Pachyonychia congenita (PC) is a rare genetic skin disorder. In 2011, a more correct classification system was introduced based on clinical findings in over 600 patients with genetically confirmed PC. This current system classifies the types based on the specific gene that carries the mutation, i.e. PC-K6a, PC-K6b, PC-K6c, PC-K16 and PC-K17. Previously, two distinct types of PC were recognized: PC-1, or Jadassohn-Lewandowsky type, and PC-2, or Jackson-Lawler type. However, the clinical findings do not match these categories and these types are now not relevant.

PC follows an autosomal dominant pattern. Autosomal means the genetic defect is carried on one of the 22 human chromosomes that do not determine sex. Dominant means the gene with the mutation dominates over normal skin and produces. A person with one dominant gene for PC and one gene for normal skin will have PC. Nearly one-half of those with genetically confirmed PC are spontaneous cases, meaning that the disorder is spontaneous and not inherited.

Keratins are proteins that are important to the normal function of hair, nails and skin. Based on the findings of the more than 600 confirmed PC patients, there are no hair changes with PC mutations. Not every affected individual exhibits every clinical feature. Each person may display a unique set of signs and symptoms, even within families.

What are the Signs & Symptoms?

The main clinical features associated with pachyonychia congenita include:

All five types (PC-K6a, PC-K6b, PC-K6c, PC-K16 and PC-K17)

- Thickened fingernails and toenails (not necessarily 20/20 nail dystrophy and some have no affected fingernails).
- Painful Plantar keratoderma - blisters and thick calluses on the soles of the feet causing extreme pain.

May be present in any PC type, but more common in specific PC types

- Palmar keratoderma – blisters and thick calluses on the palms of the hands (more likely PC-K16).
- Oral leukokeratosis – thick white plaque on the tongue and the insides of the cheeks (more likely PC-K6a).
- Follicular hyperkeratosis – bumps that form around the hair follicles (more likely PC-K6a).
- Possible involvement of the larynx – hoarseness or thickening of the voice box (more likely PC-K6a).
- Natal or prenatal teeth (associated with PC-K17).
- Cysts – including steatocystoma (epithelial/skin) and other forms of cysts (more likely PC-K17 and PC-K6a).
Possible association with PC

- Hyperhidrosis (excessive sweating) and non-epidermal cysts.

Syndromes similar to PC, including palmoplantar keratoderma (PPK), epidermolytic palmoplantar keratoderma (EPPK), and Tylosis or Unna-Thost syndrome, as well as other conditions, have been clinically misdiagnosed as PC and articles have been published as cases of PC. However, now that genetic testing is available, these other conditions can be identified. Based on the data of over 600 genetically confirmed PC patients, with PC there is no alopecia, no corneal dystrophy, no bone deformities, no deafness, no mental retardation. All of these characteristics are indications of different disorders and not PC and this should assist in better differential diagnosis in clinic.

What is the Treatment?

There is no effective treatment for PC. Several clinical studies and clinical trials are underway, sponsored by the patient advocacy group Pachyonychia Congenita Project (PC Project). Currently, treatment of PC is primarily symptomatic. Emollients (moisturizers) and keratolytics (products containing alpha-hydroxy acids) provide little improvement for the hyperkeratosis and mechanical removal of the callus several times a week is usually necessary. Routine grinding of the nail plates can minimize their interference with function. Oral retinoids such as Accutane have no positive effect for those with PC. The retinoids Soriatane, Tigason or Neo-Tigason have minimal effect, but may allow some slight relief especially of palmar keratoderma when used in low dosage or variable dosage. Retinoids are used cautiously due to their known bone toxicity and other complications.

This information is provided as a service to patients and parents of patients who have ichthyosis. It is not intended to supplement appropriate medical care, but instead to complement that care with guidance in practical issues facing patients and parents. Neither FIRST, its Board of Directors, Medical & Scientific Advisory Board, Board of Medical Editors nor Foundation staff and officials endorse any treatments or products reported here. All issues pertaining to the care of patients with ichthyosis should be discussed with a dermatologist experienced in the treatment of their skin disorder.