Gene	OMIM	Inheritence	Pt. Zygosity	Position	Nucleotide	Amino Acid	Parental Studies	Comment
DNM1L	614388	AD	Het	Chr12:32875536	c.1048G>A	p.G350R	Mother 6-8% mosaicism	Novel variant
ACADSB	610006	AR	Het	Chr10:124812677	c.1228+1G>A	N/A	Father heterozygous	One variant detected (AR disorder)
ACSF3	614265	AR	Het	Chr16:89199607	c.1303G>T	p.E435X	Father heterozygous	One variant detected (AR disorder)
CLN8	600143	AR	Het	Chr8:1728557	c.685C>G	p.P229A	ND	One variant detected (AR disorder)
COQ9	614654	AR	Het	Chr16:57485113	c.235C>T	p.P79S	ND	One variant detected (AR disorder)
AHI1	608629	AR	Het	Chr6:135778794	c.989A>G	p.D330G	ND	One variant detected (AR disorder)
DHFR	613839	AR	Het	Chr5:79950481	c.68C>T	p.S23F	ND	One variant detected (AR disorder)
SLC25A3	610773	AR	Het	Chr12:98987859	c.103G>C	p.G35R	ND	One variant detected (AR disorder)
CHKB	602541	AR	Het	Chr22:51021197	c.14C>T	p.A5V	ND	One variant detected (AR disorder)
SLC6A3	613135	AR	Het	Chr5:1403128	c.1676C>T	p.A559V	ND	One variant detected (AR disorder)
PEX1	214100	AR	Het	Chr8:92129015	c.2718+3G>A	N/A	ND	One variant detected (AR disorder)
ALG13	601110	XL	Hem	ChrX:110970229	c.1922A>G	p.H641R	Mother heterozygous	Patient had normal N-glycan analysis
MECP2	300672	XL	Hem	ChrX:153296146	c.1133C>T	p.A378V	Mother heterozygous	Most variants X-linked dominant; Male with same variant seen in ExAC data; "benign clinical significance in ClinVar; rs201314910