Title: McLeod Neuroacanthocytosis Syndrome *GeneReview* Table 3
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 3. *XK* Mutations

Exon	DNA Change	Protein Change	Reference
Whole gene	Major deletion	Absence of XK protein	Ho et al [1994], El Nemer et al [2000], Danek et al [2001a], Danek et al [2001b], Peng et al [2007]
Promotor + exon 1	Major deletion	Absence of XK protein	Ho et al [1994]
Promotor + exon 1	Major deletion	Absence of XK protein	Wendel et al [2004]
1	Major deletion	Absence of XK protein	Danek et al [2001a]
Exons 1 + 2	Major deletion	Absence of XK protein	Walker et al [2006]
1	254delG	V58YfsX71	Zeman et al [2005]
1	189G>A	W36X	Malandrini et al [1994], Danek et al [2001a]
IVS1	IVS1+1G>C	Splice-site mutation	Russo et al [2002]
2	7453 bp major deletion	Unknown	Singleton et al [2003]
2	350delT [1]	Y90TfsX39	Ho et al [1996]
2	479C>T	R133X	Dotti et al [2000], Danek et al [2001a]
2	533_534insC	Q151PfsX47	Ueyama et al [2000], Starling et al [2005]
2	545C>T	Q155X	Danek et al [2001a]
IVS2	IVS2+1G>A [2]	Splice-site mutation	Swash et al [1983], Ho et al [1994]
IVS2	IVS2+5G>A	Splice-site mutation	Daniels et al [1996]
IVS2	IVS2-1G>A [3]	Splice-site mutation	Swash et al [1983], Ho et al [1994]
3	746C>G	R222G	Russo et al [2002]
3	768_769delTT	F229YfsX35	Danek et al [2001a]
3	789G>A	W236X	Danek et al [2001a]

Exon	DNA Change	Protein Change	Reference
3	853delG	W257CfsX10	Danek et al [2001a]
3	938_942delCTCTA	L286YfsX15	Danek et al [2001a]
3	962T>C [4]	C294R	Danek et al [2001a]
3	977C>T [5]	Q299X	Jung et al [2001]
3	1020_1033del [6]	N313TfsX23	Allen et al [1961], Danek et al [2001a]
3	1023G>A [7]	W314X	Supple et al [2001]
3	1061G>A	E327K	Jung et al [2003]
3	1095delT [8]	F338SfsX70	Hanaoka et al [1999]

Notes:

A. The numbering of the cDNA starts from the first base in the GenBank entry Z32684, i.e. the A of the initiation codon is assigned as base 83.

B. For the frameshift mutation, the following nomenclature was used: the first amino acid change_fsX_the length of the shifted reading frame (www.genomic.unimelb.edu.au/mdi/mutnomen/recs.html#protein).

1. OMIM allelic variant 314850.003

2. OMIM allelic variant 314850.001

3. OMIM allelic variant 314850.002

4. OMIM allelic variant 314850.005

5. OMIM allelic variant 314850.008

6. OMIM allelic variant 314850.006

7. OMIM allelic variant 314850.007

8. OMIM allelic variant 314850.004

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