**Supporting Table 18.** Progressive dystonia with normal brain MRI or generalised atrophy

***Without cognitive impairment***

**Autosomal dominant**

Rapid onset dystonia-parkinsonism (ATP1A3 mutations)
AD dopa responsive dystonia (Segawa disease) due to GTP cyclohydrolase I mutations
DYT1 (torsin 1a mutations)
DYT6 (THAP mutations)

**Autosomal recessive**

Parkin (PARK2)
PINK1 mutations (PARK6)
DJ-1 mutations (PARK7)
FBXO7 mutations

**X-linked**

Lubag (DYT3) (TAF mutations)

***With static cognitive impairment (mental retardation)***

**Autosomal recessive**

Homocystinuria

**X-linked**

Lesch-Nyhan disease

***With progressive cognitive decline (dementia)***

**INHERITED**

**Autosomal recessive**

Neuronal ceriod lipofuscinoses
Niemann-Pick Type C
Gaucher's disease
Kufor-Rakeb disease (PARK9 due to ATP13A2 mutations) (some have NBIA)
Phospholipase A2 associated neurodegeneration (PLAN) (most have NBIA or leukoencephalopathy)

**X-linked**

Mohr-Tranebjaerg syndrome (DDP1 gene mutations)

**IDIOPATHIC**

**Sporadic**

Progressive supranuclear palsy—may have focal atrophy
Corticobasal degeneration—may have focal atrophy

***Normal T1- and T2-weighted images with abnormalities on special sequences***

**INHERITED**

**Autosomal recessive**

Some NBIAs (eg PLAN)—need iron susceptibility sequences
Cerebral creatine deficiency syndromes—need MR spectroscopy

**ACQUIRED**

**Infection**

Prion disease—may need diffusion weighted images