TABLE 451e-1 CLASSIFICATION OF THE SPINOCEREBELLAR ATAXIAS (SCA) *(CONTINUED)*

Name	Locus	Phenotype
Baltic myoclonus (Unverricht-Lundborg) (recessive)	21q22.3; cystatin B; extra repeats of 12–base pair tandem repeats	Myoclonus epilepsy; late-onset ataxia; responds to valproic acid, clonazepam; phenobarbital
Marinesco-Sjögren syndrome (recessive)	5q31; SIL 1 protein, nucleotide exchange factor for the heat-shock protein 70 (HSP70); chaperone HSPA5; homozygous 4-nucleotide duplication in exon 6; also compound heterozygote	Ataxia, dysarthria; nystagmus; retarded motor and mental maturation; rhabdomyolysis after viral illness; weakness; hypotonia; areflexia; cataracts in childhood; short stature; kyphoscoliosis; contractures; hypogonadism
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	Chromosome 13q12; SACS gene; loss of sacsin peptide activity	Childhood onset of ataxia, spasticity, dysarthria, distal muscle wasting, foot deformity, retinal striations, mitral valve prolapse
Kearns-Sayre syndrome (sporadic)	mtDNA deletion and duplication mutations	Ptosis, ophthalmoplegia, pigmentary retinal degeneration, cardiomyopathy, diabetes mellitus, deafness, heart block, increased CSF protein, ataxia
Myoclonic epilepsy and ragged red fiber syndrome (MERRF) (maternal inheritance)	Mutation in mtDNA of the $tRNA^{lys}$ at 8344; also mutation at 8356	Myoclonic epilepsy, ragged red fiber myopathy, ataxia
Mitochondrial encephalopathy, lactic acidosis, and stroke syndrome (MELAS) (maternal inheritance)	$tRNA^{leu}$ mutation at 3243; also at 3271 and 3252	Headache, stroke, lactic acidosis, ataxia
Neuropathy; ataxia; retinitis pigmentosa (NARP)	ATPase6 (Complex 5); mtDNA point mutation at 8993	Neuropathy; ataxia; retinitis pigmentosa; dementia; seizures
Episodic ataxia, type 1 (EA-1) (autosomal dominant)	12p13; potassium voltage-gated channel gene, *KCNA1*; Phe249Leu mutation; variable syndrome	Episodic ataxia for minutes; provoked by startle or exercise; with facial and hand myokymia; cerebellar signs are not progressive; choreoathetotic movements; responds to phenytoin; genetic testing available
Episodic ataxia, type 2 (EA-2) (autosomal dominant)	19p-13 (*CACNA1A*) (allelic with SCA6) (α_{1A}-voltage–dependent calcium channel subunit); point mutations or small deletions; allelic with SCA6 and familial hemiplegic migraine	Episodic ataxia for days; provoked by stress, fatigue; with down-gaze nystagmus; vertigo; vomiting; headache; cerebellar atrophy results; progressive cerebellar signs; responds to acetazolamide; genetic testing available
Episodic ataxia, type 3 (autosomal dominant)	1q42	Episodic ataxia; 1 min to over 6 h; induced by movement; vertigo and tinnitus; headache; responds to acetazolamide
Episodic ataxia, type 4 (autosomal dominant)	Not mapped	Episodic ataxia; vertigo; diplopia; ocular slow pursuit defect; no response to acetazolamide
Episodic ataxia, type 5 (autosomal dominant)	2q22-q23; CACNB4β4 protein	Episodic ataxia; hours to weeks; seizures
Episodic ataxia type 6 with seizures, migraine, and alternating hemiplegia (autosomal dominant)	SLC1A3; 5p13; EAAT1 protein; missense mutations; glial glutamate transporter (GLAST); 1047 C to G; proline to arginine	Ataxia, duration 2–4 days; episodic hypotonia; delayed motor milestones; seizures; migraine; alternating hemiplegia; mild truncal ataxia; coma; febrile illness as a trigger; MRI: cerebellar atrophy
Episodic ataxia, type 7 (autosomal dominant)	19q13	Episodic ataxia; vertigo, weakness; less than 24 h
Episodic ataxia with paroxysmal choreoathetosis and spasticity (dystonia-9) (DYT9; CSE) (autosomal dominant)	1p	Ataxia; involuntary movements; dystonia; headache; spastic paraplegia; responds on occasion to acetazolamide
Fragile X tremor/ataxia syndrome (FXTAS) X-linked dominant	Xq27.3; CGG premutation expansion in *FMR1* gene; expansions of 55–200 repeats in 5′ UTR of the FMR-1 mRNA; presumed dominant toxic RNA effect	Late-onset ataxia with tremor, cognitive impairment, occasional parkinsonism; males typically affected, although affected females also reported; syndrome is of high concern if affected male has grandson with mental retardation (fragile X syndrome); MRI shows increased T2 signal in middle cerebellar peduncles, cerebellar atrophy, and occasional widespread brain atrophy; genetic testing available
Ataxia telangiectasia (autosomal recessive)	11q22-23; *ATM* gene for regulation of cell cycle; mitogenic signal transduction and meiotic recombination; elevated serum alpha-fetoprotein level; immunoglobulin deficiency	Telangiectasia, ataxia, dysarthria, pulmonary infections, neoplasms of lymphatic system; IgA and IgG deficiencies; diabetes mellitus, breast cancer; genetic testing available; chorea; dystonia
Early-onset cerebellar ataxia with retained deep tendon reflexes (autosomal recessive)	13q11-12	Ataxia; neuropathy; preserved deep tendon reflexes; impaired cognitive and visuospatial functions; MRI, cerebellar atrophy
Ataxia with oculomotor apraxia (AOA1) (autosomal recessive)	9p21; protein is member of histidine triad superfamily, role in DNA repair; elevation of serum LDL cholesterol and low serum albumin level; APTX, aprataxin	Ataxia; dysarthria; limb dysmetria; dystonia; oculomotor apraxia; optic atrophy; motor neuropathy; late sensory loss (vibration); genetic testing available
Ataxia with oculomotor apraxia 2 (AOA2) (autosomal recessive)	9q34; senataxin protein, involved in RNA maturation and termination; helicase superfamily 1; elevated serum alpha-fetoprotein level; SETX, senataxin	Gait ataxia; choreoathetosis; dystonia; oculomotor apraxia; neuropathy, vibration loss, position sense loss, and mild light touch loss; absent leg deep tendon reflexes; extensor plantar response; genetic testing available

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