

Title: Hyperekplexia *GeneReview* Tables 3-5

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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. Hyperekplexia pathologic allelic variants/*GLRB*

Sequence change	Mutation	Reference
G920A	G229D	Rees et al 2002
IVS 5+5 G>A	IVS5+5G-A	

Table 4. Hyperekplexia pathologic allelic variants/*SLC6A5*

Sequence change	Mutation	Reference
delC{319-324}	P108L fs 25	Rees et al 2006
C916G	L306V	
C1131A	Y377	
C1274T	T425M	
G1294T / Ins[T]1295	V432F/ fs 97	
T1444C	W482R	
A1472G	Y491C	
A1526G	N509S	
C1274T	S510R	
C1888T	Q630X	

Table 5. Hyperekplexia pathologic allelic variants/*GPHN*

Sequence change	Mutation	Reference
A28T	N10Y	Rees et al 2003