Points to remember for Clinical Practice

Posterior Segment Eye Diseases

- It is important to perform fundoscopy and optical coherence tomography (OCT) to examine the posterior segment in individuals with Trisomy 21.

- Although many fundus abnormalities have been described in individuals with Trisomy 21, no association has been demonstrated between these anomalies and visual impairment in individuals with Trisomy 21.

- The retina of both children and adults with Trisomy 21 presents abnormal development in OCT scans, both in the macula and fovea, and in the choroid or retinal nerve fibers.

- No formal association has been demonstrated between the retinal anomalies found in OCT scans in older individuals with Trisomy 21 and the development of Alzheimer's disease.

- Retinal detachment should be considered in individuals with Trisomy 21, particularly late-onset RD after congenital cataract surgery.

- While retinoblastoma is rare (1/15,000 births in the general population), it may be more frequent in children with Trisomy 21, which warrants systematic screening.

- Although optic nerve anomalies are frequent in individuals with Trisomy 21, they do not appear to significantly contribute to the decrease in visual acuity commonly found in individuals with Trisomy 21.

- Brain imaging and lumbar puncture should be performed on children with Trisomy 21 presenting with papilledema or pseudopapilledema, even if asymptomatic, to confirm the diagnosis.