Title: Hypokalemic Periodic Paralysis *GeneReview* Table 2

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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

Gene	Test Method	Mutations Detected	Mutation Detection Frequency by Mutation and Test Method
CACNA1S	Targeted mutation analysis	p.Arg528His p.Arg528Gly p.Arg897Ser p.Val876Glu p.Arg900Ser p.Arg900Gly p.His916Gln p.Arg1239His p.Arg1239Gly	~35% <1% <1% private (one family) <1% <1% private (one family) ~20% <2%
SCN4A	Targeted mutation analysis	p.Arg222Trp p.Arg669His p.Arg672His p.Arg672Gly p.Arg672Ser p.Arg672Cys p.Arg1129Gln p.Arg1132Gln p.Arg1135His	<1% <2% ~10% <2% <1% <1% private (one family) <2% <2%
CACNA1S	Sequence analysis of select exons (e.g. exons 4, 11, 21 and 30 encoding S4 helixes)	See *CACNA1S* above	~60%
SCN4A	Sequence analysis of select exons (e.g. exons 5, 12, 13, 18 and 24 encoding S4 helixes)	See *SCNA4* above; Also detects mutations associated with other types of periodic paralysis associated with *SCN4A* (HYPP, NormoPP)	~20%

Gene	Test Method	Mutations Detected	Mutation Detection Frequency by Mutation and Test Method
CACNA1S	Sequence analysis	See *CACNA1S* above	~60%
SCN4A	Sequence analysis	See *SCNA4* above; Also detects mutations associated with other types of periodic paralysis associated with *SCN4A* (HYPP, NormoPP)	~20%
KCNJ18	Targeted mutation analysis	c.127C>T p.Arg43Cys c.419C>T p.Thr140Met c.428delC p.Ile144SerfsTer8 c.502G>A p.Val168Met 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