Updated complete list of hereditary paroxysmal movement disorders

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| **Designation** | **Less common movement phenotype** | **Clinical clues** | | **OMIM** | **MOI** |
| **Predominant dyskinesias** | | | | | |
| PxMD-*KCNMA1149, 150* |  | Paroxysmal non-kinesigenic dyskinesia (mainly dystonic) with no clear trigger, developmental delay, generalized epilepsy | | 609446 | AD |
| PxMD-*PNKD*  (formerly *MR1*)933 |  | Paroxysmal non-kinesigenic dyskinesia (PNKD) | | 118800 | AD |
| PxMD-*PRRT2*934 |  | Paroxysmal kinesigenic dyskinesia (PKD), rarely other paroxysmal movement disorders (PNKD, PED, episodic ataxia, writer’s cramp) or hemiplegic migraine. The PRRT2-associated disease spectrum also includes benign familial infantile epilepsy (BFIE) and combined phenotypes (paroxysmal kinesigenic dyskinesia with infantile convulsions (PKD/IC)) | | 128200 | AD |
| MYC/PxMD-*SCN8A*A,935 |  | Paroxysmal kinesigenic dyskinesia, seizure disorder (wide spectrum with benign infantile seizures in some and epileptic encephalopathy in others), intellectual disability | | 617080 | AD |
| PxMD-*SLC2A1B,*773 |  | Paroxysmal exercise (exertion)-induced dyskinesia,  Alternative phenotype: Glut1 deficiency syndrome (see footnote) | | 612126 | AD |
| **Predominant ataxias** | | | | | |
| PxMD-*CACNA1A*936 |  | Paroxysmal ataxia with vertigo, nausea, headaches, weakness and other manifestations, often favorable response to acetazolamide | | 108500 | AD |
| PxMD-*KCNA1*937 |  | Paroxysmal ataxia with interictal myokymia | | 160120 | AD |
| PxMD-*PDHA1*938 |  | Pyruvate dehydrogenase deficiency 939, 940: Paroxysmal episodes of ataxia, dystonia, occasionally parkinsonism  Additional clinical features: developmental delay/intellectual disability, encephalopathy, truncal hypotonia, seizures, microcephaly, spasticity, facial dysmorphism, peripheral neuropathy | | 300502 | XL |
| PxMD-*SLC1A3*941 |  | Paroxysmal ataxia with vertigo, nausea, seizures, migraine, weakness, alternating hemiplegia and other manifestations | | 612656 | AD |
| **Predominant dystonia** | | | | | |
| PxMD-*ECHS1144-148* | Ataxia, spasticity | Leigh syndrome: onset before age 10, paroxysmal dystonia triggered by high metabolic demand (exercise, fever, low calorie intake), developmental delay, acute episodes of encephalopathy, increased plasma lactate, and urinary excretion of organic acids | | 616277 | AR |
| **Disorders that usually present with other phenotypes but can have predominant paroxysmal dyskinesias** | | | | | |
| ***Gene*** | **Associated disease** | **OMIM** | **Clinical phenotype** | | **MOI** |
| *GLDC*942 | Glycine encephalo-pathy | 605899 | Glycine encephalopathy; intermittent chorea during febrile illness | | AR |

AD = autosomal dominant, AR = autosomal recessive, MOI = mode of inheritance, OMIM = Online Mendelian Inheritance in Man (<https://www.omim.org/about>), XL = x-linked

A Gene mutations can also cause Familial Myoclonus Type 2 (OMIM [**618364**](https://www.omim.org/entry/618364); Table 5), autosomal-dominant cognitive impairment with or without cerebellar ataxia (OMIM [**614306**](https://www.omim.org/entry/614306)), and/or autosomal-dominant developmental and epileptic encephalopathy 13 (DEE13, OMIM [**614558**](https://www.omim.org/entry/614558)).

B Mutations in this gene more commonly cause infantile-onset epileptic encephalopathy, delayed development, acquired microcephaly, ataxia, dystonia, spasticity, atypical phenotypes without epilepsy include patients with mixed movement disorders and mental retardation or adult-onset cases with minimal symptoms (Glut1 deficiency syndrome).875

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