Supporting Table 1. Dystonia with or without parkinsonism of infantile or childhood onset

INHERITED

Hereditary disorders of dopamine metabolism

Autosomal dominant

GTP-cyclohydrolase 1 (Segawa disease)

Autosomal recessive

Dopamine metabolic pathway

- Phenylketonuria (in adulthood)
- GTP-cyclohydrolase 1 homozygous mutations
- Tyrosine hydroxylase
- L-amino acid decarboxylase deficiency

Tetrahydrobiopterin synthesis pathway

- Homozygous (autosomal recessive) GTP-cyclohydrolase 1 deficiency
- 6-Pyruvoyl-tetrahydropterin synthase (PTPS)
- Sepiapterin reductase (SR)

Tetrahydrobiopterin regeneration pathway

- Pterin-4α-carbinolamine dehydratase (PCD)
- Dihydropteridine reductase (DHPR)

Other

Dopamine transporter deficiency

Other hereditary disorders causing mainly dystonia

Autosomal dominant

Huntington's disease

Autosomal recessive

Wilson's disease
Pantothenate kinase-associated neurodegeneration (PKAN)
Phospholipase A2 associated neurodegeneration (PLAN)
DYT16 (PRKRA mutations)
Manganese transporter deficiency
GM1 gangliosidosis
GM2 gangliosidosis (incl. Tay-Sach's disease)

X-linked

Rett syndrome

ACQUIRED

Infection

Japanese B encephalitis
Mycoplasma
Measles
Encephalitis lethargica (some cases)

Immune-mediated

Encephalitis lethargica (some cases)

Metabolic

Hypoxia (often delayed onset)

- Asphyxia
- Perinatal hypoxia-ischemia

Extrapontine myelinosis