Note: The following information is provided by the author(s) and has not been reviewed by *GeneReviews* staff.

Table 2. Selected Mutations in the *SLC2A1* Gene Causing Glut1-DS

Number of Individuals	Nucleotide Change	Location	Amino Acid Change	Mutation Type
1	102A>T	Exon 2	p.N34I	Missense
1	376C>T	Exon 3	p.S66F	
3	G272A	Exon 4	p.G91D	
1	279G>A	Exon 4	p.R93Q	
1	556G>T	Exon 4	p.R126L	
5	556G>A	Exon 4	p.R126H	
2	555C>T	Exon 4	p.R126C	
1	567G>A	Exon 4	p.G130S	
1	615G>A	Exon 4	p.E146K	
1	920G>C	Exon 6	p.E247D	
1	945A> G946A>T	Exon 6	p.K256V	
1	1063C>T	Exon 7	p.T295M	
1	1108C>T	Exon 7	p.T310I	
3	1176C>G	Exon 8	p.R333W	
2	1167C>T	Exon 7	p.R330X [1]	Nonsense
1	1368C>T	Exon 9	p.Q379X	
1	1526C>A	Exon 10	p.Y449X	
1	1545A>T	Exon 10	p.K456X	
1	266delc, 267A>T	Exon 2	FS [2] >STOP	Deletion
1	368-369ins23	Exon 3	FS>STOP	Insertion
2	684del3	Exon 5	FS>STOP	Deletion
1	741-742insC	Exon 5	FS>STOP	Insertion
1	888-889insG	Exon 6	FS>STOP	Deletion
1	904delA	Exon 6	FS>STOP	Deletion

Number of Individuals	Nucleotide Change	Location	Amino Acid Change	Mutation Type
1	102A>T	Exon 2	p.N34I	Missense
1	376C>T	Exon 3	p.S66F	
3	G272A	Exon 4	p.G91D	
1	279G>A	Exon 4	p.R93Q	
1	556G>T	Exon 4	p.R126L	
5	556G>A	Exon 4	p.R126H	
2	555C>T	Exon 4	p.R126C	
1	567G>A	Exon 4	p.G130S	
1	615G>A	Exon 4	p.E146K	
1	920G>C	Exon 6	p.E247D	
1	945A> G946A>T	Exon 6	p.K256V	
1	1063C>T	Exon 7	p.T295M	
1	1108C>T	Exon 7	p.T310I	
3	1176C>G	Exon 8	p.R333W	
2	969delC	Exon 6	FS>STOP	Deletion
1	1086delG	Exon 7	FS>STOP	Deletion
1	1159del2	Exon 8	FS>STOP	Deletion
1	1377del1	Exon 9	FS>STOP	Deletion
1	1562ins4	Exon 9	FS>STOP	Insertion
3	197+1G>A	Intron 1		Splice site
1	857T>G 858G>A 858+1del10	Intron 5		
1	1151+1G>T	Intron 7		
3	Large scale deletion			Hemizygosity

1. Termination codon (X)
2. Frameshift (FS) leading to a nonsense (STOP) codon, thereby predicting a premature termination of the polypeptide [Seidner et al 1998, Wang et al 2000, Klepper et al 2005, Wang et al 2005]