Understanding Pachyonychia Congenita
THE CONDITION AND THE MISSION TO END THE DEBILITATING PAIN

IT’S MORE THAN JUST THICK NAILS
While “pachy” and “onychia” together translate to thickened nails, *pachyonychia congenita* (PC) includes a spectrum of symptoms.

DO YOU HAVE PATIENTS WITH FEATURES* OF GENETICALLY-CONFIRMED PC?

- Plantar keratoderma with/without blisters
- Palmar keratoderma and thickened nails
- Follicular hyperkeratosis, oral leukokeratosis, and cysts

*Not all patients have all of these phenotypes. The most common feature is painful plantar keratoderma.

DISEASE OVERVIEW

- **PC is an ultra-rare genetic autosomal dominant skin disorder** involving a mutation in one of five keratin genes: *KRT6A, KRT6B, KRT6C, KRT16, KRT17*. While many cases are inherited, approximately 30% to 40% of patients in the International PC Research Registry (IPCRR) have spontaneous mutations.
- **Prevalence is estimated between 5,000 and 10,000 cases** globally. However, due to lack of disease state awareness and access to standardized genetic testing worldwide, these numbers likely fail to accurately represent the true prevalence of PC.
- **Diagnosis is confirmed with genetic testing.** Clinical presentation alone is insufficient as there are other inherited skin disorders with overlapping symptoms. Differential diagnoses include Clouston syndrome, Epidermolysis bullosa simplex, Olmsted syndrome, Tylosis, Striate palmoplantar keratoderma, and non-syndromic congenital nail disorders such as recessive nail dysplasia.

DEBILITATING EFFECTS OF PC

- **Disabling and unpredictable pain** from plantar keratoderma and blisters
- **Negative impacts on quality of life** due to the need for walking aids such as crutches, canes, and wheelchairs
- **Repetitive damage** due to the fragility of the skin cells results additive pain, inflammation, blistering, and potential for infections
- **No permanent pain relief** due to lack of cure for PC

THE FACES OF PC

THE VOICES OF PC

“It feels like walking on stones in your shoes. When you sit down the pain doesn’t go away.”

“I don’t know what a painless day is.”

“Walking and working are specific activities that are important to me but are also things I cannot do at all or as fully as I would like because of the condition.”

**PACHYONYCHIA CONGENITA IS A LIFELONG CONDITION THAT AFFECTS PEOPLE OF ALL AGES, RACES, NATIONALITIES, AND GENDERS**
Pachyonychia Congenita (PC) Project is the only patient advocacy organization in the world dedicated to patients with PC, a painful and debilitating skin disorder. PC Project unites patients, researchers, medical professionals, and industry partners on a global level to advance research and drug development for meaningful treatments and ultimately, a cure for PC.

SERVICES PROVIDED BY PC PROJECT

**International Pachyonychia Congenita Research Registry (IPCRR):** A WIRB-approved registry (WIRB Study #20040468) that gathers data from patients with clinical signs of PC. Registrants are offered free genetic testing, individualized support, notified of studies for PC treatments, and activities (online forums, patient support meetings, etc.).

**International PC Consortium (IPCC):** A group founded by experts in PC and fueled by dedication to patients impacted with PC. The IPCC facilitates collaboration among scientists, physicians, and industry partners interested in advancing research and translational therapeutics for PC. De-identified data from the registry are readily available for research purposes.

**Annual Patient Support Meetings:** An educational gathering where patients meet, teach and support one another and effectively defeat the overwhelming loneliness caused by an ultra rare disease. These meetings have been held each year since 2004 in Europe and in the USA with patients, physicians, scientists, and pharmaceutical representatives participating.

**Research Grants to fund PC-related projects:** Past grants have included specific gene inhibitors, small molecule drug screening, development of targeted siRNA, development of human skin equivalents, and other innovative research. Grant proposals are currently accepted by PC Project on an open application basis.

**Research and Industry Partnerships:** PC Project collaborates with stakeholders interested in developing therapeutics for PC by surveying PC registry patients, educating decision makers about PC, holding patient focus groups, helping with designing clinical endpoints, advertising of studies, and recruiting patients for studies.

NEED MORE INFORMATION? PC PROJECT CAN HELP

- Visit [www.pachyonychia.org](http://www.pachyonychia.org) for current information for patients, researchers, and clinicians, including a full-text bibliography of 700+ articles related to PC and keratin research. More than 100 articles have been published in collaboration with PC Project.
- To join the IPCC, refer a patient, or partner with PC Project, email info@pachyonychia.org or call 801-987-8758.

Scan the QR code or visit [www.pachyonychia.org](http://www.pachyonychia.org) to learn more about PC, PC Project, IPCC, and the IPCRR.