Gene	OMIM	Inheritence	Pt. Zygosity	Position	Nucleotide	Amino Acid	Parental Studies	Comment
DNM1L	614388	AD	Het	Chr12:32884003	c.1135G>A	p.E379K	*De novo*	Novel variant
PDHA1	312170; 308930	XL	Het	ChrX:19371229	c.448G>A	p.G150R	*De novo*	Novel variant
SMARCA2	601358	AD	Het	Chr9:2161785	c.4081G>Y	p.V1361I	Mother heterozygous	Mother also heterozygous
FARS2	614946	AR	Het	Chr6:5369309	c.506A>T	p.D169V	ND	One variant detected (AR disorder); variant seen in 15/3723 European American and 0/7020 African American in ESP5400 data.
C12orf65	613559; 615035	AR	Het	Chr12:123738477	c.256C>A	p.H86N	ND	One variant detected (AR disorder)
ICK	612651	AR	Het	Chr6:52878698	c.914A>C	p.K305T	ND	One variant detected (AR disorder)
GAA	232300	AR	Het	Chr17:78078606	c.221G>A	p.R74H	ND	One variant detected (AR disorder)
NDUFS1	252010	AR	Het	Chr2:207003310	c.1291C>G	p.L431V	ND	One variant detected (AR disorder); variant seen in 3/3735 European American and 39/6981 African American in ESP5400