DiGeorge Syndrome

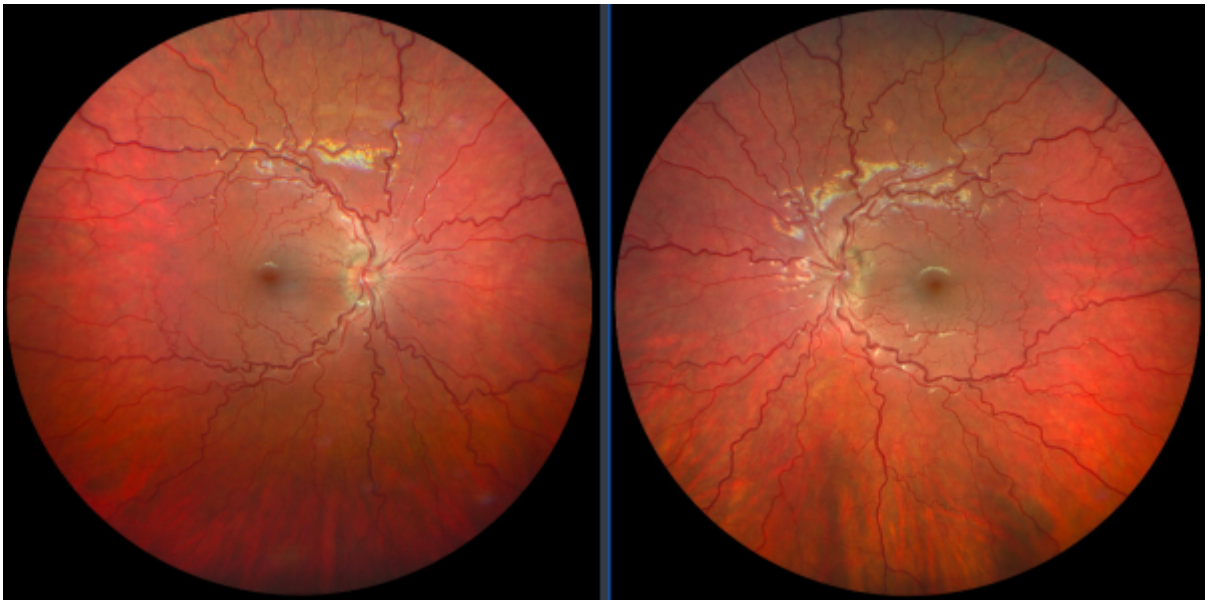
Main Features

- Various phenotypic expressions with multiorgan dysgenesis
- Most common abnormalities involve the following systems/organs:
    - cardiac
    - craniofacial
    - thymus
    - parathyroid

Eye Findings

- Anterior segment
    - Posterior embryotoxon (15%)
    - Sclerocornea
    - Peters anomaly
    - Iris remnants
    - Cataracts
    - Uveitis
    - Colobomas
    - Anterior segment dysgenesis
- Posterior segment
    - Tortuous retinal vessels (24%)
    - Optic Disc abnormalities (hypoplastic, hyperpigmented, small or tilted, excavated) (9%)
    - 22q11.2 may be an additional genetic locus for familial exudative vitreoretinopathy
- Globe
    - Microphthalmia with or without orbital cyst- rare
    - Persistent fetal vasculature- rare
- External, Orbit, Vision
    - Strabismus (10%) / Amblyopia (6%)
    - Eyelid abnormalities
        - Epicanthus (4%)
        - Ptosis (3%)
        - Distichiasis
        - Dacryostenosis (3%)

Fundus Vascular Tortuosity and Optic Nerve anomaly in teenager with DiGeorge



Other Findings

- Renal, endocrine, immunologic, and psychiatric problems

Etiology

- Partial deletion of the long arm of chromosome 22 (deletion 22q11.2)
- 22q11.2 distal deletion syndrome is considered distinct from DiGeorge and is described separately

Reference

- Ocular Findings in 22q11.2 deletion syndome: A systematic literature reveiw and results of a Dutch multicenter study
- Microphthalmia and orbital cysts in DiGeorge syndrome- JAAPOS
- Medline Plus
- Gene Reviews
- 22q11.2 Deletion Syndrome. McDonald-McGinn DM et al. Nat Rev Dis Primers 2016;1:15071
- Chromosome 22q11.2 deletion syndrome and DiGeorge syndrome. Sullivan KE. Immunol Rev. 2019;287(1):186-201
- 22q11.2 distal deletion: A Recurrent Genomic Disorder Distinct from DiGeorge Syndrome and Velocardiofacial Syndrome. Ben-Shachar, S et al. American Journal of Human Genetics. 2008;82:214-221

syndrome