DYSTONIA WITH OPHTHALMOLOGICAL ABNORMALITIES

Supranuclear gaze palsy
- *Vertical gaze*
  - Kufor-Rakeb syndrome (often upward gaze first)
  - Niemann-Pick Type C (often downward gaze first)
  - Progressive supranucear palsy (often saccadic slowing before restriction)
  - Tau-gene associated frontotemporal dementia and parkinsonism
  - SPG7 (paraplegin)
  - SPG11
  - Phospholipase A2 associated neurodegeneration (PLAN) (PLA2G6 mutations)
  - Pantothenate kinase associated neurodegeneration (PKAN) (pantothenate kinase mutations)
  - FBXO7 mutations
  - Phosphoglycerate kinase deficiency
  - Glutaric aciduria
  - Prion disease
  - Kernicterus
- *Multidirectional*
  - Huntington's disease
  - Spinocerebellar ataxias (especially SCA2 and SCA3)
  - Neuroacanthocytosis
  - AFG3L2 homozygous mutations
- *Oculomotor apraxia*
- Coriticobasal degeneration
- Ataxia-telangiectasia
- Ataxia-oculomotor apraxia type 1
- Ataxia-oculomotor apraxia type 2
- Ataxia-telangiectasia like syndrome
- GM2 gangliosidosis

Progressive external ophthalmoplegia
- Mitochondrial disease

Retinal abnormalities
- *Cherry red spot*
  - GM1 gangliosidosis
  - GM2 gangliosidosis
- *Pigmentary retinopathy*
  - Mitochondrial disease
  - Pantothenate kinase associated neurological disease
  - Aceruloplasminemia
  - Metachromatic leukodystrophy
  - SCA7
- *Optic atrophy*
  - Mitochondrial protein associated neurological disease (MPAN)
  - Mohr-Tranebjaerg syndrome (DDP1 gene mutations)
  - SPG7 (paraplegin)
  - Metachromatic leukodystrophy

Methylmalonic aciduria
CASK mutations
Retinal telangiectasia/angiomas
Cerebroretinal microangiopathy with calcifications and cysts

Cataracts

Wilson's disease
Homocystinuria
Cerebrotendinous xanthomatosis
Mitochondrial disease
FBX07 mutations

Corneal abnormalities

Wilson's disease (Kayser-Fleischer rings)
GM1 gangliosidosis (corneal clouding)
Xeroderma pigmentosum (corneal opacification, neoplasms)