Síndrome de Axenfeld-Rieder associada à maculopatia tipo "Bull's eye"

Axenfeld-Rieger syndrome associated with "Bull's eye" maculopathy

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RESUMO

A Síndrome de Axenfeld-Rieger representa um espectro de anomalias de desenvolvimento caracterizadas por alterações da periferia da córnea, íris e seio camerular, além de malformações ósseas e dentárias. Existem algumas descrições esparsas de anormalidades de fundo de olho, porém estas não foram documentadas. Nós descrevemos, pela primeira vez, um caso de Síndrome de Axenfeld-Rieger associada à maculopatia bilateral do tipo "bull's eye".

Palavras chave: Síndrome de Axenfeld-Rieder, "bull's eye", maculopatia, glaucoma.

INTRODUCTION

The Axenfeld-Rieger syndrome mesodermal dysgenesis represents a spectrum of developmental disorders characterized by abnormalities of the peripheral cornea, iris and anterior chamber angle associated with developmental defects of the teeth and facial bones.

The typical abnormality of the peripheral cornea is a prominent, anteriorly displaced Schwalbe's line. Gonioscopy frequently shows tissue strands extending from the peripheral iris to Schwalbe's line, as well as a high insertion of the peripheral iris. The iris may be normal, or may exhibit defects which range from mild stromal thinning to marked atrophy with hole formation, corectopia and ectropion uveae [1-3].

The ocular involvement is typically bilateral and glaucoma develops in approximately 50% of the cases, secondary to maldevelopment of the trabecular meshwork and the Schlemm's canal [1,2]. A family history of mesodermal dysgenesis is frequent, which most often follows an autosomal dominant pattern of transmission .

Aside from the ocular findings already mentioned, no additional ocular anomaly is frequent enough to be included as a typical feature of the syndrome. Sparse descriptions of fundus abnormalities have been made [3-9], but none have been well documented.

We report, to our knowledge for the first time, a case of Axenfeld-Rieger syndrome associated with bilateral "bull's eye" maculopathy.

CASE REPORT

A 14-year-old caucasian female was referred to the Glaucoma Service of Wills Eye Hospital for a second opinion. The referring ophthalmologist had measured intraocular pressures of 50 mmHg in both eyes after a routine examination and had prescribed timolol 0.5% and dipivalil-epinephrine 0.1% twice daily OU. A family history was not available, since the patient had been adopted.

On initial examination (October, 1987), the best corrected visual acuity

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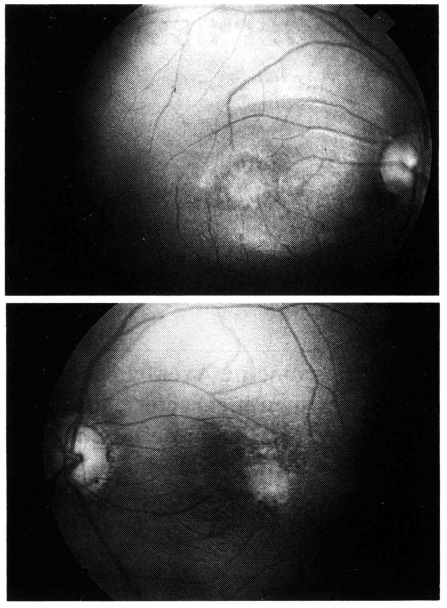
was 20/40 in the right eye and 20/50 in the left eye. Intraocular pressures were 32 mmHg bilaterally under anti-glaucoma medication before dilatation, and raised to 37 mmHg in the right eye and 38 mmHg in the left eye after dilatation. There was an afferent pupillary defect on the left side.

Corneal diameters were 11.5 mm in both eyes. There were hypoplastic changes in the iris of both eyes, most notably superiorly in the right eye (Figure 1). Remnants of pupillary membrane and ectropion uveae were seen in both eyes. Gonioscopy showed distinctly abnormal angle structures, with a very high anterior insertion of the iris to the level of the trabecular meshwork (inferiorly) and all the way to a prominent Schwalbe's line (superiorly). The anterior segment findings suggested the diagnosis of Axenfeld-Rieger syndrome.

The optic discs were markedly excavated, more significantly in the left eye. Fundus examination also showed a "bull's eye" pattern of retinal pigment epithelial alteration in the central macula of both eyes (Figures 2,3). There was a subtle "salt and pepper" pattern to the retinal pigment epithelium diffusely. The retinal vessels and periphery were unremarkable.

Humphrey computerized visual field testing showed central islands in both eyes, with a more advanced stage of loss in the left eye. Fluorescein angiography was of suboptimal quality, but revealed areas of hyperfluorescence in the central macula of both eyes. The electroretinogram was normal to photopic and scotopic stimulation, and the electro-oculogram showed normal light to dark ratios in both eyes. The Farnsworth Dichotomous Test for color blindness was within normal limits for both eyes.



Figuras 2 e 3- Fotografias do fundo de olho de ambos os olhos revelando um padrão "bull's eye" de maculopatia.

The patient underwent trabeculectomies in both eyes in October 1987. On the most recent examination (March, 1992), visual acuities were 20/30 in the right eye and 20/70 in the left eye. Intraocular pressures were 14 mmHg in the right eye and 13 mmHg in the left eye, without antiglaucoma medications. The optic nerves and the visual fields showed no progression, nor did the maculae.

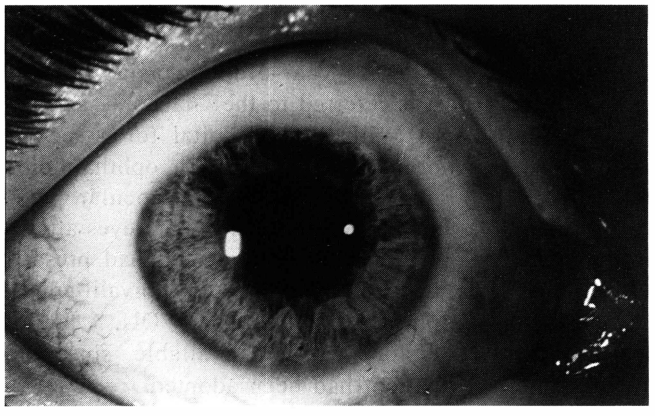


Figura 1- Fotografia do olho direito, demonstrando área de hipoplasia do mesoderma iriano superiormente e ao redor da pupila.

DISCUSSION

There are few reports about fundus abnormalities in patients with Axenfeld-Rieger syndrome. Forsius and Eriksson [5] reported a unilateral cystic macular degeneration in a 40-year-old man. Falls [4] described three patients where the macular area was "mottled with pigment clumps". Unfortunately, these macular changes were not documented, and no angiographic study was performed. Medullated nerve fibers [1,3,6], chorioretinal colobomas [7,8], choroidal hypoplasia [6,9] and retinal detachments [1] have also been described in patients with the syndrome.

This is, to our knowledge, the first report of a "bull's eye" pattern associated with the Axenfeld-Rieger syndrome. The differential diagnosis of this macular change includes chloroquine retinopathy, thioridazine retinopathy, cone dystrophy, rod-cone dystrophy, Stargardt's disease, lipofuscinosis, fenestrated sheen macular dystrophy, benign concentric annular macular dystrophy and autosomal dominant central areolar choroidal and pigment epithelial dystrophy unassociated with drusen or flecks.

Unfortunately, the genetic pattern of transmission could not be analyzed and the relatives could not be examined in our case report, since the patient was adopted. There was no history of drug ingestion, excluding the possibility of chloroquine or thioridazine retinopathies. The electrophysiologic and color vision tests were within normal limits, which goes against cone and rod-cone dystrophies (associated with impairment of color vision or abnormal photopic/ scotopic response in the electroretinogram).

The juvenile type of lipofuscinosis (Spielmeyer-Sjoegren) typically presents during the first decade (6-7 years of age). Mild changes in the RPE are observed during fundoscopic examination, but they soon progress to a

"bull's eye" type of macular atrophy. However, neurologic manifestations such as speech disturbances, loss of recent memory, and inability to learn are common and usually progress to more severe involvement [10]. This was not observed in our case.

Fenestrated sheen macular dystrophy is unlikely to be the diagnosis, since it is characterized by small red fenestrations occurring in the central macular zone [11]. On the other hand, the findings of the autosomal dominant central areolar choroidal and pigment epithelial dystrophy unassociated with flecks or drusen are compatible with our case [10]. This dystrophy is characterized by the development of fine, mottled depigmentation in the macular region that may lead to a "bull's eye" configuration. Visual acuities, ERG and dark adaptation are normal, but the EOG may be subnormal. During the fourth and fifth decades of life there is a slow progressive deterioration of visual acuity, followed by an enlargement of the depigmented area.

We can not rule out Stargardt's disease (most often reported as an autosomal recessive disease), since in some patients the atrophic changes in the macula are not associated with flecks. Furthermore, the angiographic "dark choroid" may not be present in early life. On subsequent follow-up, it is possible to observe flecks as well as signs of diffuse storage of lipofuscin in the RPE [10].

A benign, concentric annular macular dystrophy, first described by Deutman [12] in 1974, is an autosomal dominant disease characterized by the development of paracentral ring scotomas associated with a "bull's eye" maculopathy. Electrophysiologic and color vision tests were normal or slightly abnormal. However, after 10 years of follow-up [13], these patients developed a more generalized tapetoretinal dystrophy which compromised rod and cone function. Thus, this diagnosis can not be excluded.

A definite diagnosis for this patient can not be made on the basis of her recent findings. A period of follow-up will be necessary to analyze the progression of the disease, which will allow a more precise diagnosis.

SUMMARY

The Axenfeld-Rieger syndrome mesodermal dysgenesis represents a spectrum of developmental disorders characterized by abnormalities of the peripheral cornea, iris and anterior chamber angle associated with developmental defects of the teeth and facial bones. Sparse descriptions of fundus abnormalities have been made, but none have been well documented. We report, to our knowledge for the first time, a case of Axenfeld-Rieger syndrome associated with bilateral "bull's eye" maculopathy.

Key words: *Axenfeld-Rieger, "bull's eye", maculopathy, glaucoma.*

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