|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Mutation type** | **Exon** | **Gene variant** | **GRCh38** | **ExAC AF** | **1000G MAF** | **dbSNP** | **Protein consequence** | **Sex** | **CDKL5 clinical diagnosis** |
| Frameshift | 4 | c.106\_107delCA | 18564483 | - | - | - | p.His36fs | Male | Yes |
| Frameshift | 5 | c.163\_166delGAAA | 18575371 | - | - | - | p.Glu55fs | Female | Yes |
| Frameshift | 5 | c.200\_201delTC | 18575408 | - | - | - | p.Leu67fs | Female | Yes |
| Frameshift | 7 | c.435delC | 18581922 | - | - | - | p.His145fs | Female | Yes |
| Frameshift | 9 | c.665delC | 18588064 | - | - | - | p.Thr222fs | Female | Yes |
| Frameshift | 9 | c.678-691del14ins11 | 18588077 | - | - | - | p.Val226fs | Female | Yes |
| Frameshift | 9 | c.691\_692delCCinsC | 18588090 | - | - | - | p.Pro231fs | Female | Yes |
| Frameshift | 11 | c.859\_868del10 | 18598495 | - | - | - | p.Leu287fs | Female | Yes |
| Frameshift | 12 | c.1111delC | 18604035 | - | - | - | p.Leu371fs | Female | Yes |
| Frameshift | 12 | c.1268\_1274delCAAAGCC | 18604192 | - | - | - | p.Pro423fs | Female | Yes |
| Frameshift | 12 | c.1371\_1372dupA | 18604295 | - | - | - | p.Thr457fs | Female | Yes |
| Frameshift | 12 | c.1470\_1471delGG | 18604394 | - | - | - | p.Gly490fs | Female | Yes |
| Frameshift | 12 | c.1756\_1759delTCTG | 18604680 | - | - | - | p.Ser586fs | Female | Yes |
| Frameshift | 15 | c.2177\_2186delinsAATGTGTCAAC | 18613176 | - | - | - | p.Ser726Ter | Male | Yes |
| Frameshift | 16 | c.2317delG | 18619907 | - | - | - | p.Glu773fs | Female | Yes |
| Frameshift | 19 | c.2610delC | 18628484 | - | - | - | p.Ser870fs | Female | Yes |
| Frameshift | 19 | c.2663delA | 18628537 | - | - | - | p.Asn888fs | Female | Yes |

**Table e-1. Novel *CDKL5* frameshift variants in our clinical cohorts.** GRCh38 indicates the X-chromosome co-ordinate in the GRCh38/hg38 assembly. Frequencies of variants in population databases are shown as ExAC allele frequency (AF) and 1000G minor allele frequency (MAF). CDKL5 clinical diagnosis was made by the clinical centre the patient presented to.