may change, depending on a person's weight, activity level, and exposure to environmental stressors. Cysts may worsen during puberty, but bumps around the hair follicles may improve with age.

## Causes of Pachyonychia Congenita

Pachyonychia congenita is caused by mutations in one of at least five genes that help produce keratins (tough proteins that make up the skin, nails, and hair).

These mutations change keratin structure so that nails thicken, and skin cells are more sensitive to minor stress such as walking. Painful blisters form and are covered by thick calluses on the bottoms of the feet. In some cases, blisters also form on the palms of the hands.

## Diagnosis of Pachyonychia Congenita

The only way to diagnose pachyonychia congenita is with a genetic test. This is because symptoms of the disorder are similar to those of other conditions. For example, thickened nails also can be caused by [psoriasis](#), and white patches inside the mouth may be confused with thrush (a yeast infection common in babies).

## Treatment of Pachyonychia Congenita

There is no cure or medicine to treat pachyonychia congenita. Over-the-counter medications are commonly used to treat pain associated with symptoms.

Find tips to help you deal with the condition in the [Living With section](#).

## Who Treats Pachyonychia Congenita?

Because pachyonychia congenita is extremely rare, few doctors have experience treating it. Your best choices include:

- Dermatologist, who specializes in skin disorders.
- Primary care doctor: Your own doctor knows your medical history, your lifestyle, and your special needs and can help as problems occur.

# Living With Pachyonychia Congenita

There are no medicines for pachyonychia congenita, but there are things you can do to cope with the disease. They include:

- Grinding or shaving down thickened nails and skin. Take care that you don't make them too thin, as this could cause pain and infection.
- Wearing gloves to protect the hands during activities like riding a bicycle or using hand tools.
- Wearing comfortable shoes and socks that reduce moisture. This will reduce rubbing that can worsen painful calluses.

## Research Progress Related to Pachyonychia Congenita

Research on pachyonychia congenita focuses on "shutting down" the disease gene and reducing the pain caused by the disorder. Some research includes:

- **Medications.**
  - Rapamycin. Scientists are exploring whether this drug, which suppresses the immune system, can be applied to calluses to relieve pain.
  - Statins. Early research suggests that these cholesterol-lowering drugs may affect one of the genes responsible for pachyonychia congenita.
  - Pain medications. Many people with pachyonychia congenita have neuropathic pain, which is burning, tingling, numbness, or pain for no reason. Studies are looking at drugs to treat this type of pain.
- **Gene therapy.** Research is exploring ways to shut down the faulty disease genes without interfering with healthy ones.
- **Shoe inserts.** Painful calluses on the bottoms of the feet can make walking difficult. Scientists are working to develop special shoe insoles that cushion and cool the foot to reduce pain.

## For More Info

### U.S. Food and Drug Administration

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

Drugs@FDA at <https://www.accessdata.fda.gov/scripts/cder/daf>. Drugs@FDA is a searchable catalog of FDA-approved drug products.

**Centers for Disease Control and Prevention, National Center for Health Statistics**

Website: <https://www.cdc.gov/nchs>

**NIH Office of Rare Diseases Research Genetic and Rare Diseases Information Center**

Website: <https://rarediseases.info.nih.gov>

**National Library of Medicine's Genetics Home Reference**

Website: <https://ghr.nlm.nih.gov/condition/pachyonychia-congenita>

**Pachyonychia Congenita Project**

Website: <https://www.pachyonychia.org>

**American Academy of Dermatology**

Website: <https://www.aad.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at [NIAMSInfo@mail.nih.gov](mailto:NIAMSInfo@mail.nih.gov).

- [Asian Language Health Information](#)
- [Spanish Language Health Information](#)