Observations from information in the IPCRR

Patients with mutations in PC-K6a experience painful plantar keratoderma as the most challenging feature of PC and are eager to find relief especially for the plantar keratoderma pain.

- The condition is usually evident at birth usually with 20/20 nail dystrophy.
- PC-K6a nails may be extremely thickened or may not grow out completely.
- Plantar keratoderma and pain is experienced by nearly 100% of PC-K6a patients by age 10. Pain levels for PC-K6a patients are often from 6 to 10 on a 0 to 10 scale.
- Leukokeratosis in oral mucosa is often present very early and PC-K6a infants are often misdiagnosed with thrush.

In children with PC-K6a, there are several unique conditions that occur only for some PC-K6a and not with other types of PC:

1. Infants may have feeding/sucking problems with crying after a few seconds of sucking. This is solved by an enlarged hole in the nipple or use of syringe for feeding, or thickened formula. After initial crying, feeding usually can resume. Some infants undergo efforts by ENTs to remove the leukokeratosis which may make the condition worse and is not the cause of the feeding issue.
2. Follicular hyperkeratosis (FHK) is problematic for children up to age 12 or so and lessens after that time. It occurs at areas of friction on elbows, knees, waist, etc.
3. An extreme pain lasting 15 to 25 seconds occurs with first bite or first swallow which we believe may be related to salivary glands is experienced by children ages 4 to 12 and lessens after that. This is often misdiagnosed as an ‘ear’ problem.