Table SI. AAGAB mutations identified in the current study

<table>
<thead>
<tr>
<th>Nucleotide change</th>
<th>Protein change</th>
<th>Computational analysis</th>
<th>Exon</th>
<th>Protein position</th>
<th>Family (patient)</th>
<th>References</th>
</tr>
</thead>
<tbody>
<tr>
<td>Affecting start codon</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>c.1A&gt;G</td>
<td>p.0?</td>
<td>Probably damaging deleterious</td>
<td>1</td>
<td>N-terminal</td>
<td>PPKP 10 (66651), PPKP 4 (68077, 71005)</td>
<td>Current study</td>
</tr>
<tr>
<td>c.2T&gt;A</td>
<td>p.0?</td>
<td>Probably damaging deleterious</td>
<td>1</td>
<td>N-terminal</td>
<td>PPKP 3 (66350, 66351)</td>
<td>Current study</td>
</tr>
<tr>
<td>c.1-1_12del</td>
<td>p.0?</td>
<td>–1/1</td>
<td>N-terminal</td>
<td>PPKP 2 (66352)</td>
<td>Current study</td>
<td></td>
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<tr>
<td>Missense</td>
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<tr>
<td>c.415G&gt;A</td>
<td>p.Val139lle</td>
<td>Probably damaging tolerated</td>
<td>4</td>
<td>Between Rab-like GTPase and adaptin-binding domain</td>
<td>PPKP 6 (69352, 71033, 71034, 71035, 76427)</td>
<td>Current study</td>
</tr>
<tr>
<td>Frameshift</td>
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<td></td>
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<tr>
<td>c.77delT</td>
<td>p.Ile26Thrfs*11</td>
<td>Probably damaging tolerated</td>
<td>2</td>
<td>N-terminal</td>
<td>PPKP 1 (65325, 65604, 66550, 72382)</td>
<td>Current study</td>
</tr>
<tr>
<td>c.506_507insAA</td>
<td>p.Asn169Lysfs*7</td>
<td></td>
<td>2</td>
<td>N-terminal</td>
<td>PPKP 1 (65325, 65604, 66550, 72382)</td>
<td>Current study</td>
</tr>
<tr>
<td>c.17_73+17delCCT(68bp)GCC</td>
<td>p.?</td>
<td>Exon skipping</td>
<td>1/+1</td>
<td>N-terminal</td>
<td>PPKP 14 (78323)</td>
<td>Current study</td>
</tr>
<tr>
<td>Nonsense</td>
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<td>c.512G&gt;A</td>
<td>p.Trp171*</td>
<td>Exon skipping</td>
<td>1</td>
<td>N-terminal</td>
<td>PPKP 11 (73417)</td>
<td>Current study</td>
</tr>
<tr>
<td>c.370C&gt;T</td>
<td>p.Arg124*</td>
<td>Exon skipping</td>
<td>1</td>
<td>Between Rab-like GTPase and adaptin-binding domain</td>
<td>PPKP 5 (69783, 70445)</td>
<td>Giehl et al. 2012 (1)</td>
</tr>
<tr>
<td>c.370C&gt;T</td>
<td>p.Arg124*</td>
<td>Exon skipping</td>
<td>1</td>
<td>Between Rab-like GTPase and adaptin-binding domain</td>
<td>PPKP 5 (69783, 70445)</td>
<td>Pohler et al. 2012 (12)</td>
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<tr>
<td>Splice-site</td>
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<tr>
<td>c.73+1G&gt;T</td>
<td>p.?</td>
<td>Exon skipping</td>
<td>+1</td>
<td>N-terminal</td>
<td>PPKP 8 (70562, 735578)</td>
<td>Current study</td>
</tr>
<tr>
<td>c.451+1G&gt;A</td>
<td>p.?</td>
<td>Exon skipping</td>
<td>+4</td>
<td>Between Rab-like GTPase and adaptin-binding domain</td>
<td>PPKP 7 (69354)</td>
<td>Eytan et al. 2014</td>
</tr>
</tbody>
</table>

PPKP: punctate palmoplantar keratoderma.