Opitz Syndrome

AKA. Oculi-genito-laryngeal syndrome, BBB/G compound syndrome

Main Features

- Defects in midline structures
- Telecanthus (wide spaced eyes)
- Hypospadius
- Cleft lip or palate (25%)

Eye Findings

- Telecanthus / Hypertelorism
- Strabimus (exotropia more likely)
- Anisometropia

Other Findings

- Prominent forehead
- Small Jaw
- Ear abnormalities
- Laryngotracheaoesophageal malformations
- Congenital heart defects
- Brain Malformations

Etiology

- Chromosome 22q11.2 deletion
- MID1 gene defect
    - X-linked

Reference

- OMIM reference
- Gene Reviews

syndrome