**Table S2: Genotype of patients diagnosed by WES (n= 28)**

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| --- | --- | --- | --- | --- | --- | --- |
| **ID** | **Genetic finding** | **Variant(s)** | **Variant type** | **Zygosity** | **Inheritance** | **ACMG classification** |
| 2 | *GRIN2B* (NM\_000834.3) | c.1931C>A [p.(Ala644Asp)] | Missense | Heterozygous | *De novo*, AD | P |
| 4 | *KCNQ2* (NM\_004518.4) | c.716G>T [p.(Gly239Val)] | Missense | Heterozygous | *De novo*  | P |
| 5 | *ACTG2* (NM\_001615.3) | c.118C>T [p.(Arg40Cys)] | Missense | Heterozygous | *De novo*  | P |
| 6 | *EFTUD2* (NM\_001258353.1) | c.446T>A [p.(Leu149\*)] | Nonsense | Heterozygous | *De novo*  | P |
| 7 | *GLDC* (NM\_000170.2) | [Ex22-25](http://genome-euro.ucsc.edu/cgi-bin/hgTracks?db=hg19&highlight=hg19.chr9:6533016-6540146&position=chr9:6533016-6540146) deletion  | Deletion | Homozygous | AR | P |
| 8 | *ASNS* (NM\_001673.4) | c.1210C>T [p.(Arg404Cys)], mat.c.180G>C [p.(Gln60His)], pat. | Missense | Compoundheterozygous | AR | LP / LP |
| 10 | *TPM1* (NM\_000366.5)*SNAP25* (NM\_003081.3) | c.80\_97dup [p.(Ala27\_Ala32dup)]c.181G>A [p.(Glu61Lys)] | Inframe duplicationMissense | HeterozygousHeterozygous | *De novo**De novo* | LPVUS |
| 13 | *SCN2B* (NM\_004588.4) | c.194T>C [p.(Leu65Pro)] | Missense | Heterozygous | AD, maternal inherited | VUS |
| 17 | *TNNT3* (NM\_006757.3) | c.116C>G [p.(Pro39Arg)] | Missense | Heterozygous | *De novo* | LP |
| 19 | *MAST1* (NM\_014975.2) | c.669\_689del [p.(Gln223\_Asp230delinsHis)] | Inframe deletion | Heterozygous | *De novo* | LP  |
| 23 | *TPO* (NM\_000547.5) | c.1618C>T [p.(Arg540\*)] | Nonsense | Homozygous | AR | P |
| 24 | *CLCNKB* (NM\_000085.4) | Ex2-20 del | Deletion | Homozygous | AR | P |
| 25 | *FLNA* (NM\_001456.3) | c.3293G>A [p.(Gly1098Asp)] | Missense | Hemizygous | XLD, maternal | VUS |
| 26 | Del 16p11.2 | chr16:g.30,364,306\_31,372,533 Del | Deletion | Heterozygous | unknown  | VUS \* |
| 28 | *C12orf65* (NM\_152269.4) | c.62G>A [p.(Trp21\*)] | Nonsense | Homozygous | AR | P |
| 29 | Del 1q21 | chr1:g.146,491,108\_147,408,799 Del | Deletion | Heterozygous | maternal | VUS \* |
| 30 | *POU1F1* (NM\_000306.2) | c.514C>T [p.(Arg172\*)] | Nonsense | Homozygous | AR, maternal isodisomy | P |
| 31 | *PTPN11* (NM\_002834.3) | c.923A>G [p.(Asn308Ser)] | Missense | Heterozygous | *De novo* | P |
| 36 | *KMT2D* (NM\_003482.3) | c.6025delG [p.(Asp2009Metfs\*38)] | Frameshift | Heterozygous | *De novo* | P |
| 42 | *EFL1* (NM\_024580.5) | c.904T>C [p.(Trp302Arg)] | Missense | Homozygous | AR | LP |
| 43 | *PNKP* (NM\_007254.3) | c.1253\_1269dup [p.(Thr424Glyfs\*49)], mat.c.143delG[p.(Arg48Lysfs\*17)], pat. | FrameshiftFrameshift | Compound heterozygous | AR | P / P |
| 46 | *TUBA1A* (NM\_006009.3) | c.1142C>T [p.(Thr381Ile)] | Missense | Heterozygous | *De novo* | P |
| 49 | *PTPN11* (NM\_002834.3) | c.1528C>G [p.(Gln510Glu)] | Missense | Heterozygous | *De novo*  | P |
| 55 | *TBX5* (NM\_000192.3) | c.1348delC [p.Leu450Trpfs\*132)] | Frameshift | Heterozygous | *De novo*  | P |
| 56 | *CHD7* (NM\_017780.3) | c.6579delA [p.(Glu2194Lysfs\*21)] | Frameshift | Heterozygous | *De novo*  | P |
| 58 | *CACNA1C* (NM\_199460.5) | c.4769G>A [p.(Arg1590His)] | Missense | Heterozygous | *De novo*  | LP  |
| 60 | *SLC5A7* (NM\_021815.4) | c.282T>A [p.(Ser94Arg)] | Missense | Homozygous | AR | LP  |
| 61 | *TFAP2A* (NM\_001042425.2) | c.773G>A [p.(Gly258Glu)] | Missense | Heterozygous | *De novo* | LP  |

Abbreviations: AR, autosomal-recessive; AD, autosomal-dominant; XLD, X-linked dominant; CNV, Copy number variation; LP, likely pathogenic; mat., maternal; P, pathogenic; pat., paternal; VUS, variant of uncertain significance