Understanding Pachyonychia Congenita
PC Advocate Training Session 2
Pachyonychia Congenita (PC) is a...
Pachyonychia Congenita (PC) is a...

Rare (Orphan)

ULTRA rare
est 2,000-10,000

Genetic autosomal dominant

Keratin

$KRT6A$, $KRT16$, $KRT17$, $KRT6B$, $KRT6C$

Skin
Pachyonychia Congenita (PC)

Rare (Orphan)
ULTRA rare est 2,000-10,000

Genetic autosomal dominant

Keratin KRT6A KRT16 KRT17 KRT6B KRT6C

Skin Nails, Feet, Skin, Tongue
• **Genotype** – The underlying genetic cause of PC is a mutation in any one of five keratin genes, *KRT6A, KRT6B, KRT6C, KRT16* or *KRT17*

• **Phenotype** – PC is a syndrome with various overlapping characteristics across five major PC types (PC-K6a, PC-K6b, PC-K6c, PC-K16, PC-K17)
Major Clinical Findings of PC

Thick=(Pachy) o (nychia)=Nails
Major Clinical Findings of PC

Blisters and Calluses

PC = Painful Calluses
Major Clinical Findings of PC

Cysts

PC = Painful Cysts
Major Clinical Findings of PC

Follicular Hyperkeratoses (FHK)
Major Clinical Findings of PC

Leukokeratosis

K6a

K6a

K16

Pachyonychia Congenita Project
My PC Story

• The Burden of a Rare Disease
  • Misdiagnosis and misunderstanding
  • No treatment (unmet medical need)
  • Isolation

• The Burden of PC
  • Pain (limiting activities)
  • Appearance
  • Time for care
My PC Story

• How PC impacts my life.
• The hardest thing about PC.
• The ways I manage PC.
  • No effective treatment for PC.
  • Patients rely on self-care to trim callus, treat blisters, FHK and cysts.
• The difference PC Project has made.