Supplementary table 3. Recurrent *PURA* variants

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| Variant | Seen in n patients (%) | Phenotype |
| p.(Gly53Profs\*) / p.(Gly53Alafs\*) | 3 (2.1%) | Epilepsy, non-verbal, hypotonia, nystagmus |
| p.(Leu54Alafs\*) / p.(Leu54Cysfs\*) | 6 (4.2%) | Epilepsy, no epilepsy, stereotypic hand movements, feeding difficulties |
| p.(Glu69\*) | 2 (1.4%) | No epilepsy |
| p.(Tyr74His) | 2 (1.4%) | No epilepsy, exaggerated startle response |
| p.(Lys97Glu) | 3 (2.1%) | Epilepsy, no epilepsy, hypotonia, non-verbal |
| p.(Leu100Pro) / p.(Leu100Arg) | 2 (1.4%) | Epilepsy, non-verbal, non-ambulatory |
| p.(Tyr121\*) | 2 (1.4%) | Epilepsy, no epilepsy, non-ambulatory |
| p.(Glu128\*) / p.(Glu128fs\*) | 3 (2.1%) | Epilepsy, no epilepsy, hypotonia |
| p.(Lys144Glu) | 2 (1.4%) | No epilepsy, hypotonia, movement disorder |
| p.(Met157Lys) / p.(Met157Arg) | 2 (1.4%) | Epilepsy, no epilepsy |
| p.(Leu160\*) / p.(Leu160Glu) | 2 (1.4%) | No epilepsy, hypotonia |
| p.(Ile188Thr) / p.(Ile188Ser) | 3 (2.1%) | Epilepsy, no epilepsy, hypotonia |
| p.(Ala189Cysfs\*) / p.(Ala189Pro) | 2 (1.4%) | Epilepsy, CVI |
| p.(Ile196fs\*) / p.(Ile196del) | 2 (1.4%) | Epilepsy, hypotonia, non-verbal |
| p.(Ile206Phe) / p.(Ile206del) | 2 (1.4%) | Epilepsy, no epilepsy, movement disorder |
| p.(Val226Glyfs\*) / p.(Val226Serfs\*) | 4 (2.8%) | Epilepsy, no epilepsy, walking, non-verbal |
| p.(Phe233del) | 14 (9.9%) | Epilepsy, no epilepsy, hypotonia, broad-based gait |
| p.(Arg245\*) / p.(Arg245Pro) | 4 (2.8%) | Epilepsy, no epilepsy, hypotonia, unsteady gait |
| p.(Phe271del) | 4 (2.8%) | Epilepsy, hypotonia, SUDEP |