

Supporting Table 12. Dystonia with hematological abnormalities

Acanthocytes

- Chorea-acanthocytosis (chorein mutations)
- Pantothenate kinase associated neurodegeneration (PKAN)
- McLeod syndrome
- Huntington's disease-like 2 (junctophilin mutations)

Anemia

- Diarrhea-associated hemolytic uremic syndrome
- Wilson's disease
- Aceruloplasminemia
- Lesch-Nyhan syndrome
- Mitochondrial disease (e.g. Leigh's syndrome)
- Multiple system atrophy
- Neuroferritinopathy (low ferritin rather than anemia)
- Glut1 mutations
- triosephosphate isomerase deficiency
- 3-Oxothiolase deficiency
- Phosphoglycerate kinase deficiency
- Gaucher disease

Polycythemia

- Manganese transporter deficiency
- Fumarate hydratase deficiency

Leukopenia

- Gaucher disease
- Fumarate hydratase deficiency

Thrombocytopenia

- Wilson's disease
- Gaucher disease
- Methylmalonic aciduria
- Aceruloplasminemia
- Manganese transporter deficiency
- Aicardi-Goutieres syndrome
- 3-Oxothiolase deficiency

Agammaglobulinemia

- X-linked agammaglobulinemia (BTK mutations)
- Mohr-Tranebjaerg syndrome due to DDP1 deletion with contiguous BTK deletion

Other

- Methemoglobinemia type II