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Recently identified or confirmed forms of hereditary parkinsonism

Designation	Clinical features	OMIM	MOI
Classical parkinsonism			
PARK-*CHCHD2*[1-8]	Typical levodopa-responsive parkinsonism	616710	AD[#]
Atypical parkinsonism or complex phenotypes			
PARK-*DCTN1*[9-12]	Adult-onset (atypical) parkinsonism with depression or apathy, followed by weight loss and respiratory hypoventilation/failure (referred to as *Perry syndrome*); some cases reported with PSP-like phenotype	168605	AD
PARK-*RAB39B*[13-15]	Early-onset (atypical) parkinsonism, delayed psychomotor development, impaired intellectual development (referred to as *Waisman syndrome*)	311510	XLR
PARK-*VPS13C*[12, 16-20]	Early-onset parkinsonism with often rapid and severe progression and loss of response to levodopa during disease course, early cognitive impairment potentially leading to dementia	616840	AR
Reclassified primary familial brain calcification genes			
PARK-*JAM2*-(PFBC)[21-23]	Atypical parkinsonism with cognitive deficits, brain imaging: calcifications in basal ganglia, cerebellum, and white matter	618824	AR
PARK-*SLC20A2*-(PFBC)[23, 24]	Atypical parkinsonism, commonly with cognitive deficits and headaches, less commonly dystonia, chorea, and ataxia, brain imaging: calcifications in basal ganglia, thalamus, cerebellum, and white matter	213600	AD

Disorders that usually present with other phenotypes but can have (prominent) parkinsonism				
Gene	**Disease**	**OMIM**	**Clinical phenotype**	**MOI**
DYT-*DNAJC12*[25, 26]	Hyperphenylalaninemia	617384	Increased serum phenylalanine and highly variable neurological defects including movement disorder phenotypes; many cases with dystonia and variable impairment of intellectual development, phenotype can also include non-progressive or mild levodopa-responsive parkinsonism	AR
MYC/ATX-*EPM2A*[27, 28]	Progressive myoclonus epilepsy (Lafora disease)	254780	Early-onset progressive neurodegeneration with myoclonus, generalized seizures, often visual hallucinations and cognitive decline, phenotype can also include ataxia or rarely parkinsonism	AR
C9orf72[29, 30]	Frontotemporal dementia (FTD) and/or Amyotrophic Lateral Sclerosis (ALS)	105550	Broad phenotypic spectrum including frontotemporal dementia and features of motor neuron disease, parkinsonism (mostly atypical, e.g., PSP-like, MSA or CBS), and also dystonia, cerebellar signs, or chorea	AD, repeat expansion
GRN[31, 32]	Primary progressive aphasia (PPA), Frontotemporal lobar degeneration with ubiquitin-positive inclusions, and Neuronal ceroid lipofuscinosis type 11	607485, 607485, 614706	Phenotypic spectrum includes atypical parkinsonism (PSP-like, CBS, and DLB)	AD or AR
MAPT[6, 30, 33-38]	Frontotemporal dementia with or	600274, 172700,	Broad phenotypic spectrum including mostly atypical parkinsonism (PSP-like, CBS) but also	AD or AR

	without parkinsonism, Pick disease, Progressive supranuclear palsy, Progressive atypical supranuclear palsy	601104, 260540	features of motor neuron disease (eg, ALS); susceptibility locus for PD (OMIM: 168600)	
PDE8B[39-43]	Autosomal dominant striatal degeneration	609161	Neurological disorder with variable movement abnormalities due to striatal dysfunction; phenotypic spectrum includes slowly progressive parkinsonism (mainly bradykinesia and rigidity, usually no tremor) without response to levodopa, as well as dysarthria, gait disturbances, and brisk (lower limb) reflexes	AD
PDGFRB-(PFBC)[*,44]	Idiopathic basal ganglia calcification-4 (IBGC4)	615007	Many asymptomatic carriers, prominent late-onset parkinsonism and cognitive impairment in a minority of patients, brain imaging: calcification most commonly in basal ganglia	AD
XPR1-(PFBC)[*,45]	Idiopathic basal ganglia calcification-6 (IBGC6)	616413	Neurodegenerative disorder with adult onset neuropsychiatric and movement disorders including parkinsonism, dystonia, gait abnormalities, chorea, psychosis, and dementia, brain imaging: calcification most commonly in basal ganglia	AD

AD = autosomal dominant, AR = autosomal recessive, CBS = corticobasal syndrome, DLB = Dementia with Lewy bodies, MOI = mode of inheritance, MSA = multiple system atrophy, OMIM = Online Mendelian Inheritance in Man (https://www.omim.org/about), PSP = progressive supranuclear palsy, XLR = x-linked recessive

[#] In addition to pathogenic variants as monogenic causes of the disease, some of the reported variants in *CHCHD2* represent genetic risk factors also occurring in a considerable but lower number of control individuals when compared to PD patients.

[*] These genes were previously included in the list of primary familial brain calcification.

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