

Supplemental Table 1

Missense Mutations Analyzed

exon1	M1L	G196V	A356V	K478E	exon15	G613S	V677M	exon23	R904Q
	T60M	exon5 R209W	exon9 G374V	exon12 D486N		S615L	V677L	exon24	R919C
	D62N	L215P	exon10 R399C	G496C		S615W	exon18 A728T	exon25	R955Q
	E68K	A226P	S402F	A523T		L623P	G731R		R958G
	H69N	G230D	N406H	exon13 F535L		G630V	L738R		S967F
	R83W	V242A	C421R	L542P		R642C	G741R	exon26	G980R
exon3	R145H	exon6 R261C	N426K	S555L		R642H	exon22 L849H		C985Y
	V153M	G264R	C430G	P560H		R642G	L850P		R1009Q
	R158Q	exon7 T304P	G439S	exon14 A569V	exon16	V647M	R852C		N1014K
	T163M	S314F	G439V	A569E		T649R	R852S		V1015M
exon4	W172R	G316V	N442K	L571P		R655H	R852H		
	T180K	exon8 G342A	G463E	V578M		R655C	G867S		
	G186D	P349L	A464T	A588V		M672I	R871H		

Supplemental Table 2

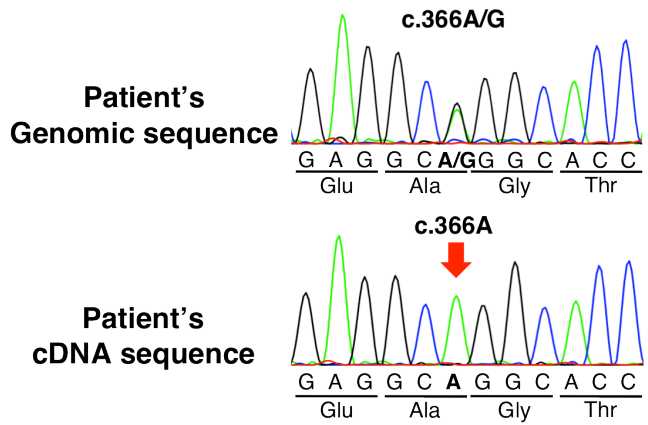
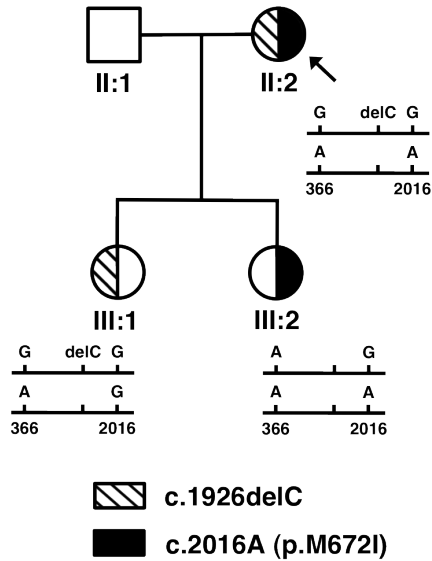
SLC12A3 Splice Site Strength*

Intron	5' donor site	3' acceptor site
6	1	0.86
7	0.98	<u>0.33</u>
8	1	0.98
9	0.99	0.99
10	0.88	0.19
11	0.99	0.09
12	1	0.01
13	0.99	0.12
14	0.96	0.82
15	0.99	0.96
16	<u>0.67</u>	0.97
17	0.54	0.98
18	0.91	0.98
19	0.71	0.97

*Scores were determined using the BDGP splicing predictor program (http://www.fruitfly.org/seq_tools/splice.html)

Supplemental Figure 1

Heterozygous Deletion in cDNA



Supplemental Table 3 Primers List

	Forward primer	Reverse primer
<For H492>		
exon8	5'-GATAGCTAGCGGACTCTACTACCCAGGAATC-3'	5'-GATAGGATCCAACAACCTCAGAACGGGAATG-3'
exon10	5'-GATAGCTAGCGGACACACCTTCATTGTCATC-3'	5'-GATAGGATCCTGGCACTTCATCCCTACTG-3'
exon16	5'-GATAGTAGCCCCCTGGATTATAGGTGGGAAG-3'	5'-GATAGGATCCGTCTCAAACCTCCTGACCTCG-3'
<For uptake assay>		
	5'-GATAGGATCCATGGCAGAAGTCCCAACACAG -3'	5'-GATAGCGGCCGCTTACTGGCAGTAAAAAGGTG-3'
<For semi-nested PCR>		
first	5'-TCCTCCTGCTCTATGTCATCTAC-3'	5'-CAGTTCTTCTTGAACCCCAACCAC-3'
second	5'-GCCTTGATCTTCCTCTTGTTCAG-3'	(-)