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Confirmed forms of mixed movement disorders

Designation	Clinical clues	OMIM	MOI
MxMD-*ADCY5*[1]	Pleiotropic dyskinesia (choreatic, myoclonic, dystonic) mainly involving the limbs, neck, and/or face, paroxysmal worsening triggered by anxiety or drowsiness, axial hypotonia, developmental delay, abnormal saccades, spasticity	600293	AD
MxMD-*ATP13A2*[A]	Broad and variable clinical spectrum including several movement disorders: 1) Kufor-Rakeb syndrome[2-4]: juvenile-onset atypical dystonia-parkinsonism, supranuclear gaze palsy, pyramidal signs, dementia, dysphagia, dysarthria and olfactory dysfunction; 2) HSP[5-8]: adult-onset, characterized by spasticity, lower limb weakness, cognitive impairment, psychiatric symptoms, axonal neuropathy, thin corpus callosum and *ear of the lynx* sign on MRI; 3) Adult-onset progressive ataxia[9-11] and action myoclonus[9, 10, 12-14]	606695 (PARK), 617225 (HSP)	AR
MxMD-*MYORG*-(PFBC)[15-18]	Dysarthria, cognitive deficits, and depression, headaches and psychosis in a lower percentage, imaging abnormalities include basal ganglia and cerebellum calcification	618317	AR
MxMD-*OPA3*[19-24]	3-Methylglutaconic Aciduria Type 3 (MGCA3; many alternative names); neuroophthalmological syndrome with early-onset bilateral optic atrophy with progressive decrease in visual acuity and horizontal nystagmus, choreoathetoid movements before age ten, which can restrict ambulation, spastic paraparesis in second decade, pyramidal dysfunction, ataxia, and variable cognitive impairment	258501	AR
MxMD-*PDGFB*-(PFBC)[25, 26]	Parkinsonism, ataxia, or chorea with possible additional headache and cognitive deficits, imaging abnormalities include thalamus, cerebellum, white matter, and basal ganglia calcifications	615483	AD
MxMD-*POLG*[27-29]	Multiple syndromes often with progressive external ophthalmoparesis and variable other neurological manifestations; rarely prominent parkinsonism	174763	AD or AR

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

[A] Mutations in this gene also cause neuronal ceroid lipofuscinosis (CLN12).[12]

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