Pachyonychia Congenita Project (PC Project) Overview

Pachyonychia Congenita Project (PC Project) is a 501(c)(3) nonprofit founded in 2003 with the specific goal of finding a cure for Pachyonychia Congenita (PC), an extremely painful and debilitating genetic skin disease which makes walking and standing difficult and sometimes even impossible. Because there is currently no effective treatment or cure for PC, everything PC Project does is dedicated to its mission: "Fighting for a cure, connecting and helping patients, and empowering research."

The most important asset of PC Project is the International Pachyonychia Congenita Research Registry. Continually growing, this Patient Registry is the connecting link to each facet of PC Project's mission. Individuals suspected of having PC join the online Registry by completing a questionnaire, consent form, and uploading photos of their symptoms at https://www.pachyonychia.org/patient-registry/. Dermatologists and other clinicians worldwide are also able to register their patients through the same online portal. Cases are then reviewed by members of a genetics team who are members PC Project's Medical and Scientific Advisory Board.

With a clinical diagnosis, participants in the registry are eligible to receive free genetic testing to determine the affected gene and resulting mutation*. Currently, over 116 mutations have been discovered within the five PC genes. Genetic testing is particularly important as other skin diseases can mimic PC. For a PC patient who has often lived many years not knowing why he or she has these symptoms, a correct genetic diagnosis of this rare disorder is empowering. For a researcher or drug developer, a genetic diagnosis assures professionals they are dealing with actual PC patients, and in specific studies, targeting the correct gene and mutation.

PC Project shares deidentified data from the Registry with interested scientists, physicians and industry partners. This data has resulted in numerous publications and has literally changed what the medical and scientific community knows about PC. Furthermore, PC Project utilizes the Registry to recruit patients for studies, clinical trials and other projects to further drug development. Those overseeing clinical studies have the confidence of knowing that PC Project, through the Registry, knows who the patients are, where the patients are, and the exact affected gene and mutation for each patient.

Patients who enroll in the Registry receive benefits such as disability assistance, access to physicians on the PC Medical Advisory Board, and connection to a worldwide patient community, which includes annual patient support meetings, monthly newsletters and thriving private social media groups. Because PC is an isolating disease, this global community provides patients and their caregivers with support, practical management helps and comradery within a group that completely understands. This fulfills an unmet need, particularly for patients who have been bullied, felt hopeless, and who have lived in shame and embarrassment as they deal physically and mentally with daily pain.

As the only organization in the world dedicated to relieving the suffering of PC patients, PC Project is passionate about helping patients live fulfilling lives while navigating this painful,

debilitating disease, all the while never losing focus of the larger goals – getting effective therapeutics to patients and eventually a cure.

*Usually, saliva is needed from only one family member.