CHOREA

Gene	Condition	Her	Variants	NGS panel	Expansion	WES	WGS	mtDNA	Other	AOO	Genetic tricks	Clinical traits	Ref
HTT	**Huntington disease**	AD	CAG expansion		x		x		LRS	Childhood to late adulthood	Anticipation on paternal inheritance. Penetrance: complete with ≥ 39 rpt; incomplete with 36-38 rpt (somatic mosaicism with possible longer expansions in certain brain areas). Correlation between rpt, AOO and severity. Genetic modifiers like *FAN1, MTMR10, RRM2B, URB5* play a significant role in disease development.	**Classic form**: chorea, dementia, psychiatric symptoms. **Juvenile form** "*Westphal variant*": rigid-hypokinetic, seizures, dementia, psychiatric symptoms. **Brain MRI:** caudate nucleus head atrophy	(1,2)
PRNP	**HDL-1**	AD	Octapeptide expansion		x		x		LRS	Adulthood	Allelic disorder to Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Straussler disease, Spongiform encephalopathy with neuropsychiatric features, and Cerebral amyloid angiopathy *PRNP*-related (also caused by SNV)	"Early-onset prion disease" with prominent psychiatric features, ataxia, chorea, rigidity, dysarthria, dementia, seizures.	(3)
JPH3	**HDL-2**	AD	CAG/CTG expansion		x		x		LRS	Adulthood	Only described in patients of African ancestry. Anticipation. Penetrance: < 33-35 rpt may be unstable in vertical transmission, 40-43 rpt: incomplete; > 43 rpt complete (impact of alleles of 36-39 rpt remain uncertain)	Psychiatric symptoms, dementia, chorea and/or rigidity/ parkinsonism, dysarthria. **Brain MRI:** cortical and striatal atrophy	(4,5)
genetic cause is unknown	**HDL-3**	AR	*unknown*	NA						Childhood - Adulthood		Normal developmental, followed by decline in cognition and movements, with chorea, dystonia, ataxia, gait instability, spasticity. Seizures. **Brain MRI:** frontal cortical and caudate atrophy	(6)
TBP	**HDL-4, SCA17**	AD (rarer AR)	CAG/CAA repeats		x		x		LRS	Adulthood	Anticipation. Penetrance: complete with >46 rpt; incomplete with 41-46 rpt; unaffected with 25-41. Digenic disease**:** patients with intermediate *TBP* expanded alleles in combination with heterozygous mutations in the *STUB1* gene (SCA48) demonstrate full penetrance	Cerebellar ataxia, chorea and/or rigidity/parkinsonism, pyramidal signs, cognitive impairment, psychosis, seizures **Brain MRI:** cerebellar atrophy	(7–10)
ATN1	**Dentatorubral-pallidoluysian atrophy (DRPLA)**	AD	CAG repeats (100%)		x		x		LRS	Early adulthood	Almost only described in Japanese (few families African Americans and Caucasians). Number rpt correlates with different phenotypes	**Juvenile form** (<20yy, typically >65 rpt): developmental delay, intellectual disability, myoclonus, and epilepsy **Adult-onset form:** ataxia and/or choreoathetosis, personality changes +/- cognitive decline	(11–13)

C9orf72	**Frontotemporal dementia and/or Motor neuron disease**	AD	Hexanucleotide repeats G_4C_2 (GGGGCC)		x		x		LRS	Adulthood	Second most common cause in Caucasians. Repeats size: 25-60 rpt uncertain significance (somatic mosaicism with possible longer expansions in certain brain regions); 61 to >4000 rpt pathogenic.	Movement disorders, including chorea, dystonia, myoclonus, tremor, parkinsonism may precede signs of FTD/ALS, or even be present in isolation	(14)
VPS13A	**Chorea-acanthocytosis**	AR	SNV, del/dup, splice site (70-90%)	x		x	x			Early adulthood	Penetrance is complete.	Triad of progressive movement disorder (chorea, dystonia, tics with prominent orofacial involvement and common bite lesions; in later stages parkinsonism) Cognitive alterations. Psychiatric issues. Epileptic seizures. Myopathy, neuropathy with weak/absent tendon reflexes. Dysphagia, dysarthria. Hepatosplenomegaly. Cardiomyopathy, arrhythmia. **Brain MRI:** caudate atrophy. Muscle and liver enzymes, markers of hemolysis. RBC acanthocytosis	(15)
			CNV				x		del/dup analyses (10-30%)				(16)
XK	**McLeod syndrome**	X-linked recessive	SNV (60%)	x		x	x			Adulthood	Only males affected. Females heterozygous have **mosaicism**: may develop clinical manifestations including chorea or late-onset cognitive decline. Penetrance is almost complete.	**CNS:** Psychiatric issues, cognitive alterations, movement disorders, sensorimotor axonopathy, muscle weakness. **Cardiac**: Dilated cardiomyopathy, atrial fibrillation, and tachyarrhythmia. **Hematological**: absent expression of the Kx erythrocyte antigen and weakened expression of Kell blood group antigens: hemolysis. RBC acanthocytosis	(17)
			CNV				x		del/dup analyses (40%), CMA (30%)				
NKX2-1 *(also known as **TTF-1**)*	**Benign Hereditary Chorea**	AD (rarer AR)	CNV (2%)				x		del/dup analyses (16%), CMA	Infancy - Childhood	Allelic disorder to: Choreoathetosis, hypothyroidism, and neonatal respiratory distress; Nonmedullary thyroid cancer. Splicing mutations in introns.	Chorea which peaks in the second decade and does not progress. Possible dysarthria, gait abnormalities, slightly decreased intelligence.	(18)
			SNV, del/dup, splice site (82%)	x		x	x						
PDE10A	**PDE10A-related chorea**	AR	SNV	x		x	x			Infancy		Infantile onset chorea, developmental and cognition delay with **normal brain MRI**	(19)
	PDE10A-related chorea with STRIATAL DEGENERATION	AD/De novo	SNV	x		x	x			Childhood		Childhood onset chorea, normal cognition and development, with **T2-hyperintense symmetrical bilateral striatal lesions**	(20)
ADCY5	**Dyskinesia with orofacial involvement**	AD (rarer AR) / often De novo	SNV, del, splice site (100%)	x		x	x			Infancy to late-adolescence	**Somatic mosaicism** occurring in later stages of embryogenesis, demonstrated in high proportion, in part responsible for intra-familial phenotypic variability. Penetrance 100%.	Dyskinesias: chorea, dystonia, myoclonus, in variable association; ++ face, + arms; constant or paroxysmal, in sleep; typical improvement. in early adulthood. Triggers: none or emotional stress, intercurrent illness, sneezing, caffeine. Sleep related motor and behavior disorder. Alternating hemiplegia of childhood. Possible axial hypotonia, delayed	(21)

												milestones, +/ MCI (often misdiagnosed as CP).	
GPR88	**Chorea, childhood-onset, with psychomotor retardation**	AR	SNV	x		x	x			Infancy		Delayed psychomotor development, Generalized chorea.	(22)
ATM	see Supplementary Table 3												
GNAO1	see Supplementary Table 2												
HPRT1	see Supplementary Table 2												

Supplementary Table 1. Most relevant genetic causes of choreic syndromes and related test selection. For each listed gene, disease nomenclature, reported genetic variants are listed, appropriate diagnostic test selection (based on described genetic variants), important genetic highlights, and main clinical traits are reported. Percentage values reported represent the amount of pathogenetic variants detected with the specific analysis or gene-targeted deletion/duplication analysis on GeneReviews® when available/applicable. **AD**: autosomal dominant. **AOO**: age of onset. **AR**: autosomal recessive. **C**: cytosine. **CMA**: Chromosomal microarray analysis. **CNV**: deletion/insertion/duplication of > 50 base pairs. **del/ins/dup**: deletion/insertion/duplication < 50 base pairs. **del/dup analyses**: gene-targeted deletion/duplication analysis (methods used may include a range of techniques such as quantitative PCR, long-range PCR (**LR-PCR**), multiplex ligation-dependent probe amplification (**MLPA**), and a gene-targeted microarray designed to detect single-exon deletions or duplications). **G**: guanine. **Her**: inheritance. **Expansion**: repeat expansion tests (method used may include long-range PCR (**LR-PCR**), repeat-primed PCR (**RP-PCR**). **LRS**: long-reads sequencing. **mtDNA**: mitochondrial DNA tests; **NGS**: next generation sequencing. **Rpt**: repeats. **SNV**: single nucleotide variant. **splice site**: splice site variant. **T**: thymidine. **UNK**: no data available. **WES**: whole exomes sequencing. **WGS**: whole genome sequencing.

DYSTONIA

Gene	Condition	Her	Variants	NGS panel	Expansion	WES	WGS	mtDNA	Other	AOO	Genetic tricks	Clinical traits	Ref
Isolated dystonia													
ANO3	**DYT-ANO3**	AD (rarely de novo)	SNV, splice site	x		x	x			Anytime (peak in adulthood)	Often de novo variants are found in childhood onset. 0.8% missense variants in HC (critical evaluation of each variant before establishing causality)	Multifocal cranio-cervical dystonia, often with laryngeal involvement and ULs tremor. Rarely can present with dystonia plus myoclonus; paroxysmal-dystonia.	(23–25)
GNAL	**DYT-GNAL**	AD	SNV, ins (100%)	x		x	x			Adulthood	Incomplete penetrance. Intrafamilial variability. AR in one family, with ID and generalized dystonia	Focal/multifocal cervical dystonia, spreading to face or arms. Dystonic tremor may precede dystonia onset. Rarely can present with dystonia plus myoclonus.	(26–28)
HPCA	**DYT-HPCA**	AR	SNV, del	x		x	x			Childhood - adolescence		Generalized dystonia, starting in limbs, later involving cranio-cervical region. +/- mild syndromic features (ID, seizure, psychiatric features)	(29,30)
KMT2B	**DYT-KMT2B**	AD (mostly de novo)	SNV, del/dupl, splice site (72%)	x		x	x			Childhood; rarely adulthood	Second most common cause of early onset generalized dystonia. Incomplete penetrance. Variable expression	Generalized dystonia starting in limbs, then involving cranial, cervical and laryngeal regions. Commonly reported mild syndromic features (ID, short stature, mild dysmorphic features etc.)	(31,32)
			Chromosomal microdel involving the whole gene (28%)				x		del/dup analyses: CMA				
PRKRA	**DYT-PRKRA**	AR	SNV, del	x		x	x			Childhood - adolescence		Dystonia with prominent oromandibular involvement, dysphagia, retrocollis. +/- mild Parkinsonian features	(33)
THAP1	**DYT-THAP1**	AD	SNV, del/ins	x		x	x			Adolescence	Incomplete Penetrance 50%. One family with large del and severe and atypical form of dystonia.	Cranial or generalized, with prominent tongue and laryngeal involvement	(34)f
TOR1A	**DYT-TOR1A**	AD (rarely de novo)	Mostly 3-bp del in exon 5 (c.907_909del GAG). Exonic SNV (>99%)	x		x	x			Childhood – adolescence; rarely adulthood	First cause of hereditary generalized dystonia (especially in AJ). Incomplete penetrance 35% (modified by the D216H polymorphism). Intrafamilial variability	Generalized dystonia, starting with actions in one limb, then progressively presenting at rest and spreading to other body parts.	(35–39)
VPS16	**DYT-VPS16**	AD, AR	SNV, dupl/del/ins, splice site	x		x	x			Childhood - adolescence	Incomplete penetrance.	Generalized dystonia starting in oromandibular, bulbar/cervical areas or in ULs, slowly progressively. +/- chorea, myoclonus, freezing, ID	(40–43)
EIF2AK2	**DYT-EIF2AK2**	AD (AR 1 case)	SNV	x		x	x			Childhood - adolescence	Incomplete penetrance. Variable expression. Allelic disorder to the Leukoencephalopathy, DD, and episodic neurologic regression	Focal/generalized dystonia. Slowly progressive, may result in gait difficulties, dysarthria/dysphagia. +/- mild DD, pyramidal signs	33236446

											syndrome (AD)		
***AOPEP*⁺**	**DYT-AOPEP**	AR	SNV, del, splice site	x		x	x			Childhood - early adulthood		Multifocal/generalized dystonia, with prominent orofacial, laryngeal involvement. Rarely associated with parkinsonism.	(44,45)
Combined dystonias (disorders where dystonia frequently coexists with other movement disorders)													
ATP1A3	**DYT/PARK-ATP1A3** **Rapid-onset Dystonia-Parkinsonism (RDP)**	AD (also de novo)	SNV, ins/del (26%: c.1838C>T) (80-90%)	x		x	x		del/dup analyses	Childhood to adulthood (4-45yo)	Incomplete penetrance. Allelic disorder to: ACH and CAPOS (AD). Variable expression: from only few features of one phenotype to intermediate overlapping phenotypes (+ACH and RPD).	Asymmetric dystonia-parkinsonism with abrupt onset (days-weeks), with later stabilization and slow progression. Cranio-caudal gradient: face/bulbar > UL > LL. Trigger: fever, trauma, exertion. +/- epilepsy, arrhythmia	(46)
TAF1	**DYT/PARK- TAF1** **Lubag syndrome**	XL-R	5 disease-specific changes: 4 SNP and a 48-bp (22); SVA retrotransposon ins (100%)	x		x	x			Adulthood	Only in Filipino descent. Female carriers are mostly asymptomatic, a small minority may have dystonia, parkinsonism or chorea (likely for an extreme X inactivation)	Dystonia-parkinsonism, typically starting with mild parkinsonism, then worsening to a multifocal/generalized dystonia. Rarely pure mild parkinsonism.	(47,48)
GCH1	**DYT/PARK-GCH1** **Dominant DRD / Segawa syndrome**	AD	SNV, ins/del splice site (87%)	x		x	x			Childhood	Penetrance females 87%, males 38%. Variable expression**:** from subtle to severe phenotype	Dystonia-parkinsonism: dystonia, typically starting in LL (equinovarus foot) and gradually progressing to generalized, with later parkinsonism, often pyramidal signs. Diurnal fluctuation. Responsive to low dose L-Dopa.	(49–51)
			large del of multiexons - whole gene (CNV) (13%)				x		del/dup analyses				
TH	**DYT/PARK-TH** **Recessive DRD / Segawa syndrome**	AR	SNV, del, splice site (99%)	x		x	x			Childhood - adolescence	Variable expression**:** from mild to severe phenotype	Broad spectrum: DRD (mild), infantile parkinsonism with DD (severe), infantile encephalopathy (very severe). Rarely can present with myoclonus-dystonia	(52–54)
			Large del (1-2 exons, promotor-exon 1) (1%)				x		MLPA				
DNAJC12	**DYT-DNAJC12** **Hyperphenylalaninemia, mild**	AR	SNV, small del, splice site	x		x	x			Infantile - adulthood	Variable expression (in homozygous twins)	Dystonia-parkinsonism: DD, ID, infantile dystonia, young-onset Dopa-responsive parkinsonism. **↑ serum phenylalanine** (NBS). Treatment: L-Dopa, 5-HTP, BH4	(55)
			CNV del (up to 6.9 kb)				x		MLPA, CMA				
SLC39A14	**DYT-SLC39A14** **Hypermanganesemia with dystonia-2**	AR	SNV, del, splice site (100%)	x		x	x			Infantile - childhood		Dystonia-parkinsonism: DD, spasticity, bulbar dysfunction, severe generalized dystonia, parkinsonism. **Brain MRI:** globus pallidus T_1 hyper- / T_2 hypo-intensity; ↑↑↑ manganesemia	(56)
KCTD17	**MYC/DYT-KCTD17**	AD	SNV, splice site	x		x	x			Childhood - adolescence		Dystonia with myoclonus: early onset myoclonus, later in life generalized dystonia (+ cranial/cervical). Rarely (2 family): DD, ID, severe lingual dystonia	(57)
SGCE	see Supplementary Table 4											Myoclonus-dystonia.	

GNAO1	**DYT/CHOR-GNAO1**	AD (typically de novo)	SNV, splice site, del (98.5%)	x		x	x			Infancy - adulthood	Variable expression**,** from mild to severe	Broad spectrum: Dystonia (from mild to *status dystonicus*), choreoathetosis, persistent or paroxysmal. +/- DD/ID, epilepsy	(58–60)
			CNV del (intragenic, involving the whole gene or contiguous genes) (1.5%)				x		del/dup analyses: MLPA, CMA				
COX20 (FAM36A)	**DYT-COX20 - Deficiency of cytochrome c oxidase 20**	AR	SNV, splice site	x		x	x			Infancy - childhood	No clear genotype/phenotype correlation	DYTCA: predominant dystonia and mild cerebellar ataxia. Severe forms: DD	(61,62)
Complex dystonias (dystonia dominates the clinical picture but occurs in the context of a complex phenotype including symptoms other than movement disorders) and													
DCAF17, PANK2, PLA2G6, CP; FA2H FTL, c19orf12 WDR45 *	**NBIA**	AR, AD (FTL), AD/AR (c19orf12), XL-D (WDR45)	SNV, del/ins/dupl, splice site	x		x	x			Infancy - adulthood	Intrafamilial variability	Dystonia +/- other movement disorders, DD, psychiatric features. Other signs: ataxia, hearing loss, retinal degeneration, hypogonadism, diabetes. **Brain MRI:** basal ganglia T_1 hyperintensity; atrophy (cortex, cerebellum)	(63)
			CNV (del single exons-multiple continuous genes)				x		del/dup analyses: MLPA, CMA				
ATP7B	**Wilson disease**	AR	SNV, del/ins, splice site (98%)	x		x	x			Childhood - elderly (70 yo)	Targeted analysis can be performed first in individuals from populations with known founder variants AJ, Canary Islands, Druze, Sardinia.	Combination of hepatic (fatty liver), psychiatric or neurological signs (movement disorders, ataxia, dysarthria, dementia) **Brain MRI:** putamina T2 hyperintensity ↓ serum ceruloplasmin & copper, ↑ 24-hr urinary copper	(64)
			rare exon/multiexons del/dupl				x		del/dup analyses: MLPA, LRS, long range PCR				
SLC30A10	**Hypermanganesemia with dystonia-1**	AR	SNV, small del/ins (96%)	x		x	x			Childhood - adulthood		Childhood: four-limb dystonia, "cock-walk gait", dysarthria, tremor, bradykinesia. Adulthood: parkinsonism (L-Dopa unresponsive). Other signs: polycythemia, hepatomegaly. **Brain MRI:** basal ganglia, subthalamic /dentate n. T_1 hyperintensity (thalamus and pons spared); ↑↑↑ manganesemia	(65–67)
			Large del (multiple exons) (4%)				x		del/dup analyses: MLPA, LRS, long range PCR				
MECR, SUCLA, ACAT1, OPA1, TIMM8A, mt-ND6	**Mitochondrial disorders**	AR, AD/AR (OPA1), XL-R (TIMM8A), mtDNA	SNV, del, splice site	x		x	x			Infancy - childhood		Various movement disorders (+dystonia), psychiatric abnormalities, ID, seizures, migraine, myopathy, neuropathy, optic/hearing loss. Dysmorphic features, cardiomyopathy, metabolic/endocrine disorders. MELAS, LHON, Leigh syndrome.	(68)i
			CNV (multi-exons/genes del)				x		del/dup analyses: MLPA, CMA				
			SNV mt-DNA					x			Maternal inheritance. Incomplete penetrance, phenotypic variability due to heteroplasmy.		
PTS, MUT, SERAC1, GCDH, PCCA/PCCB; DDC,	**Inborn error of metabolism of aminoacids and monoamine neurotransmitter**	AR	SNV, del/ins, splice site	x		x	x		x	Infancy - childhood		Various movement disorders (+ dystonia), DD/ID, psychiatric abnormalities, seizures. Systemic signs. Often detectable with NBS.	(69,70)

QDPR, SPR												**SPR "dopa responsive dystonia"**	
SLC6A3	**Dopamine Transporter Deficiency Syndrome**	AR	SNV, small del, splice site (95%)	x		x	x			Infancy (classic) - childhood --> adulthood (atypical later onset)	Protection against nicotine dependence	Typical: DD, hyperkinesia or parkinsonism, bulbar and eye movements abnormalities (including oculogyric crises). Atypical later onset: ADHD, tremor, parkinsonism-dystonia, dysarthria.	(71)
			CNV (multiexon del), translocation e ncompassing SLC6A3				x		del/dup analyses: MLPA, CMA				
SLC19A3	**Biotin-responsive basal ganglia disease (BTBGD)**	AR	SNV, del/dup, splice site (95%)	x		x	x			Childhood - adulthood	Biallelic predicted loss-of-function variants are more likely to present early and develop into the severe Leigh-like phenotype. Compound heterozygosity for one missense variant and one predicted loss-of-function variant has been associated with the classic childhood form of BTBGD	Dystonia, Parkinsonism. Classic form in childhood: subacute encephalopathy/coma (often triggered by febrile illness), cranial nerve palsy, seizures. Infancy: Leigh-like syndrome. Adulthood: Wernicke-like encephalopathy. Responsive to thiamine.	(72,73)
			45-kb del in 5'-UTR				x		del/dup analyses: MLPA, CMA				
TUBB4A	**TUBB4A-DYT**	AD	SNV (100%)	x		x	x			Early childhood - early adulthood	Variants consistently associated with phenotypes. Penetrance 100%. Variable clinical and radiological expression (MRI: hypomyelination with atrophy of the basal ganglia and cerebellum or isolated hypomyelination).	Isolated dystonia. Whispering dysphonia, spreading to the neck or limbs, +/- ataxic gait. MRI is normal.	**(74,75)**
	TUBB4A-Related Leukodystrophy	AD de novo										Hypomyelinating leukodystrophy-6: pyramidal and extrapyramidal signs (dystonia, perioral dyskinesia, choreoathetosis, oculogyric crisis), cerebellar and bulbar dysfunction, altered cognition. **Brain MRI:** Hypomyelination with atrophy of basal ganglia and cerebellum	
VPS11	**VPS11-related dystonia**	AR	SNV, CNV, splice site	x		x	x		del/dup analyses: MLPA, CMA	Childhood - adolescence	Part of HOPS complex. Biallelic LOF variants.	Complex dystonia with neurodevelopmental features. May present with dystonia, myoclonus, spasticity, cerebral atrophy, neuropathy and overlap with lysosomal storage phenotypes	42
VAC14	**Striatonigral degeneration**	AR	SNV, splice site	x		x	x			Childhood		Striatonigral degeneration, dystonia-parkinsonism, ataxia, dysarthria, hypotonia	(76)
ADAR1	**Aicardi-Goutieres syndrome (AGS)**	AR	SNV, del (100%)	x		x	x			Infancy - early adulthood	A dominant p.Gly1007Arg variant in AGS has been reported	Encephalopathy often with dystonia, DD/ID, intermittent sterile pyrexias	(77,78)
	Dyschromatosis symmetrica hereditaria	AD										Dystonia, ID. Hyper-/Hypo-pigmented macules on face and extremities	(79)
IRF2BPL	**Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures**	AD (de novo)	SNV, del/ins	x		x	x				Intronless gene	DD, hypotonia, seizures, pyramidal signs, dysarthria	(80)
FOXG1	**FOXG1 Syndrome**	AD (also de novo)	SNP, del (>95%)	x		x	x			Childhood	Del/intragenic pathogenic variants vs large del/dup (whole	Rett-like phenotype: DD, ID, hyperkinetic movements,	(81–83)

			CNV (large del, whole gene dup) (<5%)				x		del/dup analyses: MLPA		gene / contiguous gene): different phenotypes. Reported mosaicism.	stereotypies, psychiatric manifestations, impairment of social interaction	
HPRT	**Lesch-Nyhan syndrome**	XL	SNP, splice site, del/ins/dup (80%)	x		x	x			Childhood	Heterozygous females are clinically normal; rarely LND is observed for skewed X-chromosome inactivation	Dystonia, chorea, occasionally ballism, DD/ID, eye movement abnormalities, spasticity, compulsive self-injurious behavior (resembling cerebral palsy). Additional clinical features: hyperuricemia, crystalluria, gouty arthritis, nephrolithiasis, renal failure. **↓↓↓ HGprt enzyme activity**	(84,85)
			CNV (5'UTR, one exon, multiexons) (20%)				x		del/dup analyses: MLPA, CMA				
GLB1	**GM1 gangliosidosis** (lysosomal storage disorder)	AR	SNV, del, intronic SNV (>99%)	x		x	x			Infancy - adulthood	Variable expression: type I, earlier onset and more severe phenotype. Type III: childhood to III decade.	Type III: generalized dystonia, gait and speech problems, akinetic rigid parkinsonism, cardiomyopathy, cognitive decline. **↓ beta-galactosidase enzyme activity**	(86)
			CNV (del) (<1%)						del/dup analyses: MLPA				
SQSTM1	**Neurodegeneration with ataxia, dystonia, and gaze palsy (NADGP)**	AR	SNV, del/ins, splice site	x		x	x				Allelic disorder to Frontotemporal Dementia and/or Amyotrophic Lateral Sclerosis 3	Childhood-onset neurodegeneration, gait ataxia, cognitive decline, oculomotor abnormalities including vertical gaze palsy and nystagmus, and hypergonadotropic hypogonadism	(87)
ACTB	**Dystonia-deafness**	AD (mainly de novo)	SNV (100%)	x		x	x			Adolescence - adulthood		Sensory hearing loss, generalized dystonia, skeletal abnormalities. +/- DD, seizures	
BCAP31		XL-R	SNV, splice site	x		x	x			Infancy		Sensorineural hearing loss. generalized dystonia. Central hypomyelination, ID, microcephaly, ophthalmoplegia	(88)
			CNV (del 5.3 kb)				x		del/dup analyses: MLPA, CMA				
FITM2		AR	SNP, dup	x		x	x			Childhood		Sensorineural hearing loss. Generalized dystonia. Other signs: DD, poor growth	(89)
ATXN3*	**SCA-3** (see Supplementary Table 3)	AD	exonic CAG expansion		x		x		LRS	Adult	Penetrance depending on number of expansions. Anticipation, especially with paternal transmission.	May have dystonia, parkinsonism >> ataxia. Altered eye movements (+horizontal), lower motoneuron involvement, neuropathy	(90)
KIF1C*	**HSP/ATX-KIF1C SPASTIC ATAXIA 2**	AR	SNP, splice site	x		x	x			Adulthood		Pure and complicated; variable additional features including dystonia, ataxia, chorea, myoclonus, dysarthria, developmental delay, mild mental retardation, hypodontia, ptosis, short stature, sensorineural deafness, pes planus, white matter lesions	(91)
			CNV (del multiexons)				x		del/dup analyse: MLPA, CMA				
DNAJC6*	**PARK-DNAJC6**	AR	SNP, del splice site (95%)	x		x	x			Early adulthood (<21 years)		Atypical parkinsonism: DD/ID, parkinsonism, dystonia, spasticity, seizures, bulbar/gastrointestinal disfunction	(92,93)
			CNV (del 80 kb) (5%)				x		del/dup analyse: MLPA,				

									CMA				
Other candidate genes associated with dystonia													
NUP54 (2023)	**Dystonia with striatonigral degeneration**	AR	SNV, del	x		x	x			Childhood - adolescence		Generalized dystonia, DD, limb-choreoathetoid and/or ataxic movements, dysarthria, dysphagia. **Brain MRI:** T2/FLAIR hyperintensities in basal ganglia	(94)
NUP62 (2006)	**Dystonia with striatonigral degeneration**	AR	SNV	x		x	x			Infancy		Choreoathetosis, dystonia, and abnormal high T2-weighted MRI signals in the striatum	(95)
DRD1 (2023)	**-**	AR	SNV	x		x	x			Infancy		Severe infantile parkinsonism-dystonia, oculogyric crises, dysautonomia. severe DD	(96)
DRD2 (2021)	**-**	AD	SNV	x		x	x			Childhood - Early adulthood		Focal or multifocal Dystonia, generalized chorea. Psychiatric symptoms	(97)
ATP5F1B (2023)	**-**	AD	SNV (2 families)	x		x	x			Infancy - adolescence	Encoding a subunit of the mitochondrial ATP synthase	Early-onset isolated dystonia	(98)

Supplementary Table 2. Most relevant genetic causes of dystonic syndromes and related test selection. For each listed gene, disease nomenclature, reported genetic variants are listed, appropriate diagnostic test selection (based on described genetic variants), important genetic highlights, and main clinical traits are reported. Percentage values reported represent the amount of pathogenetic variants detected with the specific analysis or gene-targeted deletion/duplication analysis on GeneReviews® when available/applicable. **A**: adenine. **ACH**: alternating hemiplegia of childhood; **AD**: autosomal dominant; **AJ**: Ashkenazi Jews; **AOO**: age of onset. **AR**: autosomal recessive; **C**: cytosine. **CAPOS**: cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss. **CMA**: Chromosomal microarray analysis. **CNV**: deletion/insertion/duplication of > 50 base pairs. **DD**: developmental delay. **del/ins/dup**: deletion/insertion/duplication < 50 base pairs. **del/dup analyses**: gene-targeted deletion/duplication analysis (methods used may include a range of techniques such as quantitative PCR, long-range PCR (**LR-PCR**), multiplex ligation-dependent probe amplification (**MLPA**), and a gene-targeted microarray designed to detect single-exon deletions or duplications). **DRD**: Dopa responsive dystonia. **Expansion**: repeat expansion tests (method used may include long-range PCR (**LR-PCR**), repeat-primed PCR (**RP-PCR**). **G**: guanine. **HC**: healthy controls. **Her**: inheritance. **ID**: intellectual disability. **LHON**: Leber hereditary optic neuropathy. **LL**: lower limb. **MELAS**: Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes. **mtDNA**: mitochondrial DNA tests. **NBS**: new born screening. **NGS**: next generation sequencing. **Rpt**: repeats. **SNV**: single nucleotide variant. **Splice site**: splice site variant. **SVA**: SINE-VNTR-Alu. **T**: thymidine. **UL**: upper limb. **UNK**: no data available. **WES**: whole exomes sequencing. **WGS**: whole genome sequencing. *****: Disorders that usually present with other phenotypes but can have predominant dystonia.

ATAXIA

Gene	Condition	Her	Variants	NGS panel	Expansion	WES	WGS	mtDNA	Other	AOO	Genetic tricks	Clinical traits	Ref
ATXN1, ATXN2, ATXN3, ATXN7, ATXN8, PPP2R2B	SCA1,2,3,7,8, 12	AD	Exonic CAG expansion (SCA12: promoter) (100%)		x		x		LRS	Adults	Anticipation (>> with parental transmission)	Different additional features in different subtypes (neuropathy: SCA1; slow horizontal saccades: SCA2; parkinsonism and dysautonomia: SCA1,2,3; spasticity: SCA3; prominent tremor: SCA12)	(99,100)
ATXN10	SCA10	AD	Intronic ATTCT expansion (100%)		x		x		LRS	Adults	Anticipation (>> with parental transmission), Mexican and Brasilian	Cerebellar symptoms and seizure	(101)
TBP	SCA17	AD	Exonic CAG or CAA expansion (100%)		x		x		LRS	Adults	Anticipation (>> with parental transmission)	Great mimic: chorea, motoneuron disease, cognitive impairment	(102)
NOP56	SCA36	AD	Intronic GGCCTG expansion (100%)		x		x		LRS	Adults	Anticipation (>> with parental transmission)	Associated with motoneuron symptoms	(103)
BEAN1	SCA31	AD	(TAGAA)n repeat (100%)		x		x		LRS	Adults	Anticipation (>> with parental transmission), common in Japan	Variable presence of hearing loss, common in Japan	(104)
CACNA1A	SCA6, EA2	AD	CAG expansion		x		x		LRS	Childhood/ adolescence		SCA, episodic ataxia type 2 (prolonged attacks of cerebellar symptoms and blurred vision)	(105)
			SNV	x		x	x						
KCNA1, CACNB4, SLC1A3, SCN2A	EA1, EA5, EA6, EA9	AD	SNV, in/del	x		x	x			Childhood/ adolescence		Brief (EA1,EA9) or prolonged (EA5,EA6,EA9) attacks of cerebellar symptoms, dizziness, blurred vision, nausea/vomiting, possible epilepsy	(106)
RFC1	CANVAS	AR	Intronic expansions (100%)		x		x		LRS	Adults		Neuropathy, vestibular areflexia	(107)
FGF14	SCA27B	AR	GAA Intronic expansions (100%)		x		x		LRS	Adults		Downbeat ny, episodic symptoms	(108,109)
ATN1	DRPLA	AD	CAG expansion (100%)		x		x		LRS	Childhood to adulthood	Age and CAG expansion correlation (<21 y: 63-79; 21-40y: 61-69; >40y: 48-67)	Early onset: seizure, ataxia, myoclonus, ID Late onset (>20y): ataxia, choreoatetosis, psychiatric symptoms and dementia	(110,111)
FMR1	FXTAS	X-linked	CGG repeats (100%)		x		x		LRS	Adults	Premutation alleles (55-200	Changes in personality and psychiatric	(112)

											repeats)	symptoms. Women: rare (with POCS)	
Mitochondrial DNA	NARP, MERRF, KSS, Leigh syndrome							x			Maternal inheritance	Myopathy, neuropathy, hearing loss, optic atrophy, dementia, and short stature, myopathy, neuropathy, hearing loss, optic atrophy, dementia, and short stature	(113)
TTPA	AVED	AR	SNV (>97%)	x		x	x			I-II decade	One case reported of whole gene deletion	Cerebellar features plus areflexia and loss of proprioception, Babinski sign, macular degeneration and pigmentary retinopathy. Blood work: low levels of vitamin E	(114,115)
			SVs/CNV	x			x		del/dup analyses: MLPA, LRS, long range PCR				
APOB, MTTP	Ataxia and vitamin E deficiency	AR	SNV, in/del	x		x	x			I-II decade	*MTTP*: small indels more frequent than missense variants	Neuropathy, retinopathy, acanthocytosis, hepatomegaly, deficiency of fat-soluble vitamins	(116)
FXN	Friedreich ataxia (FRDA)	AR	GAA repeats (96%)		x		x		LRS	Usually < 25y (25-39y: LOFA; >40y: VLOFA)	5-33 GAA: normal; 34-65: premutation; >66: full penetrance	Reduced reflexes, positive Babinski sign; hypertrophic cardiomyopathy; DM; hypoacusia. Late onset forms can have present reflexes	(117–120)
			SNV	x		x	x			4% cases (mostly compound heterozygous with expansion of the other allele)			
			SVs/CNV				x		del/dup analyses: MLPA, LRS, long range PCR	Rare cases			
APTX, SETX, ATM	AOA	AR	SNV (90-95% *ATM*, 80-92% *SETX*)	x		x	x		LRS	Childhood to adulthood	*ATM*: deep intronic variants reported	Oculomotor apraxia, dystonia, increased AFP (*ATM*), hypoalbuminemia (*APTX*), increased cancer risk (*ATM*)	(121–125)
			SVs/CNV (5-10% *ATM*, 8-20% *SETX*)	x		x	x		del/dup analyses: MLPA, LRS, long range PCR				
SACS	Charlevoix-Saguenay	AR	SNV (95%)	x	x	x				Adults	Frequent in French Canadians	Ataxia and spasticity	(126)
			CNV (5%)				x		del/dup analyses:				

									MLPA, LRS, long range PCR, CMA				
Paraplegin	SPG7	AR,	SNV (>98%)	x		x	x			Adults		Ataxia and spasticity	(99)
			SVs/CNV (2%)	x		x	x		del/dup analyses: MLPA, LRS, long range PCR, CMA				
ANO10	SCAR10	AR	SNV, in/del (100%)	x		x	x			III-V decade		Reported spasticity, neuropathy, reduced levels of CoQ10	(127,128)
SYNE1	SCAR8	AR	SNV (~100%)	x		x	x			Childhood to adulthood	Multiple isoforms with variable tissue expression (Variants causing SCAR8 involve CNS isoform); frequent intronic variants affecting the splicing	Hotspot in Quebec, Canada (where it manifests as pure, slow progressive cerebellar ataxia). Other cases are associated with spasticity and brainstem dysfunction. Allelic with Arthrogryposis multiplex congenita 3 and Emery-Dreifuss muscular dystrophy 4	(129–132)
			CNV				x		del/dup analyses: MLPA, LRS, long range PCR, CMA				
ITPR1	SCA15 and 29	AD	SNV	x		x	x			Adult (SCA15) and childhood (SCA29)	Allelic with Gillespie syndrome (AD/AR)	SCA15: adult-onset, pure ataxia, slowly progressive; SCA29: non-progressive, infantile onset, with possible cognitive impairment	(133,134)
			SVs/CNV				x		del/dup analyses: MLPA, LRS, long range PCR				
KCND3	SCA19/22	AD	SNV, in/del	x		x	x			Childhood to adulthood	Loss of function pathogenic variants are associated with SCA19/22, gain of function pathogenic variants are associated with Brugada syndrome	Ataxia with possible cognitive deficit/developmental delay, other movement disorders	(135,136)
SPTBN2	SCA5 / SCAR14	AD/AR	SNV	x		x	x			I-V decade	Anticipation has been reported	Ataxia, possible cognitive impairment, downbeat nystagmus, spasticity	(137,138)
			SVs/CNV				x		del/dup analyses: MLPA, LRS, long range PCR				

AFG3L2	SCA28/SCAR5	AD/AR	SNV, in/del (>99%)	x		x	x			Childhood to adulthood	Deletions and duplications are very rare	Associated with mitochondrial features (PEO, ptosis) and spasticity	(139)
			CNV				x		del/dup analyses: MLPA, LRS, long range PCR, CMA				
STUB1	SCA48/SCAR16	AD/AR	SNV, in/del	x		x	x			Mid-adulthood (SCA48); I-II decade (SCAR16)		SCA48: ataxia, cognitive impairment, psychiatric symptoms and other movement disorders; SCAR16: ataxia, neuropathy, hyperreflexia, some with gonadal dysfunction and /or cognitive impairment	(140,141)
DAB1	SCA37	AD	ATTTC(n) repeat insertion (100%)		x				LRS, PCR, Southern blot	Adult	ATTTC(n) insertion in 5'-UTR of *DAB1* intron 3 or in noncoding regulatory region of *DAB1*	Pure ataxia	(142)
ADCK3	SCAR9	AR	SNV, in/del	x		x	x			Childhood		Primary coenzyme Q10 deficiency-4 (COQ10D4), Ataxia with possible epilepsy and cognitive deficits	(99,143)

Supplementary Table 3. Most relevant genetic causes of ataxia syndromes and related test selection. For each listed gene, disease nomenclature, reported genetic variants are listed, appropriate diagnostic test selection (based on described genetic variants), important genetic highlights, and main clinical traits are reported. Percentage values reported represent the amount of pathogenetic variants detected with the specific analysis or gene-targeted deletion/duplication analysis on GeneReviews® when available/applicable. **AD**: autosomal dominant; **AFP**: alpha fetoprotein; **AOA**: ataxia with oculomotor apraxia; **AVED**: ataxia with vitamin E deficiency; **AR**: autosomal recessive; **CMA**: Chromosomal microarray analysis. **CNV**: deletion/insertion/duplication of > 50 base pairs; **del/ins/dup**: deletion/insertion/duplication < 50 base pairs. **del/dup analyses**: gene-targeted deletion/duplication analysis (methods used may include a range of techniques such as quantitative PCR, long-range PCR (**LR-PCR**), multiplex ligation-dependent probe amplification (**MLPA**), and a gene-targeted microarray designed to detect single-exon deletions or duplications); DM: diabetes mellitus; **FXTAS**: Fragile X-associated tremor/ataxia syndrome; **KSS**: Kearns-Sayre syndrome; **LRS**: long read sequencing; **MERRF**: myoclonic epilepsy with ragged-red fibers; **NARP**: Neuropathy, Ataxia, Retinitis Pigmentosa; **NGS**: next generation sequencing; **PEO**: progressive external ophthalmoplegia; **POCS**: premature ovarian failure; **SCA**: spinocerebellar ataxia; **SCAR**: spinocerebellar ataxia recessive; **SNV**: single nucleotide variant; SVs: structural variants; **WES**: whole exomes sequencing. **WGS**: whole genome sequencing.

MYOCLONUS

Gene	Condition	Her	Variants	NGS panel	Expansion	WES	WGS	mtDNA	Other	AOO	Genetic tricks	Clinical traits	Ref
CSTB	Unverricht-Lundborg disease (EPM1)	AR	CCC-CGC-CCC-GCG expansion (90%)		x		x		LRS	Late childhood or adolescence	2-3 repeats: normal; 12-17 repeats: uncertain: ≥ 30 repeats: full penetrance (4-11 and 18-29 not been observed). High incidence in Finland and North Africa	Frequent generalized seizures + cerebellar signs +/- cognitive decline/dementia Compound heterozygous may cause more severe phenotype, mostly in makes	(144–148)
			SNV (10%)	x		x	x						
Laforin (EPM2A), NHLRC1 (Malin, EPM2B)	Lafora disease (EPM2)	AR	SNV (85%-90% EPM2A, >90% *NHLRC1*)	x		x	x			Adolescence		Lafora disease: occipital and generalized seizures, Lafora bodies (polyglucosans or glycogen) on skin biopsy	(149,150)
			SVs/CNV (10-15% EPM2A, <10% *NHLRC1*)				x		del/dup analyses: MLPA, LRS, long range PCR				
MT-TK (90%) MT-TF, -TH, -TI, -TL1, -TP, TS1, -TS2	Myoclonic epilepsy with ragged-red fibers (MERRF)	MT	SNV					x		Childhood to adulthood	Consider testing different tissues to account for heteroplasmy	Myopathy, neuropathy, hearing loss, optic atrophy, dementia, and short stature, myopathy, neuropathy, hearing loss, optic atrophy, dementia, and short stature	(151,152)
ATN1	DRPLA	AD	CAG expansion (100%)		x		x		LRS	Childhood to adulthood	Age and CAG expansion correlation (<21 y: 63-79; 21-40y: 61-69; >40y: 48-67)	Early onset: seizure, ataxia, myoclonus, ID Late onset (>20y): ataxia, choreoatetosis, psychiatric symptoms and dementia	(111,153)
NEU1	Sialidosis type 1	AR	SNV	x		x	x			II-III decade		Cherry red spot myoclonus (+ ataxia), Neuroaminidase activity on leucocytes	(154)
GBA	Gaucher disease type 3	AR	SNV (99%)	x		x	x						
			SV (complex rearrangement *GBA-GBAP1*) (1%)						Targeted gene sequencing for complex arrangements and del/dup analyses: MLPA, LRS, long range	I decade	*GBAP1* (pseudogene) has >96% homology with *GBA1*	Oculomotor apraxia (mostly horizontal gaze), systemic symptoms (hepatomegaly, splenomegaly, bone disease, cytopenia, pulmonary disease)	(155)

									PCR, CMA				
SCARB2/ LIMP2	Action myoclonus renal failure syndrome (EPM4)	AR	SNV (100%)	x		x	x			Childhood to adulthood		Renal failure due to steroid-resistant nephrotic syndrome (not present in 100% cases)	(156,157)
PRICKLE1	PRICKLE1-PME with ataxia (EPM5)	AR (AD)	SNV (100%)	x		x	x			I decade		Heterozygous patients with epilepsy, autism, developmental delay and brain malformation have been reported	(158–162)
GOSR2	North see PME with ataxia (EPM6)	AR	SNV, in/del	x		x	x			< 1 year	G144W most frequent variant	Loss of ambulation in the II decade; cognition may be initially spared	(163)
ASAH1	SMA-PME	AR	SNV (92%)	x		x	x			Childhood	Allelic with Farber disease (neonatal onset of painful joint deformity, subcutaneous nodules, granulomas of larynx and epiglottis, life expectancy <2 years)	Proximal weakness, respiratory difficulties, possible ID	(164)
			SVs/CNV (8%)	x		x	x		del/dup analyses: MLPA, LRS, long range PCR				
DHDDS	CDG	AR	SNV	x		x	x			Childhood		Variable phenotype, including: ataxia, ID, PME, facial, myoclonus, dystonia, chorea	(165)
NUS1	CDG, PMA	AR, AD	SNV, in/del	x		x	x		del/dup analyses: MLPA, LRS, long range PCR	Childhood		Variable phenotype, including: ataxia, ID, PME, facial, myoclonus	(165–167)
SGCE	Myoclonus-dystonia	AD	SNV, in/del (75%)	x		x	x			Childhood	Paternal inherited due to maternal imprinting (5% imprinting escape)	Shivering myoclonus, Alcohol response, OCD/anxiety Silver-Russel syndrome (myoclonus/dystonia + growth retardation and dysmorphism)	(168–174)
			SVs/CNV (25%)	x		x	x		del/dup analyses: MLPA, LRS, long range PCR				
KCTD17, ANO3, ADCY5, ATP1A3, YY1, VPS16	Dystonia with myoclonus	AD/AR	SNV	x	x	x				Childhood to adulthood	SVs (including CNV: microdeletion) reported for *VPS16*	Various degrees of combination of dystonia (mostly upper limbs and neck) and myoclonus (mostly cortical)	(43,175)
			SVs/CNV (*VPS16*)	x		x	x		del/dup analyses: MLPA, LRS, long range PCR				
HTT	Huntington disease	AD	CAG expansion (100%)		x		x			Adulthood > childhood	Anticipation (< 26 CAG expansion: normal; 27-35:	Classic form: chorea, dementia, psychiatric symptoms. Juvenile form Westphal	(176,177)

											intermediate; >46: pathogenic)	variant): rigid-hypokinetic	
SCA20, *GFAP* , *NF, POLG, GM2, CYP27A1*	PAPT	AD/AR	SNV, in/dell	x		x	x			Adulthood	SCA20: identification of a duplicated region that segregates with the disease but no specific gene has been identified yet	Adult onset of palatal tremor (myoclonus) and associated with ataxia; brain MRI: hyperintensity of the olives. *GFAP*: associated with Alexander disease; POLG: possible mitochondrial features; *NF*: iron accumulation at brain MRI; *GM2*: cherry red spot (retina) and neuropathy; *CYP27A1* associated with CTX: tendon xanthomas, cataracts, diarrhea, neonatal jaundice, brain MRI: T2 hyperintensities of the dentate nuclei, basal ganglia, cerebral peduncles, cerebellar white matter, and periventricular white matter	(63,178–183)
			SVs/CNV (duplication 11q12.2-11q12.3 In SCA20)				x		CMA, MLPA, LRS, long range PCR, FISH				
SAMD12, STARD7, MARCHF6, YEATS2, TNRC6A, RAPGEF3	Cortical tremor	AD	Intronic expansion				X*		RP-PCR, LRS	Adulthood		Family history of cortical myoclonus, epilepsy in at least one family member, AD inheritance	(184,185)
	Epileptic syndromes	AD/AR	SNV	x		x	x		Microarray	Childhood			(186)
			SVs/CNV	x		x	x		MLPA, LRS, long range PCR				
			Chromosomal abnormalities						CMA				

Supplementary Table 4. Most relevant genetic causes of myoclonic syndromes and related test selection. For each listed gene, disease nomenclature, reported genetic variants are listed, appropriate diagnostic test selection (based on described genetic variants), important genetic highlights, and main clinical traits are reported. Percentage values reported represent the amount of pathogenetic variants detected with the specific analysis or gene-targeted deletion/duplication analysis on GeneReviews® when available/applicable. *Not available diagnostically. **AD**: autosomal dominant; **AR**: autosomal recessive; **CDG**: congenital disorder of glycosylation; **CMA**: Chromosomal microarray analysis. **CNV**: deletion/insertion/duplication of > 50 base pairs; CTX: Cerebrotendinous Xanthomatosis; **del/ins/dup**: deletion/insertion/duplication < 50 base pairs. **del/dup analyses**: gene-targeted deletion/duplication analysis (methods used may include a range of techniques such as quantitative PCR, long-range PCR (**LR-PCR**), multiplex ligation-dependent probe amplification (**MLPA**), and a gene-targeted microarray designed to detect single-exon deletions or duplications); **ID**: intellectual disability; **EPM1/EPMI2A/EPM4/EPM5**: progressive myoclonic epilepsy, type 1/2A/4/5; **LRS**: long read sequencing; **NGS**: next generation sequencing; PAPT: progressive ataxia with palatal tremor; PMA: progressive myoclonus ataxia; **PME**: progressive myoclonic epilepsy; **SMA-PME**: spinal muscular atrophy-progressive myoclonic epilepsy; **SNV**: single nucleotide variant; **SVs**: structural variants; **WES**: whole exomes sequencing. **WGS**: whole genome sequencing.

PAROXYSMAL MOVEMENT DISORDERS (PMD)

Gene	Condition	Her	Variants	NGS panel	Expansion	WES	WGS	mtDNA	Gene-targeted del/dup analysis	AOO	Genetic tricks	Clinical traits	REF
Paroxysmal dyskinesia													
PRRT2	***PRRT2 disorders***	AD	SNV, splice site (>99%)	x		x	x		del/dup analyses (<1%)	Infancy - childhood (epilepsy), adolescence - adulthood (PKD or migraine)	Penetrance 50-90%. Intrafamilial and interfamilial variability. Biallelic variants (<1%): more severe phenotype.	Three core phenotypes (independent or overlapped): 1) Seizures: IC, BFIE; 2) PMD: +PKD, EA (rarely PED, PNKD); 3) Headache: migraine w/ or w/o aura, HM. Between 1-2: ICCA Responsive to CMZ.	(187)
MR1	***PKND***	AD	SNV, del (100%)	x		x	x			Childhood - adolescence (rarely adulthood)	Penetrance 98%	PKND. Triggers: coffee, tea, or alcohol, excitement, stress, or fatigue, or spontaneous	(188)
SLC2A1	***GLUT1 deficiency syndrome***	AD (90% de novo)	SNV, del (84%)	x		x	x				Complete penetrance. Variable expression with two phenotypes.	**Classic** 90%: epilepsy, DD, dysarthria, PEID with complex movement including ataxia, dystonia, chorea. **Non-classic** 10%, milder: PD with ataxia, choreoathetosis, dystonia, alternating hemiplegia. Responsive to ketogenic diet. **CSF glucose concentration <60 mg/dL**	(189,190)
			CNV (multiexon or whole-gene del) (13%)				x		del/dup analyses				
Episodic ataxia (EA)													
KCNA1	***EA Type 1***	AD	SNV (>90%)	x		x	x			Childhood - adolescence (<20yo)	Potassium channelopathy	**Brief** attacks (Secs-Mins) with variable symptoms including vertigo, blurred vision, diplopia, nausea, headache, diaphoresis, clumsiness, stiffening, dysarthria, difficulty in breathing. **Interictal** myokymia, episodes of spastic contractions of the muscles with loss of coordination/balance. +/- epilepsy, DD, ID.	(191)
CACNA1A	***EA Type 2***	AD	SNP, del/dup/ins, splice site (95%)	x		x	x			Childhood - adulthood	Phenotypic variability. Allelic disorder to: familial HM; SCA6 (due to repeated CAG expansions). *Progressive cerebellar ataxia +/- migraine*	**Long** attacks (mins-hours-days) **Interictal** nystagmus; +/- chronic ataxia and epilepsy, 50% migraine	(192)
			CNV (7-140 kb, multiexons)				x		del/dup analyses				(192)
			CAG expansion		x		x						(193)
unknown (locus 1q42)	***EA Type 3***	AD	unknown	NA						Childhood - adult		**Brief** attacks (mins), like EA1 plus tinnitus; +/- epilepsy	(194)
unknown (linkage excluded with EA1-2)	***EA Type 4***	AD	unknown	NA						Adulthood		**Long** attacks (hours), like EA2 without nystagmus; +/- epilepsy	(194)
CACNB4	***EA Type 5***	AD	SNV	x		x	x			Adulthood		**Long** attacks (hours), like EA2 **Interictal** nystagmus, epilepsy	(195)
SLC1A3	***EA Type 6***	AD	SNV	x		x	x			Childhood - adulthood		**Long** attacks (hours), like EA2 +/- migraine, alternating hemiplegia	(194)
unknown (Maps 19q13)	***EA Type 7***	AD	unknown	NA						unk		Long attacks (hours), like EA2 without nystagmus	(194)

UBR4	***EA Type 8?***	AD	SNV	x		x	x			Infancy		**Brief and long** attacks (mins-hours), overlaps with EA1 and 2, +/- myokymia or nystagmus, intentional tremor	(196)
FGF14	***EA Type 9?***	AD	SNV, ins	x		x	x			Childhood - adult	Phenotypic variability. Allelic disorder to: SCA27	**Brief and long** attacks (secs-days), like EA2 with tremor	(197)
			GAA expansion		x		x		LRS				
SCN1A		AD	SNV	x		x	x			Infancy - childhood	Allelic disorder to: Dravet syndrome and other epileptics syndromes, familial HM	**Brief and long** attacks (mins-hours), **Interictal** epilepsy, neonatal seizures	(194)
CACNA1G	***EA Type 10? or Episodic vestibulocerebellar ataxias***	AD	SNV	x		x	x			Adulthood	Allelic disorder to: SCA42	**Long** attacks (up to months), **Interictal** chronic ataxia; also: facial numbness, movement induced vertigo, bilateral vestibulopathy	(198)

Supplementary Table 5. Most relevant genetic causes of paroxysmal movement disorders and related test selection. For each listed gene, disease nomenclature, reported genetic variants are listed, appropriate diagnostic test selection (based on described genetic variants), important genetic highlights, and main clinical traits are reported. Percentage values reported represent the amount of pathogenetic variants detected with the specific analysis or gene-targeted deletion/duplication analysis on GeneReviews® when available/applicable. **A**: adenine. **AD**: autosomal dominant. **AOO**: age of onset. **AR**: autosomal recessive. **BFIE**: benign familial infantile epilepsy. **CNV**: deletion/insertion/duplication of > 50 base pairs. **del/ins/dup**: deletion/insertion/duplication < 50 base pairs. **del/dup analyses**: gene-targeted deletion/duplication analysis (methods used may include a range of techniques such as quantitative PCR, long-range PCR (**LR-PCR**), multiplex ligation-dependent probe amplification (**MLPA**), and a gene-targeted microarray designed to detect single-exon deletions or duplications). **EA**: Episodic ataxia. **Expansion**: repeat expansion tests (method used may include long-range PCR (**LR-PCR**), repeat-primed PCR (**RP-PCR**). **G**: guanine. **HM**: Hemiplegic migraine. **Her**: inheritance. **IC**: Infantile convulsions. **ICCA**: Infantile convulsions with paroxysmal choreoathetosis**. LRS**: long-reads sequencing. **mtDNA**: mitochondrial DNA tests; **NGS**: next generation sequencing. **PD**: paroxysmal dyskinesia. **PED**: Paroxismal exercise-induced dyskinesia. **PKD**: Paroxismal kinesigenic dyskinesia. **PNKD**: Paroxismal non kinesigenic dyskinesia. **SNV**: single nucleotide variant. **splice site**: splice site variant. **T**: thymidine. **UNK**: no data available. **WES**: whole exomes sequencing. **WGS**: whole genome sequencing.

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