

POINTS TO REMEMBER FOR CLINICAL PRACTICE

Neuro-ophthalmology

- There is a high prevalence of strabismus, nystagmus, and amblyopia in individuals with Trisomy 21 (T21).
- The association between hyperopia, accommodative weakness, and esotropia is demonstrated in individuals with T21.
- In clinical practice, it is important to rule out pseudoesotropia; strabismus is the leading cause of abnormal head posture.
- Strabismus in individuals with T21 is usually esotropia, acquired, and with a later onset (around 4.5 years).
- Treating strabismus is a functional priority because untreated strabismus can lead to amblyopia. It is important to fully correct optical issues, including low-grade hyperopia and accommodation disorders (bifocal glasses).
- The success rates of esotropia surgery in children with T21 are similar to those in the general population.
- Nystagmus in individuals with T21 is often infantile nystagmus syndrome, which starts between 4 and 12 weeks of age and is associated with esotropia or myopia.
- The presence of infantile nystagmus in children with T21 clearly worsens the baseline visual deficit in these individuals.
- Amblyopia is a common sensory pathology in children and young adults with T21.

- Amblyopia in individuals with T21 is often of functional origin and can be either strabismic or high-grade refractive.
- The management of amblyopia is primarily preventive through early screening, as crucial intervention should occur before the age of 6, namely full correction of refractive anomalies and strabismus.
- Trisomy 21 accounts for approximately 50% of Cerebral Visual Impairment cases of genetic origin.

