Table S2. Segregating, rare variants discovered in the affected individuals using whole-exome sequencing.

Family	Chr	Position	Ref Allele	Mut Allele	Gene	Function	AA Change	AA Position	Conservation	Allele Frequency	phast Cons	GERP
Family 1426												
	16	87874689	G	A	*SLC7A5*	missense	ALA/VAL	246/508	AAAAAAA-AAAAAAA-AA-AAAAAAAA-AA-A-AAAAA-AAAAAAA	0.001	1	4.72
Family 1465												
	2	111542319	CA	C	ACOXL	frameshift	T29	29/580	TTTTTTTT-TTT-TT-TRTRRTTTT-KTKM-TKK----------	0.0004	0.261	0.185
	16	89986498	A	G	MC1R TUBB3	missense	K278E	278/317	KK-KKKKK-/-KKQK-RK/KQKKQK-KK-KK-K-QQ/SSSNYRSRRR-/--	0.013	0.93	-0.119
	22	25131681	G	T	PIWIL3	missense	A543E	543/882	AAAAAA---/PAAG-----/-A---A---PA-------/---AA-A-AA-/PG	0.0064	0.004	0.716
	22	24226213	T	C	SLC2A11,	splice_donor	R259Q	259/496	RX-R--WLW/R--R-RWL/-R--RRR-Q-RR-R---/-----KKKKKE-/--	0.0026	1	1.57
	16	87871467	G	A	SLC7A5	missense	P375L	375/508	PPPPP--P-/--PP---P/PPPPPPPPP-PP-P-PP/PPP-PPPPPPP/PP	0.0003	1	4.79