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The proposed new list of paroxysmal movement disorders

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| --- | --- | --- | --- | --- |
| **New designation**  | **Less common movement phenotype** | **Clinical clues** | **Inheri-tance** | **Locus symbol** |
| **Predominant dyskinesias** |
| PxMD-*PRRT21* |  | 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))GeneReview: http://www.ncbi.nlm.nih.gov/books/NBK1460/OMIM 128200 | AD | *DYT10 or DYT19* |
| PxMD-*PNKD*(formerly *MR1*)2 |  | Paroxysmal non-kinesigenic dyskinesia (PNKD)GeneReview: http://www.ncbi.nlm.nih.gov/books/NBK1221/OMIM 118800 | AD | *DYT8* |
| PxMD\*-*SLC2A13* |  | Paroxysmal exercise (exertion)-induced dyskinesia,Allelic phenotype: Glut1 deficiency syndrome (see footnote) GeneReview (for GLUT1-deficiency syndrome): http://www.ncbi.nlm.nih.gov/books/NBK1430/OMIM 612126 | AD | *DYT18/DYT9* |
| PxMD-KNCMA14, 5 |  | Paroxysmal non-kinesigenic dyskinesia (mainly dystonic) with no clear trigger, developmental delay, generalized epilepsy  | AD |  |
| **Disorders that usually present with other phenotypes but can have predominant paroxysmal dyskinesias** |
| PxMD – *SCN8A6* |  | Phenotype: Paroxysmal kinesigenic dyskinesia, seizure disorder (wide spectrum with benign infantile seizures in some and epileptic encephalopathy in others), intellectual disability |  |  |
| *GLDC* | PxMD7 | Glycine encephalopathy Intermittent chorea during febrile illnessGeneReviews: http://www.ncbi.nlm.nih.gov/books/NBK1357/OMIM [605899](http://omim.org/entry/605899)  |  |  |
| CHOR/DYT/PxMD-ADCY58 |  | Pleiotropic dyskinesia (choreiform, myoclonic, dystonic) mainly involving the limbs, neck, and/or face. Paroxysmal worsening triggered by anxiety or drowsiness. Axial hypotonia, developmental delay, abnormal saccades, spasticity.GeneReviewshttps://www.ncbi.nlm.nih.gov/books/NBK263441/OMIM 600293 | AD |  |
| **Predominant ataxias** |
| PxMD-*KCNA19* |  | Paroxysmal ataxia with interictal myokymiaGeneReviewshttp://www.ncbi.nlm.nih.gov/books/NBK25442/OMIM 160120 | AD | EA1 |
| PxMD-*CACNA1A10* |  | Paroxysmal ataxia with vertigo, nausea, headaches, weakness and other manifestations, often favorable response to acetazolamideGeneReview http://www.ncbi.nlm.nih.gov/books/NBK1501/OMIM 108500 | AD | EA2 |
| PxMD-*SLC1A311* |  | Paroxysmal ataxia with vertigo, nausea, seizures, migraine, weakness, alternating hemiplegia and other manifestationsGeneReview n/aOMIM 612656 | AD | EA6 |
| PxMD-*PDHA112* |  | Pyruvate dehydrogenase deficiency 13, 14: Paroxysmal episodes of ataxia, dystonia, occasionally parkinsonismAdditional clinical features: Developmental delay / intellectual disability, encephalopathy, truncal hypotonia, seizures, microcephaly, spasticity, facial dysmorphism, peripheral neuropathy14GeneReviews n/aOMIM 300502 | X-linked | None |

\* 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).118

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