

ULTRARARE NEONATAL EPILEPSIES IN OUR COHORT

Patient ID	Gene	Inheritance	Zigosity	Variant description	Type	Origin	Variant classification	Reported	Testing method
1	STAMBP	AR	Compound heterozygous	c.867G>A, p.(Leu289Leu) c.946G>A, p.(Glu316Lys)	Synonymous substitution in a highly conserved nucleotide Missense	Father Mother	Class 3 Class 3	Novel rs1340000256	WES
2	HPDL	AR	Compound heterozygous	c.1024C>T, p.(Gln342Ter) c.155C>A, p.(Ala52Glu)	Nonsense Missense	Father Mother	Class 4 Class 4	rs2139621075 Novel	Targeted Sanger sequencing
3	CPS1	AR	Compound heterozygous	c.2403delT, p.(Ile801Metfs*20) c.3069C>G, p.(Asp1023Glu)	Nonsense Missense	Mother Father	Class 5 Class 4	Novel rs1559123272	WES
4	BRAT1	AR	Compound heterozygous	c.638dup p.(Val214Glyfs*189) c.2125_2128del p.(Phe709Thrfs*17)	Nonsense Nonsense	Mother Father	Class 5 Class 5	rs730880324 rs763527391	Single gene filtered NGS
5	BRAT1	AR	Compound heterozygous	c.638dup p.(Val214Glyfs*189) c.2005C>T p.(Arg669Trp)	Nonsense Missense	Mother Father	Class 5 Class 4	rs730880324 rs35045151	Single gene filtered NGS
6	ALDH18A1	AR	Homozygous	c.1923+1G>A c.1923+1G>A	Nonsense Nonsense	Mother Father	Class 5 Class 5	rs86223315 rs86223315	WES
7	PHF6	X-linked	Hemizygous	c.729G>A p.(Met243Ile)	Missense	De novo	Class 4	Novel	WES
8	KCNT1	AD	Heterozygous	c.2800G>A p.(Ala934Thr)	Missense	De novo	Class 5	rs397515403	Epilepsy panel
9	KCNT1	AD	Heterozygous	c.2798G>C p.(Arg933Pro)	Missense	Father (mosaic 10%)	Class 3	Novel	Epilepsy panel
10	FGF12	AD	Heterozygous	c.341G>A p.(Arg114His)	Missense	De novo	Class 5	rs886039903	Epilepsy panel
11	SCN3A	AD	Heterozygous	c.1592G>C p.(Ser531Thr)	Missense	Father	Class 3	rs1559229929	Epilepsy panel