On-line Table 1: Degenerative pattern

Disease	Posterior Fossa MRI Finding	Additional MRI Findings	Confirmatory Test
Friedrich ataxia	Normal, mild cord atrophy	CA (mild), T2 cord SC, iron deposition at DN	Genetic testing
AOA	CA (<vermis)	Reduced NAA and elevated mIns on MRS	Clinical and lab findings (albumin, cholesterol, α-fetoprotein)[g]
Ataxia-telangiectasia	CA (<vermis)	WM and BG lesions, spinal cord atrophy	Clinical, lab findings (α-fetoprotein), and molecular genetic testing
Infantile-onset spinocerebellar ataxia	CA	Brain stem and cord atrophy, cerebellum SC	Clinical and genetic testing
X-linked congenital ataxia[a]	CA	Variable	Genetic testing
Mitochondrial disorders[b]	CA	WM, BG, and cortex SC; restricted DWI; lactate on MRS	Clinical, neuroimaging, and genetic testing
Ataxia with vitamin E deficiency	Normal or mild CA	Posterior and lateral columns of the cord SC	Clinical, lab findings (vitamin E, lipoprotein, and acanthocytosis)[h]
Dentatorubral-pallidoluysian atrophy	CA and midbrain atrophy	Cerebral WM and brain stem SC, brain atrophy	Genetic testing
Cerebrotendinous xanthomatosis	CA and SC in the DN	WM, CST, BG, and brain stem SC; brain atrophy	Clinical and lab findings (cholestanol in serum and tendons)
Episodic ataxia (types I to VII)	CA (<vermis)	None	Clinical, neuroimaging, and genetic analysis
Infantile refsum disease	SC in the dentate nucleus	Brain atrophy; WM, CC, CST, and brain stem SC	Clinical and lab findings (phytanic and pristanic acid)
Infantile neuroaxonal dystrophy	SC and atrophy in the cerebellar cortex	Brain, WM, and optic nerve atrophy; Low T2 signal at GP	Clinical, neuroimaging, and genetic testing
Adrenomyeloneuropathy	SC in the cerebellar WM and CST	Brain involvement is less common than classic ADL	Clinical and lab findings (very long-chain fatty acids)
Aminoacidurias and organic acidurias[c]	CA in some subtypes	Variable	Clinical, neuroimaging, and genetic testing
HABC	CA (<vermis)	Diffuse hypomyelinization; BG, CST, and WM atrophy and SC	Clinical and neuroimaging
Gangliosidosis GM2[d]	CA (>vermis)	Reduced NAA at MRS in cerebellum and WM	Clinical and lab findings (β-hexosaminidase)
Neuronal ceroid lipofuscinoses[e]	CA	Brain atrophy, low T2 signal in the thalamus, lactate on MRS	Clinical, genetic, and lab testing (lipopigments from the skin)
AR ataxia of Charlevoix-Saguenay	CA (<superior vermis), T2 pons SC	Small CST and large pontocerebellar fibers at DTI	Clinical and genetic testing
Marinesco-Sjögren syndrome	CA (or hypoplasia)	Agenesis CC, small pituitary gland, cortex SC	Clinical and genetic testing
4H syndrome	CA and WM lesions	Cerebral hypomyelinization, thin CC	Clinical findings
Alexander disease[e]	SC brain stem with enhancement	Frontal subcortical WM and BG SC	Clinical, lab findings (glial fibrillary acidic protein 5), and genetic testing
HCC	SC cerebellar WM, pons hypoplastic	Diffuse WM hypomyelinization at the periventricular WM	Genetic testing
PPCARP	SC dorsal cord	None	Clinical, neuroimaging, and genetic testing
Wilson disease[f]	SC dorsal midbrain, CP and SCP	Signs of hepatic failure; BG, thalamus, and WM SC	Clinical, neuroimaging, and lab testing (copper/ceruloplasmin)

Note:—AOA indicates ataxia with oculomotor apraxia (types 1 and 2); ADL, adrenoleukodystrophy; CA, cerebellar atrophy; CC, corpus callosum; CP, cerebral peduncle; CST, corticospinal tract; DN, dentate nucleus; GP, globus pallidus; mIns, myo-inositol; HABC, hypomyelination with atrophy of the basal ganglia and cerebellum; GM2 = ganglioside 2; HCC, hypomyelination with congenital cataracts; SCP, superior cerebellar peduncle; PPCARP, posterior column ataxia and retinitis pigmentosa; SC, T2 high signal changes; BG, basal ganglia.

[a] Including X-linked sideroblastic anemia and ataxia.

[b] Including *coenzyme q10* and *POLG* mutations.

[c] Ataxia can occur in some diseases, including urea cycle disorders and mevalonic aciduria.

[d] Late-onset.

[e] Infantile-onset.

[f] Ataxic-onset in children.

[g] Reduced albumin, elevated cholesterol and α-fetoprotein in the plasma.

[h] Acanthocytosis in abetalipoproteinemia.

On-line Table 2: Malformative pattern

Disease	Posterior Fossa MRI Finding	Additional Imaging Findings	Confirmatory Test
Joubert syndrome and related diseases	Molar tooth malformation	Abnormal foliation and fissuration of CH, CC dysgenesis, migration disorders, ventriculomegaly	Clinical and neuroimaging
PCH[a]	PC hypoplasia, "dragon-fly" configuration (>PCH I or II)	Microcephaly, abnormal CC, polymicrogyria	Clinical, neuroimaging, and genetic testing
Rhomboencephalossinapsis	Agenesis of the CV, fusion of the CH and DN	CC dysgenesis, ventriculomegaly	Clinical and neuroimaging
Congenital disorders of glycosylation	PC hypoplasia with atrophy superimposed (>CV)	Brain stem hypoplasia, WM hypoplasia or atrophy	Clinical, neuroimaging, lab, and genetic testing
X-linked nonprogressive congenital ataxia[b]	PC hypoplasia with atrophy superimposed	Not described	Clinical, neuroimaging, and genetic testing
Vermian and hemispheric cerebellar dysplasia	Abnormal folia orientation and cortical dysplasia	Variable according to the genetic mutation (cerebral malformation, brain atrophy, and ventriculomegaly can occur)	Neuroimaging
VLDLR-associated cerebellar hypoplasia[c]	Vermis and CH hypoplasia (>inferior portion of the vermis)	Mild cortical gyral simplification	Clinical, neuroimaging, and genetic testing
Dandy-Walker malformation	Enlarged PF, agenesis vermis, and elevated torcula	Hydrocephalus, CC anomalies, PMG, heterotopia, cephalocele	Neuroimaging
Pontine tegmental cap dysplasia	Cap tegmental appearance	Dysgenesis of the corpus callosum, small inner auditory canal and/or VII/VIII nerves, abnormal tegmentum transverse fibers (DTI)	Neuroimaging
Cortex malformation disorders[d]	Cerebellar hypoplasia	Cerebellar cyst, brain stem hypoplasia, and WM lesions; cobblestone lissencephaly and hydrocephalus, usually with more severe types of CMD	Clinical, neuroimaging, and genetic testing
Brain stem malformation with abnormal segmentation pattern	Short pons, short midbrain/long pons or thick medulla	Variable and includes narrow isthmus, brain stem clefts, large CH, absent CC, ventriculomegaly	Clinical and neuroimaging.
Unertan syndrome	Cerebellar hypoplasia (>vermis)	Not described	Clinical and neuroimaging.
Höyeraal-Hreidarsson syndrome	Cerebellar hypoplasia	Small pons, thin CC, hypomyelinization, bone lesion	Clinical, neuroimaging, and genetic testing
Revesz syndrome	Cerebellar hypoplasia	Multiple cerebral calcifications, leukodystrophy	Clinical, neuroimaging, and genetic testing

Note:—PCH indicates pontocerebellar hypoplasia; PF, posterior fossa; CMD, congenital muscular dystrophy; CC, corpus callosum; CH, cerebellar hemispheres; CV, cerebellar vermis; DN, dentate nucleus; PC, pontocerebellar; PMG, polymicrogyria; VLDLR, very low density lipoprotein receptor.
[a] Types I-VII.
[b] Including isolated and X-linked subtypes.
[c] Dysequilibrium syndrome.
[d] Four most common CMD groups (mild phenotype).