PACHYONYCHIA CONGENITA

Pachyonychia congenita is a rare inherited disorder that affects the nails, skin, hair, and teeth. The term “pachyonychia congenita” comes from the Greek words “pachy” meaning thick, “onychia” meaning nails, and “congenita” meaning present from birth. The disorder is caused by a mutation in one of five genes that code for keratin proteins, which are building blocks for hair, skin, and nails. The mutations prevent keratins from forming the strong network of filaments that normally gives skin cells strength and resilience.

The symptoms of pachyonychia congenita can vary widely among affected individuals, even within the same family. The most common symptoms include thickened nails (pachyonychia), painful calluses on the soles of the feet (plantar keratoderma), painful blisters on the hands and feet, white patches on the inside of the cheeks and throat (oral leukokeratosis), and cysts in the skin. The symptoms typically appear at birth or during childhood and gradually worsen over time.

Pachyonychia congenita can be diagnosed through a physical examination of the nails, skin, and mouth. Genetic testing can also be used to confirm a diagnosis. It is important to diagnose the disorder early in life to provide appropriate care and management. There is no cure for pachyonychia congenita, but treatment options are available to manage the symptoms. Treatment may include regular foot care to manage calluses and blisters, wearing soft shoes with padded soles, and avoiding activities that cause friction or pressure on the skin. Medications such as pain relievers and antibiotics may also be prescribed as needed. In some cases, surgery may be necessary to remove cysts or correct nail deformities.

Living with pachyonychia congenita can be challenging, but it is possible to manage the symptoms and lead a fulfilling life. Support groups such as the Pachyonychia Congenita Project provide resources and a community of people who understand the challenges of living with the disorder. It is important to work closely with healthcare providers to develop a care plan that addresses the specific needs of each individual. Early diagnosis and treatment can help manage the symptoms and improve the quality of life for those living with the disorder.