Turner Syndrome and Mosaic Turner Syndrome

Main Features

- Usually phenotypically female
- Short stature
- Webbed neck
- “shield chest” with wide-spaced nipples
- cubitus valgus
- hearing loss
- Cardiac malformations
    - Bicuspid Aortic valve
    - Coarctation of the aorta
    - Partial anomalous pulmonary venus return
    - Aortic dilation and dissection

Eye Findings

- Refractive error
    - Hyperopia more common
- Strabismus
    - Esotropia more than exotropia
- Amblyopia
- Rare problems (few case reports)
    - Early onset cataracts
    - Keratoconus
    - Limbal stem cell deficency
    - Anterior lenticonus
    - Retinal detachment
- Mosaic Turner Findings
    - Anterior segment dysgenesis
    - microphthalmia
    - Sclerocornea
    - Congenital Glaucoma
    - Reiger Malformation

Other Findings

- 1:2000

Etiology

- Monosomy X

Resources

- Turner Syndrome: Ocular Manifestations and Considerations for Corneal Refractive Surgery. Moshifar Majid et al. J Clin Med. 2022;11:6853
- Anterior Segment Dysgenesis in Mosaic Turner Syndrome. Lloyd IC, Haigh PM, Clayton-Smith J, et al. The British Journal of Ophthalmology. 1997;81(8):639-43. doi:10.1136/bjo.81.8.639. https://pubmed.ncbi.nlm.nih.gov/9349149/
- Microphthalmia With Linear Skin Defects Syndrome in a Mosaic Female Infant With Monosomy for the Xp22 Region: Molecular Analysis of the Xp22 Breakpoint and the X-Inactivation Pattern. Ogata T, Wakui K, Muroya K, et al. Human Genetics. 1998;103(1):51-6. doi:10.1007/s004390050782. https://pubmed.ncbi.nlm.nih.gov/9737776/

syndrome

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