Cobalamin C Disease

Methylmalonic acuduria with homocystinuria

Main Features

- Most common inborn error of vitamin B12 metabolism
- Early Onset (First year of life)
    - Neurologic & Cognitive Deterioration
    - Pancytopenia
    - Megaloblastic anemia
    - Progressive Retinopathy
- Later Childhood (>6 years) Onset
    - Gait abnormalities
    - Extrapyramidal symptoms
    - Psychiatric disturbances
    - Dementia
    - Cognitive Disability
    - No retinopathy

Eye Findings

- Retinopathy
    - Spectrum from mild nerve fiber layer loss to advanced macular and optic nerve atrophy with “bone spicule” pigmentation
    - Reduced ERG responses
- Nystagmus

Other Findings

- Failure to thrive, poor feeding, vomiting
- Microcephaly, brain malformations
- Hypotonia
- Speech delay
- Seizures

Etiology

- Mutation in MMACHC gene
    - Gene product converts dietary vitamin B12 to the metabolically active forms
- Treatment includes supplementation with:

- hydroxycobalamin
- betaine
- folinic acid

Reference

- Ocular Phenotype in Patients with methylmalonic aciduria and homocystinuria, cobalamin C type. J AAPOS 2008
- Methylmalonic aciduria and homocystinuria-associated maculopathy. Eye 2010
- The Adolescent and adult form of cobalamin C disease: clinical and molecular spectrum. J Neurol Neurosurg Psychiatry 2008

syndrome