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

Name	Locus	Phenotype
Cerebellar ataxia with muscle coenzyme Q10 deficiency (autosomal recessive)	9p13	Ataxia; hypotonia; seizures; mental retardation; increased deep tendon reflexes; extensor plantar responses; coenzyme Q10 levels reduced with about 25% of patients with a block in transfer of electrons to complex 3; may respond to coenzyme 10
Refsum's disease (autosomal recessive)	10pter; elevated serum phytanic acid level; phytanoyl-COH hydroxylase and PEX7	Retinitis pigmentosa; ataxia; sensorineural deafness; demyelinating neuropathy
Cerebrotendinous xanthomatosis (autosomal recessive)	2p33; elevated cholesterol level; CYP27; sterol 27 hydroxylase	Spastic ataxia; mental retardation; dementia; tendon xanthomas; diarrhea; cataracts
Joubert's syndrome (autosomal recessive)	9q34.3	Ataxia; ptosis; mental retardation; oculomotor apraxia; nystagmus; retinopathy; rhythmic tongue protrusion; episodic hyperpnea or apnea; dimples at wrists and elbows; telecanthus; micrognathia
Sideroblastic anemia and spinocerebellar ataxia (X-linked recessive)	Xq13; ATP-binding cassette 7 (ABCB7; ABC7) transporter; mitochondrial inner membrane; iron homeostasis; export from matrix to the intermembrane space	Ataxia; elevated free erythrocyte protoporphyrin levels; ring sideroblasts in bone marrow; heterozygous females may have mild anemia but not ataxia
Infantile-onset spinocerebellar ataxia of Nikali et al (autosomal recessive)	10q23.3-q24.1; twinkle protein (gene); homozygous for Tyr508Cys missense mutations	Infantile ataxia, sensory neuropathy; athetosis, hearing deficit, reduced deep tendon reflexes; ophthalmoplegia, optic atrophy; seizures; primary hypogonadism in females
Hypoceruloplasminemia with ataxia and dysarthria (autosomal recessive)	Ceruloplasmin gene; 3q23-q25 (trp 858 ter)	Gait ataxia and dysarthria; hyperreflexia; cerebellar atrophy by MRI; iron deposition in cerebellum, basal ganglia, thalamus, and liver; onset in the fourth decade
Spinocerebellar ataxia with neuropathy (SCAN1) (autosomal recessive)	Tyrosyl-DNA phosphodiesterase-1 (TDP-1) 14q31-q32	Onset in second decade; gait ataxia, dysarthria, seizures, cerebellar vermis atrophy on MRI, dysmetria
Cerebellar ataxia type 1 (autosomal recessive)	6p25 SCAR8; SYNE1; spectrin repeats-nuclear envelope 1	Pure ataxia
Cerebellar ataxia type 2 (autosomal recessive)	1q42;ADCK3 (CABC1); aarf-domain containing kinase 3; elevation of serum lactate and decreased coenzyme Q10 level	Ataxia; mental retardation; myoclonus; epilepsy; exercise intolerance; stroke or transient ischemic-like episodes
Niemann-Pick type C disease	18q11; NPCI; NPCH1 and 2; skin biopsy (filipin staining)	Ataxia; vertical supranuclear ophthalmoplegia; splenomegaly; dystonia; impaired cognition

Abbreviations: CSF, cerebrospinal fluid; CT, computed tomography; EMG, electromyogram; LDL, low-density lipoprotein; MRI, magnetic resonance imaging; REM, rapid eye movement; UTR, untranslated region.