Points to remember for Clinical Practice

Pathologies of the Ocular Adnexa

- Congenital palpebral anomalies are the ocular abnormalities most frequently associated with Trisomy 21. In most cases, they have no impact on visual development.

- A so-called "Mongolian" slant oriented upwards and outwards from the palpebral fissure is the most commonly observed facial dysmorphia in individuals with T21.

- In individuals with T21, the epicanthal fold is epicanthus palpebralis and has no impact other than the possibility of falsely diagnosing strabismus, due to pseudoesotropia.

- The high frequency of epiblepharon, in which the eyelid margin itself is not turned inward, unlike entropion, and which does not tend to spontaneously regress in individuals with T21.

- Congenital entropion is rare but not exceptional in individuals with T21; it tends to worsen over time and can result in chronic corneal damage.

- Congenital ectropion and euryblepharon are rare anomalies that can be associated with Trisomy 21.

- Congenital eversion of the upper eyelids is a rare neonatal disorder more often observed in certain pathologies, including Trisomy 21. Although spectacular, it usually regresses spontaneously in the first month of life.

Ophthalmic Disorders

- Blepharitis and blepharo-conjunctivitis are the most commonly reported infectious/inflammatory eyelid pathologies in individuals with T21. They should not be neglected due to the possibility of chronic corneal and conjunctival damage and progression to keratoconus due to eye rubbing.

- Congenital nasolacrimal duct obstruction is common in individuals with T21, particularly in the first year of life. Compared to the general population, they are:
  - More frequently bilateral;
  - More often complex;
  - Less likely to spontaneously resolve;
  - Less effectively treated by initial nasolacrimal duct probing.

- Congenital lacrimal fistulas are rare pathologies but sometimes associated with T21.