Points to remember for Clinical Practice

Anterior Segment Eye Diseases

- Brushfield spots and peripheral iris hypoplasia are typical phenotypes of individuals with Trisomy 21 and have no consequences for vision.
- Structural abnormalities of the cornea are common in 75% of individuals with Trisomy 21 at the age of 15 years, with corneal indices similar to those indicating mild keratoconus in the general population.
- A higher prevalence (6.8 to 30 times higher) of keratoconus (KC) in individuals with Trisomy 21 compared to the general population.
- Given the higher frequency and faster progression of KC towards severe forms in individuals with T21, it is important to increase ophthalmological monitoring and use corneal topo/tomography techniques repeatedly to enable early diagnosis and closely monitor the progression.
- The presence of fluffy or punctate lens opacities is very common in young adults with Trisomy 21, representing over 50% of the lens opacities reported, but they have no consequences for vision.
- The prevalence of congenital cataracts in children with Trisomy 21 is 1.4-1.7%, which is 80-100 times more frequent than in the general population. They are mostly bilateral, and one-third of cases are diagnosed at birth, justifying neonatal screening using the red reflex test.

Ophthalmic Disorders

- Age-related cataracts in individuals with Trisomy 21 are more frequent and occur earlier than in the general population, with a rapid increase in prevalence starting at the age of 40.

- Examining the cornea and measuring intraocular pressure are essential for diagnosing infantile or secondary glaucoma in children, especially in cases of unexplained tearing or in the medium- to long-term follow-up after congenital cataract surgery. This is also important in adults with Trisomy 21, as they may have a higher risk of glaucoma.