Supporting Table 16. Dystonia with leukoencephalopathy

With basal ganglia abnormalities

INHERITED

Autosomal dominant

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)

Autosomal recessive

Phospholipase A2 associated neurodegeneration (PLAN)
Fatty acid hydroxylase associated neurodegeneration (FAHN)
Woodhouse-Sakati syndrome
Leigh's syndrome (complex 1 deficiency)
Hypomyelination with atrophy of the basal ganglia and cerebellum (H-ABC)
Sterol carrier protein X (SCPx) mutations
Glutaric aciduria
Cerebroretinal microangiopathy with calcifications and cysts
Fucosidosis
Aicardi-Goutieres syndrome

Mitochondrial

Leigh's syndrome (various mutations)

ACQUIRED

Infection

Progressive multifocal leukoencephalopathy

Immune-mediated

Acquired disseminated encephalomyelitis (ADEM)

Without basal ganglia abnormalities

INHERITED

Autosomal dominant

Alexander disease (GFAP mutations)

Autosomal recessive

Globoid leukodystrophy (Krabbe's disease)
Metachromatic leukodystrophy
Complex 1 deficiency
POL III-related leukodystrophies
Megalencephalic leukoencephalopathy with subcortical cysts (MLC)
Glutaric aciduria
Xeroderma pigmentosum

Mitochondrial

Complex 1 deficiency

X-linked

X-linked adrenoleukodystrophy
Pelizaeus-Merzbacher disease (PMD)
Allan-Herndon-Dudley syndrome (monocarboxylate transporter 8 mutations, MCT8)

ACQUIRED

Perinatal brain injury

Periventricular hemorrhage
Perinatal stroke

Infection

HIV infection

Toxic

Heroin inhalation (“chasing the dragon”)