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Pachyonychia congenita

What is pachyonychia congenita?

Pachyonychia congenita is a rare, inherited disorder of keratinisation. This is the process by which keratin is formed and deposited in the outer most layer of the skin. It occurs due to mutation in the genes encoding keratin 6a, 6b, 16 and 17.

How is it inherited?

Pachyonychia congenita is most often autosomal dominantly inherited. That means the defective gene comes from one parent. An affected person has a 50% chance of passing on the disease to his or her offspring at each pregnancy.

A few cases of autosomal recessive pachyonychia congenita have also been reported, in which a family history may not be known as the abnormal gene is inherited from both parents. In this case, on average, one in every four of the offspring are affected.

Sporadic cases may also arise, due to mutations during conception.

What are the features of pachyonychia congenita?

Pachyonychia congenita affects the skin (especially palms and soles), nails, hair and mucous membranes.

Pachyonychia congenita



Clinical feature of pachyonychia congenita

Palms and soles	<ul style="list-style-type: none"> • Painful thickened palms and soles (palmoplantar hyperkeratosis or keratoderma), resulting in difficulty walking • Hyperhidrosis (excessive sweating) • Blisters precipitated by friction and warm weather
Nails	<ul style="list-style-type: none"> • Abnormal nails apparent early in life • Nails are thickened with brownish discolouration • All finger nails are commonly involved; toe nails to a lesser degree

Hair	<ul style="list-style-type: none"> • Coarse unruly hair • Sometimes loss of hair (alopecia)
Eyes	<ul style="list-style-type: none"> • Corneal opacities • Cataracts
Larynx	<ul style="list-style-type: none"> • Hoarseness may occur • Rarely, respiratory obstruction and death in infancy

Classification of pachyonychia congenita

Pachyonychia congenita is classified into at least two types according to the clinical features,. Particular features may be more prominent in some patients than in others.

Pachyonychia congenita type 1

Type 1 (MIM# 167200) is also known as the Jadassohn–Lewandowsky type of pachyonychia congenita. Features are:

- Nail changes
- Palmoplantar hyperkeratosis
- Follicular hyperkeratosis (thickened horny plugs) on the face & extensor surfaces of the limbs
- Oral leukokeratosis (whitish areas inside the mouth)

Pachyonychia congenita type 2

Type 2 (MIM# 167201) or the Jackson–Lawler type of pachyonychia congenita includes the features of type 1 plus:

- Natal (birth) teeth
- [Steatocystoma multiplex](#)
- [Epidermoid cysts](#)
- Hair abnormalities

How is Pachyonychia congenita diagnosed?

Pachyonychia congenita is diagnosed by its clinical appearance.

Skin biopsies of the affected tissues will only show nonspecific changes.

Molecular genetic studies can be done by specialist laboratories to detect mutations in the affected keratin genes.

Treatment

Unfortunately there is no cure for pachyonychia congenita. The effectiveness of treatment depends on the severity of the specific problem.

Treatment might include:

- [Emollients](#)
- Keratolytics e.g. [salicylic acid](#) ointment
- Mechanical debridement
- Oral retinoids e.g. [acitretin](#)

Related information

References:

- [OMIM – Online Mendelian Inheritance in Man](#) (search term pachyonychia congenita)
- [OMIM – Online Mendelian Inheritance in Man](#) (search term Pachyonychia congenita)

On DermNet NZ:

- [Scaly skin conditions](#)
- [Palmoplantar keratoderma](#)
- [Nail disease](#)

Other websites:

- [Pachyonychia Congenita](#) – emedicine dermatology

Books about skin diseases:

See the [DermNet NZ bookstore](#)

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DermNet does not provide an on-line consultation service.
If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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