Supporting Table 15. Dystonia with basal ganglia lesions (other than brain iron accumulation)

Acute/subacute onset

INHERITED

Autosomal recessive

Glutaric aciduria
Methylmalonic aciduria
Leigh's syndrome (eg PDH deficiency)
3-Oxothiolase deficiency
Biotin responsive encephalopathy (thiamine transporter deficiency)
Autosomal recessive infantile bilateral striatal necrosis (can also be gradual)
Aicardi-Goutiere syndrome

Mitochondrial

Leigh's syndrome (eg A2343G mutations)

ACQUIRED

Perinatal brain injury

Hypoxic-ischemic insult

Infection

Japanese B encephalitis
Mycoplasma
Herpes simplex (especially infants)
Human herpes virus 6
Diarrhea-associated hemolytic uremic syndrome
Measles
Cryptococcus
Toxoplasma
Prion disease

Immune-mediated

Acquired disseminated encephalomyelitis (ADEM) [CHECK]
NMDAR antibody associated encephalitis (especially children)
Sydenham's chorea (atypical)

Toxic

Wasp sting

Vascular

Stroke
Moyamoya disease

Metabolic

Hypocalcemia
Hypoparathyroidism
Uremia with diabetes

Gradual onset

INHERITED

Autosomal dominant

Huntington's disease
Autosomal dominant striatal degeneration
Neurofibromatosis type 1

Autosomal recessive

Wilson’s disease
Manganese transporter deficiency
MEGDEL (3-methylglutaconic aciduria with sensorineural deafness, encephalopathy, and Leigh-like syndrome) syndrome (SERAC1 mutations)
Cerebral creatine deficiency
GM1 gangliosidosis (adult variant)
GM2 gangliosidosis (incl. Tay-Sach’s disease)
Succinic semialdehyde dehydrogenase deficiency
Creatine transporter deficiency
Hypomyelination with atrophy of the basal ganglia and cerebellum (H-ABC)
Nephrin mutations (congenital nephrotic syndrome, Finnish type)
Sterol carrier protein X (SCPx) mutations (thalamus and brainstem)
Fucosidosis

Mitochondrial

Mitochondrial disease (eg Leigh syndrome, Leber’s hereditary optic neuropathy, polymerase gamma mutations)

ACQUIRED

Perinatal brain injury

Hypoxia-ischemia
Stroke

Infection

HIV
Prion disease

Immune-mediated

Multiple sclerosis
Sydenham’s chorea (atypical)
Juvenile rheumatoid arthritis

Toxic

Carbon monoxide (delayed onset)
Methanol (delayed onset)
Disulfiram (delayed onset)
Cyanide (delayed onset)
Manganese
- Manganese miners
- Welders
- Chronic liver disease (hepatolenticular degeneration)
- TPN
- Ephedrone recreational use

Vascular

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

Metabolic

Hypoxia (often delayed onset)

- Asphyxia
- Perinatal hypoxia-ischemia

Hyperglycemia (hyperosmolar non-ketotic acidosis, HONK)

Hypoglycemia

Extrapontine myelinosis

Hypocalcemia

Hypoparathyroidism

Hepatolenticular degeneration

Neoplastic

Glioma

Lymphoma

Germ cell tumor

Metastatic

IDIOPATHIC

Sporadic

Multiple system atrophy