Title: Hypokalemic Periodic Paralysis  
Date: July 2014  
Note: The following information is provided by the authors listed above and has not been reviewed by GeneReviews staff.

Table 2. Summary of Mutations Detection Frequencies by Method for Molecular Genetic Testing of Hypokalemic Periodic Paralysis

<table>
<thead>
<tr>
<th>Gene</th>
<th>Test Method</th>
<th>Mutations Detected</th>
<th>Mutation Detection Frequency by Mutation and Test Method</th>
</tr>
</thead>
<tbody>
<tr>
<td>CACNA1S</td>
<td>Sequence analysis of select exons (e.g. exons 4, 11, 21 and 30 encoding S4 helixes)</td>
<td>See CACNA1S above</td>
<td>~60%</td>
</tr>
<tr>
<td>SCN4A</td>
<td>Sequence analysis of select exons (e.g. exons 5, 12, 13, 18 and 24 encoding S4 helixes)</td>
<td>See SCN4A above; Also detects mutations associated with other types of periodic paralysis associated with SCN4A (HYPP, NormoPP)</td>
<td>~20%</td>
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</tbody>
</table>
| KCNJ18   | Targeted mutation analysis | c.127C>T p.Arg43Cys  
c.419C>T p.Thr140Met  
c.428delC p.Ile144SerfsTer8  
c.598G>C p.Ala200Pro  
c.1195C>T p.Arg399Ter  
c.1219C>T p.Gln407Ter  
c.1061C>T p.Thr354Met  
c.1097A>G p.Lys366Arg | 1/60 cases (s-HOKPP)  
1/30 cases (TPP)  
1/30 cases (TPP)  
2/120 cases (TPP)  
1/60 cases (s-HOKPP)  
1/30 cases (TPP)  
5/30 cases (TPP)  
1/30 cases (TPP)  
1/30 cases (TPP) |
| KCNJ18   | Sequence analysis      | See KCNJ18 above                                                                  | Between 1.5 and 33% (TPP)  
3.5% (s-HOKPP) |

s-HOKPP = sporadic hypokalemic periodic paralysis  
TPP = thyrotoxic hypokalemic periodic paralysis