Services Supported by PC Project

The International PC Research Registry (IPCRR) is one of the most important achievements of PC Project. The registry received IRB approval in May 2004 (WIRB Study #20040468) and has enrolled over 2100 patients in over 60 countries. More than 990 patients are genetically confirmed with PC. Free genetic testing is provided to those in the IPCRR. De-identified data is freely shared with interested researchers and patients are supported in a caring, global community. For more information, visit www.pachyonychia.org/patient-registry/

The International PC Consortium (IPCC) includes scientists, physicians, and industry partners from around the world to encourage and support collaborative research and drug development related to keratin disorders and finding treatments for PC. Since 2004, PC Project has hosted the annual IPCC Symposium.

Annual Patient Support Meetings 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. Applications are accepted on an open application schedule.

Research and Industry Partnerships in which 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.

The PC Website at www.pachyonychia.org provides accurate and current information for patients, researchers, and clinicians and includes a full-text bibliography of 700+ articles related to PC and keratin research. More than 100 articles have been published in connection with some aspect of PC Project.

Newsletters for both the patient community and for IPCC members to inform and unite these groups. To join the IPCC or the newsletter mailing list, email info@pachyonychia.org.

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Fighting for a cure.
Connecting and helping patients.
Empowering research.

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What is Pachyonychia Congenita (PC)?

PC is an ultra-rare genetic autosomal dominant keratin disorder. It is a chronically debilitating disease due to impaired ambulation associated with plantar keratoderma, blistering, and pain that make walking difficult or impossible.

Other common features of PC include palmar keratoderma, nail dystrophy, follicular hyperkeratosis and cysts, which can also be extremely painful.

In some patients, leukokeratosis may form in the throat and laryngeal area as well as on the oral mucosa. In children, treatment of the larynx can cause increased overgrowth, but may sometimes be necessary to avoid obstruction.

PC is caused by a mutation in one of five keratin genes: KRT6A, KRT6B, KRT6C, KRT16, or KRT17. There are both unique and overlapping features across the PC types. Individuals with the same mutation may have varied phenotypic signs. The most consistent finding is intense plantar pain.

Nails vary widely between patients. There is no typical ‘PC nail’. No patients in the PC registry with genetically confirmed PC have only nail dystrophy.

Oral Leukokeratosis may be misdiagnosed as thrush in infants and young children with PC.

Cysts are a common feature of PC-K17 but are seen in all types of PC. They rarely occur prior to puberty.

Follicular Hyperkeratosis at the waist, knees, elbows and other high friction areas is most severe in children and often lessens after age 20.

Mutant Keratin

Current Therapies for PC

Presently, there is no cure for PC. No successful drug therapy for PC has been reported that provides long term benefits. Patients manage their symptoms in a variety of ways. Effective personal care is essential for PC patients.

Two basic elements of care for keratoderma and nail dystrophy are:

1. soaking to soften skin, followed by
2. debridement. The debridement may be done at home or with professional care.

The most consistent concern for all PC patients is pain.

- Plantar pain is the most consistent finding of PC. Blisters can form under and around the calluses and neurovascular structures can grow in the calluses.
- Sometimes thickened nails result in painful nail infections.
- Cysts, especially when inflamed, can cause pain.
- Pain is caused by follicular hyperkeratosis.
- Some patients report intense ear pain lasting about 25 seconds. This ear pain may be related to salivary glands and is called ‘first bite syndrome’.

Please refer PC patients to the International PC Research Registry (IPCCR) sponsored by PC Project that provides free genetic testing and patient support.

Referring physicians are invited to be part of the International PC Consortium, participate in publications, and have access to research and clinical studies.

“...If you have PC you get blisters all over the bottoms of your feet (and sometimes on your hands). The blisters hurt a lot. It is difficult to explain how hard it is to live with PC. You have to find a way to survive and live with pain.” 16 year old patient

Photos courtesy of Prof. W. H. Irwin McLean, University of Dundee, Scotland

Keratin Immunofluorescence