Axenfeld anomaly - Posterior Embryotoxon

Axenfeld anomaly, a component of Axenfeld-Rieger syndrome, is a rare congenital ocular defect characterized by anterior segment dysgenesis. This condition is marked by an anteriorly displaced Schwalbe's line, known as posterior embryotoxon, and iris bands extending into the cornea. It is distinct from Rieger's anomaly, which includes additional iris and pupil anomalies. This syndrome is often associated with glaucoma and can present congenital malformations of the face, teeth, and skeletal system. The genetic basis of this condition is complex, involving mutations in genes like PITX2, FOXC1, and PAX6

Posterior embryotoxon is an ocular feature characterized by a prominent, anteriorly displaced Schwalbe's line, which is the anatomical border between the cornea and the trabecular meshwork in the eye. It appears as a visible ring on the posterior corneal surface. This condition is relatively common and can occur in the general population (10-15%) without associated eye abnormalities. However, it is also frequently observed in various anterior segment dysgenesis syndromes, including Axenfeld-Rieger syndrome. While posterior embryotoxon itself usually does not impair vision, its presence can be a marker for other ocular or systemic abnormalities.

On its own it does not cause an increase risk of chronic open angle glaucoma.

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