GENETIC DISORDERS THAT MASQUERADE A CEREBRAL PALSY

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AACPDM 2011

- **BRAIN MALFORMATIONS**

  - X-Linked hydrocephalus/aqueductal stenosis
  - Posterior fossa abnormalities – including Dandy Walker malformation
  - Holoprosencephaly spectrum
  - Neuronal migration disorders – Lissencephaly, heterotopias, polymicrogyria
  - Schizencephaly
  - Septo-Optic dysplasia

- **DISEASES WITH ABNORMAL BASAL GANGLIA**

  - Mitochondrial disorders
  - Glutaric aciduria type I
  - Neurodegeneration with brain iron accumulation (NBIA) – including PKAN and INAD.
  - Succinyl-Semialdehyde dehydrogenase deficiency
  - Lesch-Nyhan syndrome
  - Thiamine Transporter Deficiency
  - Biotinidase deficiency
  - Methylmalonic academia
  - Wilson disease
  - Huntington Disease
  - Leukodystrophies
  - Pseudo-TORCHes

WHITE MATTER DISEASES

- **Hypomyelination**

  - Pelizaeus Merzbacher Disease (PMD)
  - PMD-like disease – GJA12 gene
  - Monocarboxylate Transporter 8 deficiency (Allan-Hernon-Dudley syndrome)
  - Hypomyelination and Congenital Cataract
  - 4H Syndrome (Hypomyelination, hypodontia, hypogonadotropic gonadism).
  - Sialic Acid Storage Disease
  - Cree Leukoencephalopathy (EIF2B-related vanishing white matter syndrome)

- **Demyelination**

  - Krabbe leukodystrophy (globoid cell leukodystrophy)
  - Metachromatic leukodystrophy
  - Peroxisomal Biogenesis disorders/Zellweger Spectrum

- **Diseases mimicking cystic PVL** (destructive white matter diseases)
    - Sulfite Oxidase Deficiency/Molybdenum Cofactor Deficiency
    - Mitochondrial disorders
    - Pyruvate dehydrogenase deficiency
    - Fumarase deficiency

- **Delayed myelination**
    - Neuronal ceroid lipofuscinoses (NCL)/Batten disease
    - Sanfilippo syndrome (mucopolysaccharidosis type 3)
    - Mucosidosis
    - Fucosidosis
    - Tay-Sachs Disease
    - Mitochondrial disorders
    - Biotinidase Deficiency

- **PSEUDO-TORCH**
    - Aicardi-Goutieres syndrome
    - RNASET2-Deficient CMV-like leukoencephalopathy
    - LaBrune disease – Cerebroretinal microangiopathy with calcifications and cysts (CRMCC)
    - OCLN-related Band-Like Calcification with Polymicrogyria (BLCPMG).

- **SPASTIC PARAPLEGIAS**
    - Hereditary spastic paraplegias
    - Arginase deficiency

- **ATAXIAS**
    - Spinocerebellar Ataxias
    - Ataxia Telangiectesia
    - Angelman syndrome
    - Joubert syndrome
    - Congenital Disorders of glycosylation
    - Mitochondrial diseases
    - Episodic ataxias

- **DISORDERS OF CNS NEUROTRANSMITTERS**
    - Disorders of monoamine synthesis
        - GCH1 deficiency (Dopa-responsive dystonia/Segawa disease)
        - Tyrosine hydroxylase (TH) deficiency
        - Aromatic Acid Decarboxylase (AADC) deficiency
        - Others: DHPR, PCD, PTPS, SR deficiencies
    - Cerebral folate deficiency.