

Photo Essay

Axenfeld-Rieger Syndrome: Photo Essay

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Abstract

We report the case of a patient aged 47 who consult for a decrease in visual acuity. Examinations of the anterior segment spotting unilateral irrido-trabecular dysgenesis of the right eye with abnormal visibility of the Schwabe line corresponding to a posterior Embryotoxon and associated angular abnormalities. The eye tone measured with Goldman tonometer showed 14mmHg in the right eye and 15mmHg in the left eye. The examination of the fundus of the eye finds symmetrical morphology of the optic discs without pathological papillary excavation.

Keywords: Axenfeld-Rieger syndrome; Irrido-trabecularly dysgenesis; Embryotoxon

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We report the case of a patient aged 47 who consult for a decrease in visual acuity. Examination of the anterior segment spotting unilateral irrido-trabecular dysgenesis of the right eye with abnormal visibility of the Schwabe line corresponding to a posterior embryotoxon and associated angular abnormalities (Figures 1,2&3). The eye tone measured with Goldman tonometer showed 14mmHg in the right eye

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and 15mmHg in the left eye. The examination of the fundus of the eye finds symmetrical morphology of the optic discs without pathological papillary excavation.

The remainder of the somatic examination reveals no abnormality associated especially the absence of dental malformation. Chronic glaucoma is seen in 50% of patients [1].

The diagnosis of Axenfeld-Reiger syndrome uncomplicated of chronic glaucoma has been established, despite the absence of signs of Rieger [1].

No treatment has been established. Regular checks have been proposed to detect any complications including glaucoma.

This syndrome is inherited as an autosomal dominant manner. 2 genes are mainly involved in the transmission: the PITX2 gene in 4q25, present in 10-60% of patients, mainly associated with systemic alterations such as dental malformations [2,3]. The other gene responsible is FOXC1 located in 6q25, present in 50% of cases and manifested by ocular alterations, especially glaucoma [2-4].

The differential diagnosis arises with the Peters anomaly which consists of a defect of the posterior surface of the cornea associated with a stromal opacity. Currently, it is suggested that all these abnormalities are actually part of the same syndrome: Axenfeld-Rieger syndrome [5].

Conflict of interest

The authors declare no conflict of interest.

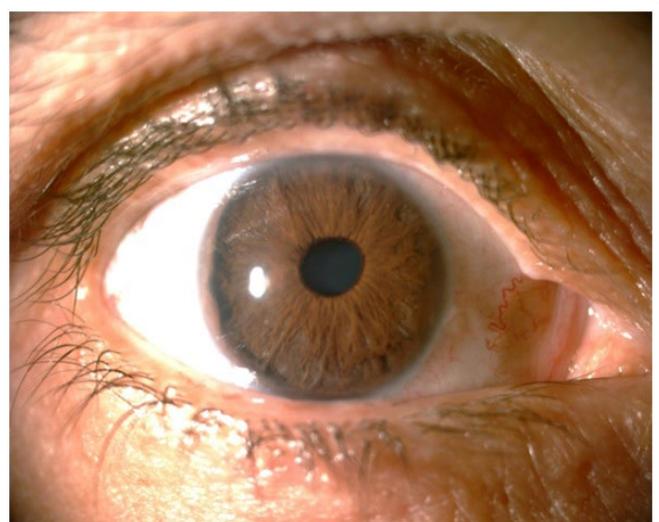


Figure 1: Embryotoxon on the temporal side of the right eye.

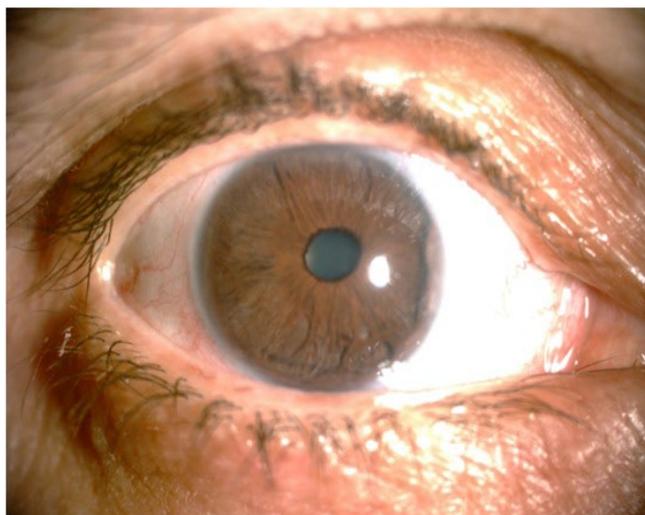


Figure 2: Embryotoxon on the nasal side of the right eye.

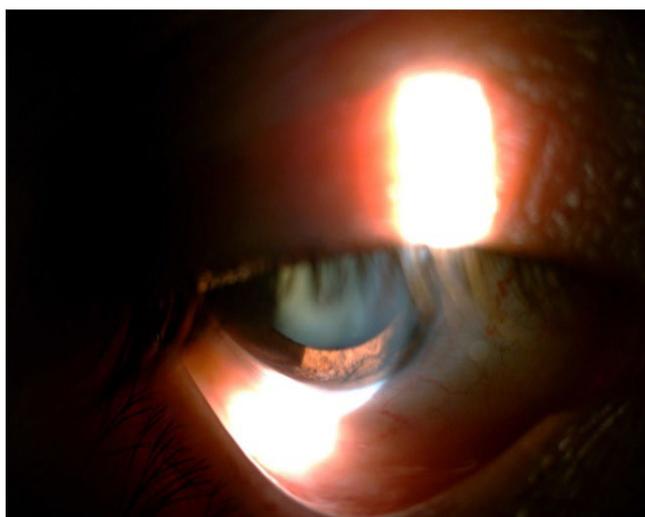


Figure 3: Angular abnormalities of the right eye.

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