**Supplementary Table 1**

**Potassium channel gene mutations reported to associate with global developmental delay/intellectual disability**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNN3*  | ZLS | 602983 | S436C | Missense | GOF | Moderate ID | Bauer et al., 2019 |
| *KCNN3* | ZLS | 602983 | K269E  | Missense | GOF | GDD | Bauer et al., 2019 |
| *KCNN3* | ZLS | 602983 | G350D | Missense | GOF | GDD | Bauer et al., 2019 |
| *KCNJ10* | ID, EP and ASD  | 602208 | R18Q | Missense | GOF | Moderate ID  | Sicca et al., 2011 |
| *KCNJ10* | ID, EP and ASD | 602208 | R18Q  | Missense | GOF | Mild ID | Sicca et al., 2011 |
| *KCNJ10* | ID, EP and ASD | 602208 | V84M | Missense | GOF | Severe ID | Sicca et al., 2011 |
| *KCNJ10* | EP and ID | 602208 | R65C and F119GfsX25 | Missense and frameshift | Unknown | Mild ID | Papavasiliou et al., 2017 |
| *KCNJ10* | EP and ID | 602208 | I60T  | Missense | Unknown | Moderate ID | Al Dhaibani et al., 2018 |
| *KCNJ10* | EP and ID | 602208 | I60T | Missense | Unknown | Moderate ID | Al Dhaibani et al., 2018 |
| *KCNJ10* | EAST syndrome | 602208 | N232Qfs\*14  | Nonsense | LOF | Moderate ID | Severino et al., 2018 |
| *KCNJ10* | EAST syndrome | 602208 | G275Vfs\*7 | Nonsense | LOF | Moderate ID | Severino et al., 2018 |
| *KCNJ10* | SeSAME syndrome | 602208 | R65P and R199\* | Compound missense and nonsense | LOF | Mild ID | Scholl et al., 2009 |
| *KCNJ10* | SeSAME syndrome | 602208 | C140R  | Missense | LOF | Mild ID | Scholl et al., 2009 |
| *KCNJ10* | SeSAME syndrome | 602208 | T164I | Missense | LOF | Mild ID | Scholl et al., 2009 |
| *KCNJ10* | SeSAME syndrome | 602208 | A167V and R297C | Compound missense | LOF | Mild ID | Scholl et al., 2009 |
| *KCNJ10* | ID and EP, truncal ataxia and dysmorphic features and electrolyte imbalance | 602208 | R204H | Missense | Unknown | Severe ID | Kara et al., 2013 |
| *KCNA2* | Severe GDD and EP | 176262 | T374A | Missense | LOF | Severe GDD | Hundallah et al., 2016 |
| *KCNA2* | Hereditary spastic paraplegia, ID and ataxia. | 176262 | R294H | Missense  | LOF | Mild ID | Helbig et al., 2016 |
| *KCNA2* | Hereditary spastic paraplegia, ID and ataxia. | 176262 | R294H | Missense | LOF | Mild ID | Helbig et al., 2016 |
| *KCNA2* | Hereditary spastic paraplegia, ID and ataxia. | 176262 | R294H | Missense | LOF | Mild ID | Helbig et al., 2016 |
| *KCNA2* | Hereditary spastic paraplegia, ID and ataxia. | 176262 | R294H | Missense | LOF | Mild ID | Helbig et al., 2016 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNA2* | Hereditary spastic paraplegia, ID and ataxia. | 176262 | R294H | Missense | LOF | Mild ID | Helbig et al., 2016 |
| *KCNA2* | EP and ID | 176262 | P405L | Nonsense | LOF | Moderate ID | Syrbe et al., 2015 |
| *KCNA2* | EP and ID | 176262 | I263T | Nonsense | LOF | Moderate ID | Syrbe et al., 2015 |
| *KCNA2* | EP and ID | 176262 | P405L | Nonsense | LOF | ID | Syrbe et al., 2015 |
| *KCNA2* | EP and ID | 176262 | P405L | Nonsense | LOF | Moderate ID | Syrbe et al., 2015 |
| *KCNA2* | EP and ID | 176262 | L298F | Missense | GOF | Severe ID | Syrbe et al., 2015 |
| *KCNA2* | EP and ID | 176262 | R297Q | Missense | GOF | Moderate ID | Syrbe et al., 2015 |
| *KCNA4* | ID, striatal thinning, congenitalcataract and ADHD | 176266 | R89Q | Missense | LOF | Mild ID | Kaya et al., 2016 |
| *KCNA4* | ID, striatal thinning, congenitalcataract and ADHD | 176266 | R89Q | Missense | LOF | Mild ID | Kaya et al., 2016 |
| *KCNA4* | ID, striatal thinning, congenitalcataract and ADHD | 176266 | R89Q | Missense | LOF | Mild ID | Kaya et al., 2016 |
| *KCNA4* | ID, striatal thinning, congenitalcataract and ADHD | 176266 | R89Q | Missense | LOF | Mild ID | Kaya et al., 2016 |
| *KCNA4* | FHEIG | 176266 | A172E | Missense | GOF | Severe ID | Bauer et al., 2018 |
| *KCNA4* | FHEIG | 176266 | A244P | Missense | GOF | Moderate ID | Bauer et al., 2018 |
| *KCNA4* | FHEIG | 176266 | A172E | Missense | GOF | Severe ID | Bauer et al., 2018 |
| *KCND3* | Cerebellar ataxia complicated by ID, EP, and ADHD | 605411 | A293F295dup | Duplication | LOF | Mild ID | Smets et al., 2015 |
| *KCND3* | ID, ataxia, myoclonus, and dystonia. | 605411 | G384S | Missense | Unknown | Mild ID | Kurihara et al., 2018 |
| *KCNH1* | TBS | 603305 | G348R | Missense | GOF | Severe ID | Megarbane et al., 2016 |
| *KCNH1* | TBS | 603305 | G503R | Missense | GOF | Severe ID | Simons et al., 2015 |
| *KCNH1* | TBS | 603305 | L489F | Missense | GOF | Severe ID | Simons et al., 2015 |
| *KCNH1* | TBS | 603305 | I494V | Missense | GOF | Severe ID | Simons et al., 2015 |
| *KCNH1* | TBS | 603305 | K217N | Missense | GOF | Severe ID | Simons et al., 2015 |
| *KCNH1* | TBS | 603305 | I494V | Missense | GOF | Severe ID | Simons et al., 2015 |
| *KCNH1* | TBS | 603305 | I494V | Missense | GOF | Severe ID | Simons et al., 2015 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNH1* | ZLS | 603305 | I467V | Missense | GOF | Severe ID | Kortum et al., 2015 |
| *KCNH1* | ZLS | 603305 | S325Y and V356L | Missense | Both were GOF | Profound ID | Kortum et al., 2015 |
| *KCNH1* | ZLS | 603305 | G348R | Missense | GOF | Profound ID | Kortum et al., 2015 |
| *KCNH1* | ZLS | 603305 | G469R | Missense | GOF | Mild ID | Kortum et al., 2015 |
| *KCNH1* | ZLS | 603305 | L352V | Missense | GOF | Severe ID | Kortum et al., 2015 |
| *KCNH1* | ZLS | 603305 | I467V | Missense | GOF | Severe ID | Kortum et al., 2015 |
| *KCNQ2* | ID, EP and behavioral disorder | 602235 | R210C  | Missense | LOF | Moderate ID | Hewson et al., 2017 |
| *KCNQ2* | EP and moderate ID | 602235 | R210C  | Missense | LOF | Moderate ID | Hewson et al., 2017 |
| *KCNQ2* | EP and moderate ID | 602235 | R210C  | Missense | LOF | Moderate ID | Hewson et al., 2017 |
| *KCNQ2* | EP and moderate ID | 602235 | R210C  | Missense | LOF | Moderate ID | Hewson et al., 2017 |
| *KCNQ2* | EP and moderate ID | 602235 | R210C  | Missense | LOF | Moderate ID | Hewson et al., 2017 |
| *KCNQ2* | EP and moderate ID | 602235 | R210C  | Missense | LOF | Moderate ID | Hewson et al., 2017 |
| *KCNQ2* | Epileptic encephalopathy | 602235 | E140Q | Missense | LOF | Moderate GDD | Soldovieri et al., 2019 |
| *KCNQ2* | ID and EP | 602235 | R213Q | Missense | LOF | Profound ID | Weckhuysen et al., 2012 |
| *KCNQ2* | ID and EP | 602235 | T274M | Missense | LOF | Profound ID | Weckhuysen et al., 2012 |
| *KCNQ2* | ID and EP | 602235 | I205V | Missense | LOF | Moderate ID | Weckhuysen et al., 2012 |
| *KCNQ2* | ID and EP | 602235 | R560W | Missense | LOF | Profound ID | Weckhuysen et al., 2012 |
| *KCNQ2* | ID and EP | 602235 | M265V | Missense | LOF | Profound ID | Weckhuysen et al., 2012 |
| *KCNQ2* | ID and EP | 602235 | M546V | Missense | LOF | Profound ID | Weckhuysen et al., 2012 |
| *KCNQ2* | ID and EP | 602235 | G290D | Missense | LOF | Profound ID | Weckhuysen et al., 2012 |
| *KCNQ2* | ID and EP | 602235 | G290D | Missense | LOF | Profound ID | Weckhuysen et al., 2012 |
| *KCNQ2* | West syndrome | 602235 | Y284H | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | West syndrome | 602235 | R291G | Missense | LOF | Severe ID | Zhang et al., 2017 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNQ2* | West syndrome | 602235 | Y237F | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | Non syndromic | 602235 | G290S | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | West syndrome | 602235 | Y280H | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | Ohtahara syndrome | 602235 | E484D | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | West syndrome | 602235 | Q429Rfs\*5 | Nonsense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | West syndrome | 602235 | F305L | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | West syndrome | 602235 | A246P | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | West syndrome | 602235 | A265T | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | Non syndromic | 602235 | V250L | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | West syndrome | 602235 | T274M | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | Non syndromic | 602235 | R213W | Missense | LOF | Severe ID | Zhang et al., 2017 |
| *KCNQ2* | Ohtahara syndrome | 602235 | T217N | Missense | GOF | Profound ID | Kato et al., 2013 |
| *KCNQ2* | EOEE, unclassified | 602235 | A265V | Missense | Unknown | Moderate ID | Kato et al., 2013 |
| *KCNQ2* | Ohtahara syndrome | 602235 | A265V | Missense | Unknown | Profound ID | Kato et al., 2013 |
| *KCNQ2* | Ohtahara syndrome | 602235 | P285H | Missense | Unknown | Moderate ID | Kato et al., 2013 |
| *KCNQ2* | EOEE, unclassified | 602235 | G290S | Missense | Unknown | Profound ID | Kato et al., 2013 |
| *KCNQ2* | Ohtahara syndrome | 602235 | A294V | Missense | LOF | Profound ID | Kato et al., 2013 |
| *KCNQ2* | Ohtahara syndrome | 602235 | A294V | Missense | LOF | Profound ID | Kato et al., 2013 |
| *KCNQ2* | Ohtahara syndrome | 602235 | R333W | Missense | Unknown | Severe MR | Kato et al., 2013 |
| *KCNQ2* | Ohtahara/West syndrome | 602235 | R553W | Missense | Unknown | Profound ID | Kato et al., 2013 |
| *KCNQ2* | Ohtahara syndrome | 602235 | R553L | Missense | Unknown | Profound ID | Kato et al., 2013 |
| *KCNQ2* | Ohtahara syndrome | 602235 | P561L | Missense | Unknown | Profound ID | Kato et al., 2013 |
| *KCNQ2* | Ohtahara syndrome | 602235 | D563E | Missense | Unknown | Moderate ID | Kato et al., 2013 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNQ2* | EOEE | 602235 | G315R | Missense | Unknown | Severe ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | G281R | Missense | Unknown | Moderate ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | R210H | Missense | Unknown | GDD | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | D563N | Missense | LOF | GDD | Weckhuysen et al., 2013; Ambrosino et al., 2018b |
| *KCNQ2* | EOEE | 602235 | F305L | Missense | Unknown | Moderate ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | S195P | Missense | Unknown | Profound ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | G315R | Missense | Unknown | Profound ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | R201C | Missense | GOF | Profound ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | A265T | Missense | Unknown | Profound ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | R325G | Missense | LOF | Profound ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | R325G | Missense | LOF | Profound ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | R325G | Missense | LOF | Profound GDD | Soldovieri et al., 2016 |
| *KCNQ2* | EOEE | 602235 | R325G | Missense | LOF | Profound GDD | Soldovieri et al., 2016 |
| *KCNQ2* | EOEE | 602235 | R325G | Missense | LOF | Profound GDD | Soldovieri et al., 2016 |
| *KCNQ2* | EOEE | 602235 | R325G | Missense | LOF | Profound GDD | Soldovieri et al., 2016 |
| *KCNQ2* | EOEE | 602235 | G281R | Missense | Unknown | Profound ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | R210H | Missense | Unknown | Moderate ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | R210H | Missense | Unknown | Moderate ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | K552T | Missense | Unknown | Moderate ID. | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | D563N | Missense | Unknown | Severe ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EOEE | 602235 | K556E | Missense | Unknown | Moderate ID | Weckhuysen et al., 2013 |
| *KCNQ2* | EIEE | 602235 | S122L | Missense | LOF | Severe ID | Zhang Y. et al., 2015 |
| *KCNQ2* | EIEE | 602235 | K319T | Missense | Unknown | Severe ID | Zhang Y. et al., 2015 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNQ2* | EIEE | 602235 | T277I | Missense | Unknown | Severe ID | Zhang Y. et al., 2015 |
| *KCNQ2* | EIEE | 602235 | K552T | Missense | Unknown | Severe ID | Zhang Y. et al., 2015 |
| *KCNQ2* | West syndrome | 602235 | A306T | Missense | Unknown | Severe ID | Dimassi et al., 2016 |
| *KCNQ2* | ID and benign familial neonatal convulsion | 602235 | K554N | Missense | LOF | Profound ID | Borgatti et al., 2004 |
| *KCNQ2* | ID and benign familial neonatal convulsion | 602235 | K554N | Missense | LOF | Moderate ID | Borgatti et al., 2004 |
| *KCNQ2* | EOEE | 602235 | A294V | Missense | LOF | GDD | Abidi et al., 2015 |
| *KCNQ2* | EOEE | 602235 | A294V | Missense | LOF | GDD | Abidi et al., 2015 |
| *KCNQ2* | EOEE | 602235 | A294V | Missense | LOF | GDD | Abidi et al., 2015 |
| *KCNQ2* | EOEE | 602235 | A294V | Missense | LOF | GDD | Abidi et al., 2015 |
| *KCNQ2* | EOEE | 602235 | A294V | Missense | LOF | GDD | Abidi et al., 2015 |
| *KCNQ2* | EOEE | 602235 | A294V | Missense | LOF | Severe GDD | Abidi et al., 2015 |
| *KCNAB1* | EIEE | 601141 | L355Hfs\*5 | Nonsense | LOF | Severe ID | Zhang Y. et al., 2015 |
| *KCNQ3* | ID and EP | 602232 | R330L | Missense | LOF | Mild ID | Miceli et al., 2015b |
| *KCNQ3* | ID and EP | 602232 | R330L | Missense | LOF | Severe ID | Miceli et al., 2015b |
| *KCNQ3* | ID and EP | 602232 | R230L | Missense | Unknown | Mild ID | Yamamoto et al., 2019 |
| *KCNQ3* | ID | 602232 | R230C | Missense | GOF | Moderate ID | Sands et al., 2019 |
| *KCNQ3* | ID and ASD | 602232 | R230C | Missense | GOF | Severe ID | Sands et al., 2019 |
| *KCNQ3* | ID | 602232 | R227Q | Missense | GOF | Moderate ID | Sands et al., 2019 |
| *KCNQ3* | EOEE | 602232 | V359L and D542N | Missense | LOF | Severe GDD | Ambrosino et al., 2018b |
| *KCNQ5* | ID and EP | 607357 | S448I | Missense | LOF | Severe ID | Lehman et al., 2017 |
| *KCNQ5* | ID and EP | 607357 | V145G | Missense | LOF | Mild ID | Lehman et al., 2017 |
| *KCNQ5* | ID and EP | 607357 | L341I | Missense | LOF | Moderate ID | Lehman et al., 2017 |
| *KCNQ5* | ID and EP | 607357 | P369R | Missense | GOF | Profound ID | Lehman et al., 2017 |
| *KCNQ5* | ID and EP | 607357 | V133\* | Nonsense | LOF | Mild ID | Rosti et al., 2019 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNMA1* | ID, EP and cerebellar atrophy | 600150 | R458\* | Nonsense | LOF | Severe ID | Yesil et al., 2018 |
| *KCNMA1* | ID, EP and ASD | 600150 | A138V | Missense | LOF | Severe ID | Laumonnier et al., 2006 |
| *KCNMA1* | ID | 600150 | S351Y | Missense | LOF | Mild ID | Liang et al., 2019 |
| *KCNMA1* | ID | 600150 | G356R | Missense | LOF | Borderline ID | Liang et al., 2019 |
| *KCNMA1* | ID | 600150 | G375R | Missense | LOF | Severe ID | Liang et al., 2019 |
| *KCNMA1* | ID | 600150 | G375R | Missense | LOF | Severe ID | Liang et al., 2019 |
| *KCNMA1* | ID. | 600150 | G375R | Missense | LOF | Severe ID | Liang et al., 2019 |
| *KCNMA1* | ID | 600150 | N449fs and C413Y | Missense | LOF | ID | Liang et al., 2019 |
| *KCNMA1* | ID | 600150 | I663V | Missense | LOF | ID | Liang et al., 2019 |
| *KCNMA1* | ID | 600150 | P805L | Missense | LOF | ID | Liang et al., 2019 |
| *KCNMA1* | ID | 600150 | D984N | Missense | LOF | ID | Liang et al., 2019 |
| *KCNMA1* | GDD and paroxysmal non-kinesigenic dyskinesia | 600150 | E884K | Missense | Unknown | GDD | Zhang Z.B. et al., 2015 |
| *KCNMA1* | GDD and paroxysmal non-kinesigenic dyskinesia | 600150 | N1053S | Missense | Unknown | GDD | Zhang Z.B. et al., 2015 |
| *KCNMA1* | GDD, EP and severe cerebellar atrophy | 600150 | Y676Lfs∗7 | Nonsense | LOF | GDD | Tabarki et al., 2016 |
| *KCNMA1* | GDD, EP and severe cerebellar atrophy | 600150 | Y676Lfs∗7 | Nonsense | LOF | GDD | Tabarki et al., 2016 |
| *KCNC1* | ID | 176258 | R339\* | Nonsense  | LOF | Severe ID | Poirier et al., 2017 |
| *KCNC1* | ID | 176258 | R339\* | Nonsense | LOF | Moderate ID | Poirier et al., 2017 |
| *KCNC1* | ID | 176258 | R339\* | Nonsense | LOF | Severe ID | Poirier et al., 2017 |
| *KCNC1* | ID | 176258 | T399M | Missense | LOF | Moderate ID | Park et al., 2019 |
| *KCNC1* | ID and EP | 176258 | A421V | Missense | LOF | Severe ID | Cameron et al., 2019 |
| *KCNC1* | ID and EP | 176258 | A421V | Missense | LOF | Severe ID | Cameron et al., 2019 |
| *KCNC1* | ID and EP | 176258 | A421V | Missense | LOF | Moderate-severe ID | Cameron et al., 2019 |
| *KCNC1* | ID and EP | 176258 | A421V | Missense | LOF | Moderate-severe ID | Cameron et al., 2019 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNC1* | ID and EP | 176258 | A421V | Missense | LOF | GDD | Cameron et al., 2019 |
| *KCNC1* | ID and EP | 176258 | A421V | Missense | LOF | Moderate ID | Cameron et al., 2019 |
| *KCNC1* | ID and ASD | 176258 | R317H | Missense | LOF | Mild ID | Cameron et al., 2019 |
| *KCNC1* | ID | 176258 | Q492X | Missense | LOF | Moderate ID | Cameron et al., 2019 |
| *KCNC1* | ID | 176258 | Q492X | Missense | LOF | Moderate ID | Cameron et al., 2019 |
| *KCNJ6* | Keppen–Lubinsky Syndrome | 600877 | T152del | Deletion | LOF | Severe ID | Masotti et al., 2015 |
| *KCNJ6* | Keppen–Lubinsky Syndrome | 600877 | T152del | Deletion | LOF | Severe ID | Masotti et al., 2015 |
| *KCNJ6* | Keppen–Lubinsky Syndrome | 600877 | G154S | Missense | Unknown | Severe ID | Masotti et al., 2015 |
| *KCNB1* | Epileptic encephalopathy | 600397 | S347R | Missense | LOF | Severe ID | Torkamani et al., 2014 |
| *KCNB1* | Epileptic encephalopathy | 600397 | G379R | Missense | LOF | Severe ID | Torkamani et al., 2014 |
| *KCNB1* | Epileptic encephalopathy | 600397 | T374I | Missense | LOF | Severe ID | Torkamani et al., 2014 |
| *KCNB1* | Atypical Rett syndrome | 600397 | G379R | Missense | LOF | Severe ID | Srivastava et al., 2018 |
| *KCNB1* | Atypical Rett syndrome | 600397 | T374I | Missense | LOF | Severe ID | Srivastava et al., 2018 |
| *KCNB1* | West syndrome. | 600397 | W370\* | Nonsense | LOF | Severe ID | Krey et al., 2019 |
| *KCNB1* | GDD, ADHD, andASD | 600397 | R583\* | Nonsense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | West syndrome  | 600397 | K502fs | Frameshift | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD, ADHD, andASD | 600397 | F416L | Missense | Unknown | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and EP | 600397 | F416L | Missense | Unknown | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and EP | 600397 | G401R | Missense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and EP | 600397 | C397Y | Missense | Unknown | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and EP | 600397 | K391N | Missense | Unknown | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | West syndrome and ASD | 600397 | P385T | Missense | Unknown | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD | 600397 | G381R | Missense | Unknown | Severe GDD | de Kovel et al., 2017 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNB1* | GDD, EP, and ASD | 600397 | G379R | Missense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD, EP | 600397 | V378A | Missense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD, EP and ASD | 600397 | T374I | Missense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD, EP, ADHD andASD | 600397 | W369\* | Nonsense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and EP | 600397 | S363fs\*13 | Nonsense | LOF | Moderate GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and EP | 600397 | S347R | Missense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | West syndrome  | 600397 | R312H | Missense | Unknown | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD, EP, and ASD  | 600397 | R312H | Missense | Unknown | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD, EP, and ASD  | 600397 | R306C | Missense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and ADHD | 600397 | R306C | Missense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD, EP and ADHD | 600397 | Q228\* | Nonsense | LOF | Severe GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and EP | 600397 | L211P | Missense | Unknown | Moderate GDD | de Kovel et al., 2017 |
| *KCNB1* | GDD and EP | 600397 | T210K | Missense | Unknown | Moderate GDD | de Kovel et al., 2017 |
| *KCNB1* | ID, EP and ASD | 600397 | I199F | Missense | LOF | ID | Calhoun et al., 2017 |
| *KCNC3* | ID and ataxia | 176264 | D129N | Missense | GOF | Severe ID | Duarri et al., 2015 |
| *KCNC3* | ID | 176264 | V535M | Missense | GOF | Mild ID | Duarri et al., 2015 |
| *KCNC3* | ID and ataxia | 176264 | R423H | Missense | LOF | Mild ID | Duarri et al., 2015 |
| *KCNT1* | EP and ID | 608167 | R428Q | Missense | GOF | Severe ID | Alsaleem et al., 2019 |
| *KCNT1* | EIEE and severe dystonia | 608167 | L437F | Missense | GOF | Severe ID | Gertler et al., 2019 |
| *KCNT1* | EIMFS | 608167 | R428Q | Missense | GOF | Severe ID | Bearden et al., 2014 |
| *KCNT1* | EIMFS | 608167 | R933G | Missense | Unknown | Severe ID | Zhang et al., 2017 |
| *KCNT1* | EIMFS | 608167 | K629E | Missense | Unknown | Severe ID | Zhang et al., 2017 |
| *KCNT1* | EIMFS | 608167 | V271F | Missense | GOF | Severe ID | Zhang et al., 2017 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNT1* | EIMFS | 608167 | R428Q | Missense | GOF | Severe ID | Zhang et al., 2017 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense | GOF | Severe ID | Zhang et al., 2017 |
| *KCNT1* | EIMFS | 608167 | R474H | Missense | GOF | Severe ID | Zhang et al., 2017 |
| *KCNT1* | EIMFS | 608167 | R1114W | Missense  | GOF | Severe ID | Numis et al., 2018 |
| *KCNT1* | EIMFS | 608167 | A259D | Missense  | GOF | Severe ID | Numis et al., 2018 |
| *KCNT1* | EIMFS | 608167 | M516V | Missense  | GOF | Severe ID | Numis et al., 2018 |
| *KCNT1* | EIMFS | 608167 | R428Q | Missense  | GOF | Severe ID | Numis et al., 2018 |
| *KCNT1*  | EIMFS | 608167 | R428Q | Missense | GOF | Profound GDD | Barcia et al., 2019 |
| *KCNT1*  | EIMFS | 608167 | L437F | Missense | GOF | Profound GDD | Barcia et al., 2019 |
| *KCNT1*  | EIMFS | 608167 | R474C | Missense | GOF | Profound GDD | Barcia et al., 2019 |
| *KCNT1*  | EIMFS | 608167 | R474H | Missense | GOF | Profound GDD | Barcia et al., 2019 |
| *KCNT1*  | EIMFS | 608167 | R474H | Missense | GOF | Profound GDD | Barcia et al., 2019 |
| *KCNT1*  | EIMFS | 608167 | M516V | Missense | GOF | Profound GDD | Barcia et al., 2019 |
| *KCNT1*  | EIMFS | 608167 | M896I | Missense | GOF | Profound ID | Barcia et al., 2019 |
| *KCNT1*  | EIMFS-West syndrome | 608167 | A934T | Missense | GOF | Profound ID | Barcia et al., 2019 |
| *KCNT1*  | EIMFS | 608167 | A934T | Missense | GOF | Profound ID | Barcia et al., 2019 |
| *KCNT1*  | EIMFS | 608167 | A965V | Missense | GOF | Profound DD | Barcia et al., 2019 |
| *KCNT1*  | EIMFS-West syndrome | 608167 | R1106P | Missense | GOF | Profound DD | Barcia et al., 2019 |
| *KCNT1* | Ohtahara syndrome | 608167 | A966T | Missense | GOF | Profound DD | Martin et al., 2014 |
| *KCNT1* | EIMFS or Ohtahara syndrome | 608167 | R474C | Missense | GOF | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | EIMFS | 608167 | Q270E | Missense | Unknown | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | EIMFS | 608167 | P409S | Missense | Unknown | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | EIMFS | 608167 | G288S | Missense | Unknown | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | EIMFS | 608167 | R474H | Missense | GOF | Profound GDD | Ohba et al., 2015 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNT1* | EIMFS | 608167 | A934T | Missense | GOF | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | West syndrome | 608167 | R474H | Missense | GOF | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | EIMFS | 608167 | R474H | Missense | GOF | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | EIMFS | 608167 | P924L | Missense | Unknown | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | EIMFS | 608167 | A477T | Missense | Unknown | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | EIMFS | 608167 | R428Q | Missense | GOF | Profound GDD | Ohba et al., 2015 |
| *KCNT1* | ID, EP and cerebellar ataxia | 608167 | R565H | Missense | Unknown | Severe ID | Hansen et al., 2017 |
| *KCNT1* | ID, EP and cerebellar ataxia | 608167 | R565H | Missense | Unknown | Severe ID | Hansen et al., 2017 |
| *KCNT1* | EIMFS | 608167 | R474C | Missense | Unknown | Severe GDD | Shimada et al., 2014 |
| *KCNT1* | Leukoencephalopathy, ID and severe myoclonic EP | 608167 | F932I | Missense | Unknown | Severe ID | Vanderver et al., 2014 |
| *KCNT1* | ID, EP, delayed myelination and leukoencephalopathy | 608167 | F932I | Missense | LOF | Severe GDD | Evely KM et al., 2017 |
| *KCNT1* | EIMFS | 608167 | R428Q | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | D480N | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS/West syndrome | 608167 | R950Q | Missense  | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS/West syndrome | 608167 | A934T | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | K629E | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | R474H | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | G288S | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | ID | 608167 | G288S | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | West syndrome | 608167 | G288S | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | G288S | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | Unclassified EOEE | 608167 | C377S | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNT1* | EIMFS | 608167 | R474C | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | L962P | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | R950Q | Missense  | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | ID and EP | 608167 | R474H | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | Unclassified EOEE | 608167 | R85S | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | L274I | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | R474H | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | Unclassified EOEE | 608167 | R961H | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | Unclassified EOEE | 608167 | R474C | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | R428Q | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | Unclassified EOEE | 608167 | A934T | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | Q270E | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | ID | 608167 | R398Q | Missense  | Unknown | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense | GOF | Severe GDD | Borlot F et al., 2020 |
| *KCNT1* | EIMFS | 608167 | G288S | Missense  | Unknown | Profound GDD | Rizzo F et al., 2016 |
| *KCNT1* | EIMFS | 608167 | M516V | Missense | GOF | Profound GDD | Rizzo F et al., 2016 |
| *KCNT1* | EIMFS | 608167 | C377S | Missense | Unknown | Profound GDD | Kawasaki Y et al., 2017 |
| *KCNT1* | EIMFS | 608167 | R474C | Missense | GOF | Profound GDD | Kawasaki Y et al., 2017 |
| *KCNT1* | EIMFS | 608167 | R474H | Missense | GOF | Profound GDD | Kawasaki Y et al., 2017 |
| *KCNT1* | EIMFS | 608167 | V271F | Missense | GOF | Profound GDD | McTague A et al., 2018 |
| *KCNT1* | EIMFS | 608167 | L274I | Missense | GOF | Profound GDD | McTague A et al., 2018 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNT1* | EIMFS | 608167 | M896K | Missense | Unknown | Profound GDD | McTague A et al., 2018 |
| *KCNT1* | EIMFS | 608167 | R950Q | Missense  | GOF | Severe GDD | McTague A et al., 2018 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense  | GOF | Profound GDD | McTague A et al., 2018 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense  | GOF | Profound GDD | McTague A et al., 2018 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense  | GOF | Profound GDD | McTague A et al., 2018 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense  | GOF | Profound GDD | McTague A et al., 2018 |
| *KCNT1* | EIMFS | 608167 | A934T | Missense  | GOF | Profound GDD | McTague A et al., 2018 |
| *KCNT1* | EIMFS | 608167 | R950Q | Missense  | GOF | Severe GDD | Dilena R et. Al., 2018 |
| *KCNT1* | EIMFS | 608167 | E893K | Missense  | GOF | Severe GDD | Dilena R et. Al., 2018 |
| *KCNT2* | EOEE | 610044 | F240L | Missense | LOF | Severe GDD | Gururaj S et al., 2017 |
| *KCNT2* | West syndrome then Lennox-Gastaut syndrome | 610044 | R190H | Missense | GOF | Severe GDD | Ambrosino P et al., 2018a |
| *KCNT2* | EIMFS | 610044 | R190P | Missense | GOF | Severe GDD | Ambrosino P et al.,2018a |
| *KCNT2* | EIMFS-like EOEE | 610044 | L48Qfs43\* | Nonsense | LOF | Severe GDD | Mao X et al., 2020 |
| *KCNT2* | EIMFS | 610044 | K564\* | Nonsense | LOF | Severe GDD | Mao X et al., 2020 |
| *KCTD3* | ID and EP | 613272 | P346Tfs\*4 | Nonsense | LOF | Severe GDD | Faqeih et al., 2018 |
| *KCTD3* | ID and EP | 613272 | P346Tfs\*4 | Nonsense | LOF | Severe GDD | Faqeih et al., 2018 |
| *KCTD3* | ID and EP | 613272 | P346Tfs\*4 | Nonsense | LOF | Severe GDD | Faqeih et al., 2018 |
| *KCTD3* | ID and EP | 613272 | P346Tfs\*4 | Nonsense | LOF | Severe GDD | Faqeih et al., 2018 |
| *KCTD3* | ID and EP | 613272 | R56\* | Nonsense | LOF | Severe GDD | Faqeih et al., 2018 |
| *KCTD3* | ID and EP | 613272 | R56\* | Nonsense | LOF | Severe GDD | Faqeih et al., 2018 |
| *KCTD3* | ID and EP | 613272 | P346Tfs\*4 | Nonsense | LOF | Moderate GDD | Faqeih et al., 2018 |
| *KCTD3* | ID and EP | 613272 | P346Tfs\*4 | Nonsense | LOF | Severe ID | Alazami et al., 2015 |
| *KCTD3* | ID and EP | 613272 | P346Tfs\*4 | Nonsense | LOF | Severe ID | Trujillano et al., 2017 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNJ11* | DEND  | 600937 | Q52R | Missense | GOF | GDD | Flanagan et al., 2006 |
| *KCNJ11* | DEND  | 600937 | G53D | Missense | GOF | GDD | Flanagan et al., 2006 |
| *KCNJ11* | DEND  | 600937 | V59G | Missense | GOF | GDD | Flanagan et al., 2006 |
| *KCNJ11* | DEND  | 600937 | C166Y | Missense | GOF | Profound GDD | Flanagan et al., 2006 |
| *KCNJ11* | DEND  | 600937 | I296L | Missense | GOF | GDD | Flanagan et al., 2006; Proks et al., 2005 |
| *KCNJ11* | I-DEND  | 600937 | R201C | Missense | GOF | GDD | Flanagan et al., 2006 |
| *KCNJ11* | I-DEND | 600937 | R201C | Missense | GOF | GDD | Flanagan et al., 2006 |
| *KCNJ11* | I-DEND  | 600937 | V59M | Missense | GOF | GDD | Flanagan et al., 2006 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | GDD | Flanagan et al., 2006 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Flanagan et al., 2006 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Flanagan et al., 2006 |
| *KCNJ11* | I-DEND | 600937 | H46L | Missense | GOF | Moderate ID | Fendler et al., 2013 |
| *KCNJ11* | I-DEND | 600937 | G53D | Missense | GOF | Moderate ID | Fendler et al., 2013 |
| *KCNJ11* | I-DEND | 600937 | G53D | Missense | GOF | Moderate ID | Fendler et al., 2013 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Fendler et al., 2013 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Shah et al., 2012 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Shah et al., 2012 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Shah et al., 2012 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Shah et al., 2012 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Shah et al., 2012 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Shah et al., 2012 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Shah et al., 2012 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| **Name of the gene** | **Syndrome/Phenotype** | **OMIM number** | **Protein change** | **Type of mutation** | **Electrophysiological study result** | **Severity of the GDD/ID** | **Reference** |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND  | 600937 | V59M | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND | 600937 | V59A | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND | 600937 | Y330C | Missense | GOF | Moderate ID | Carmody et al., 2016 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Mohamadi et al., 2010 |
| *KCNJ11* | I-DEND | 600937 | homS225T, del | Missense and deletion | GOF and LOF | Moderate ID | Lin et al., 2013 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Massa et al., 2005 |
| *KCNJ11* | I-DEND | 600937 | V59M | Missense | GOF | Moderate ID | Massa et al., 2005 |
| *KCNJ11* | I-DEND | 600937 | K170N | Missense | GOF | Moderate ID | Massa et al., 2005 |

**Abbreviations: ADHD; attention deficit hyperactive disorder, ASD; autism spectrum disorder, DEND: developmental delay, epilepsy, and neonatal diabetes, EEG; electroencephalograph, EP; epilepsy, EIMFS; epilepsy of infancy with migrating focal seizures, EAST: epilepsy, ataxia, sensorineural deafness, and tubulopathy, FHEIG: facial dysmorphism, hypertrichosis, epilepsy, intellectual disability/developmental delay, and gingival overgrowth, GDD; global developmental delay, GOF: gain of function, ID: intellectual disability, I-DEND: neonatal diabetes with moderate developmental delay and/or muscle weakness but not epilepsy, LOF: loss of function, MRI; magnetic resonance imaging, SeSAME: seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance, TBS: Temple–Baraitser Syndrome, ZLS; Zimmermann–Laband syndrome.**

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