Optic Atrophy in Children

Causes

- Eye Malformation/Injury/Infection
  - Aniridia
  - Accidental or Non-Accidental trauma
  - Neuroretinitis
- Retinal Dystrophy
  - CSNB
  - Leber congenital amaurosis
  - Stargart disease
  - X-linked retinoschisis
  - Incontientia pigmenti
- Brain malformation/injury/infection/degeneration
  - Optic neuritis
    - Neuromyelitis optica
  - Brain Tumor
    - especially suprasellar tumors
  - Intraventricular hemorrhage
  - Dandy Walker
  - Congenital Hydrocephalus
  - Perinatal Insult
    - Periventricular leukomalacia
  - Meningitis
  - Batten Disease
  - Zellweger spectrum (peroxisomal disorder)
  - Charcot-Marie-Tootl
  - Brown-Vailetto-Van Laere syndrome
  - Sandhoff Disease
  - CNS Syphilis
  - Lyme disease
- Inheritable Optic Atrophy
  - Dominant Optic Atrophy
    - OPA 1
  - Leber Hereditary Optic Neuropathy
    - Mitrochondrial DNA mutation- mothers pass to children
  - DIDMOA (Diabetes Insipidus/Diabetes mellitus/ Optic Atrophy)
    - a.k.a Wolfram Syndrome
- Skeletal Malformation
- Toxic/Metabolic
  - Anemia
  - Lead poisoning (or other heavy metals; arsenic, mercury)
  - B12 deficiency

    - Vitamin D Deficiency (Rickets)

Suggested Initial Evaluation

- Ask about History of Prematurity, Trauma, Neurological decline or seizure, birth history, Post natal growth and development
- Family history of low vision and heritable eye disease
- Exam:
    - Visual acuity
    - Pupil reactivity
    - Color vision
    - Refraction
    - Anterior Segment and Posterior segment exam
    - Visual fields
    - OCT nerves/macula
    - Fundus Photos
- MRI orbits and brain
- Labs:
    - CBC (anemia)
    - Heavy metal screen in blood (toxic exposure of lead and others)
    - B12 (GI malabsorption / nutritional deficiency)
    - Lactate levels in whole blood (neurogenerytative diseases)
    - Syphilis Total Serum Antibodies
    - consider Lyme serum antibody titers for acute or late infection
    - consider hereditary optic neuropathy genetic panel

Subsequent Evaluation

- OCT nerves and macula
- ERG
- VEP
- Molecular Genetic testing
    - Hereditary Optic Neuropahty
    - Dominant Optic Atrophy (OPA1)
- Lumbar Puncture
    - CNS infection
    - Hydrocephalus

References

1. Optic Atrophy in Children: Current Causes and diagnostic approach. Jones R et al. EJO. 2020;30(6):1499-1505

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