Comments on PC-K6b

Observations from information in the IPCRR

Patients with mutations in PC-K6b have a milder form of PC, but experience painful plantar keratoderma as the most challenging feature.

- The condition is often not evident at birth but onset is before age 15 for 90% of patients with PC-K6b.
- While nearly 100% have some toenail dystrophy, less than 50% have any fingernail dystrophy.
- Oral leukokeratosis is found in only a small percentage of PC-K6b patients and is usually mild.
- Cysts and follicular hyperkeratosis FHK are reported in many patients, although the cysts in PC-K6b are not as extensive as those in PC-K17.
- Plantar keratoderma and pain is experienced by nearly 100% of PC-K6b. Pain levels for PC-K6b patients are often from 3 to 5 on a 0 to 10 scale. This is less than PC-K6a, but significant pain levels.

<table>
<thead>
<tr>
<th>IPCRR Data</th>
<th>PC-K6b N=84</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>82 of 84 (98%)</td>
</tr>
<tr>
<td>Toenails Dystrophy</td>
<td></td>
</tr>
<tr>
<td>all 10 toenails thickened</td>
<td>30 of 84 (36%)</td>
</tr>
<tr>
<td>7-9 toenails thickened</td>
<td>16 of 84 (19%)</td>
</tr>
<tr>
<td>4-6 toenails thickened</td>
<td>26 of 84 (31%)</td>
</tr>
<tr>
<td>1-3 toenails thickened</td>
<td>10 of 84 (12%)</td>
</tr>
<tr>
<td><strong>Toenails - Onset</strong></td>
<td><strong>N = 82</strong></td>
</tr>
<tr>
<td>Birth or less than 1 year</td>
<td>12 of 82 (15%)</td>
</tr>
<tr>
<td>1 to 4 years old</td>
<td>20 of 82 (24%)</td>
</tr>
<tr>
<td>5 to 14 years old</td>
<td>42 of 82 (51%)</td>
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<tr>
<td>15 years and over</td>
<td>9 of 82 (11%)</td>
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<tr>
<td>Fingernail Dystrophy</td>
<td>37 of 84 (44%)</td>
</tr>
<tr>
<td>all 10 fingernails thickened</td>
<td>6 of 84 (07%)</td>
</tr>
<tr>
<td>7-9 fingernails thickened</td>
<td>5 of 84 (06%)</td>
</tr>
<tr>
<td>4-6 fingernails thickened</td>
<td>19 of 84 (23%)</td>
</tr>
<tr>
<td>1-3 fingernails thickened</td>
<td>7 of 84 (08%)</td>
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<tr>
<td><strong>Fingernails - Onset</strong></td>
<td><strong>N = 37</strong></td>
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<tr>
<td>Birth or less than 1 year</td>
<td>5 of 37 (14%)</td>
</tr>
<tr>
<td>1 to 4 years old</td>
<td>8 of 37 (22%)</td>
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<tr>
<td>5 to 14 years old</td>
<td>12 of 37 (32%)</td>
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<tr>
<td>15 years and over</td>
<td>14 of 37 (38%)</td>
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<tr>
<td>Plantar Keratoderma</td>
<td>81 of 84 (96%)</td>
</tr>
<tr>
<td>Always (never goes away)</td>
<td>80 of 84 (95%)</td>
</tr>
<tr>
<td>Sometimes (clear up at times)</td>
<td>1 of 84 (01%)</td>
</tr>
<tr>
<td>Seldom (feet usually clear)</td>
<td>0 of 84 (00%)</td>
</tr>
<tr>
<td><strong>Plantar Keratoderma - Onset</strong></td>
<td><strong>N = 81</strong></td>
</tr>
<tr>
<td>Birth or less than 1 year</td>
<td>4 of 81 (05%)</td>
</tr>
<tr>
<td>1 to 4 years old</td>
<td>27 of 81 (33%)</td>
</tr>
<tr>
<td>5 to 14 years old</td>
<td>48 of 81 (59%)</td>
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<tr>
<td>15 years and over</td>
<td>2 of 81 (02%)</td>
</tr>
<tr>
<td>Plantar Pain</td>
<td>81 of 81 (100%)</td>
</tr>
<tr>
<td>Often require meds for pain</td>
<td>16 of 81 (20%)</td>
</tr>
<tr>
<td>Very painful, but do not use meds</td>
<td>37 of 81 (46%)</td>
</tr>
<tr>
<td>Somewhat painful</td>
<td>28 of 81 (35%)</td>
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<tr>
<td>Palmar Keratoderma</td>
<td>41 of 84 (49%)</td>
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<tr>
<td>Always (never goes away)</td>
<td>18 of 84 (21%)</td>
</tr>
<tr>
<td>Sometimes (clear up at times)</td>
<td>8 of 84 (10%)</td>
</tr>
<tr>
<td>Seldom (hands usually clear)</td>
<td>15 of 84 (18%)</td>
</tr>
<tr>
<td>Other</td>
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<tr>
<td>Oral Leukokeratosis</td>
<td>20 of 84 (24%)</td>
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<tr>
<td>Cysts</td>
<td>56 of 84 (67%)</td>
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<tr>
<td>Follicular Hyperkeratosis</td>
<td>34 of 84 (40%)</td>
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<tr>
<td>Natal or Prenatal Teeth</td>
<td>0 of 84 (00%)</td>
</tr>
</tbody>
</table>