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ABSTRACT

Background:

Retinoschisis is an uncommon disease that affects the macula and peripheral retina most frequently in males. This association to optic disk pit and papilar coloboma has also been reported previously, but not their association with optic disc drusen.

Methods:

This is a case report.

Result:

We present a female, 23 years old, with unilateral peripheral retinoschisis linked to congenital optic disc drusen in the right eye, and optic nerve head hypoplasia in her left eye. We did not observe a macular lesion in the right eye, but peripheral vitreous veil-like membranes were present. The electroretinogram presented a diminished b-wave with a normal a-wave and diminished oscillatory potential in the same eye. In the left eye optic nerve head hypoplasia was present.

Conclusion:

The authors present a case of a female with unilateral juvenile retinoschisis associated to congenital optic disc drusen, and optic nerve head hypoplasia in the left eye that has not previously been described.

Keywords:

congenital retinoschisis,
congenital optic disc drusen,
hypoplastic optic nerve head.

Female with Unilateral Retinoschisis and Congenital Optic Disc Drusen associated with Contralateral Optic Disc Hypoplasia

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Background

Retinoschisis may be hereditary, degenerative or secondary.¹ The former may also be x-linked juvenile retinoschisis or Goldmann-Favre vitreotapetal-retinal degeneration. X-linked juvenile retinoschisis has been called congenital hereditary retinoschisis, an uncommon bilateral disease that develops early in life and is probably present at birth. It is transmitted as an X-linked recessive trait,¹ occurring most frequently in males, although female cases have been reported as transmitted in autosomal dominant² or recessive³ forms. Retinoschisis has been associated with optic disk pit and coloboma,^{4,5} but never with optic disc drusen.

Drusen of the optic disc were first described clinically by Liebreich in 1868. Men and women are equally affected, and bilateral drusen occur in 6% to 85% of cases. The evolution of disc drusen is a dynamic process that is present at birth and continues throughout life, although there have been reported cases of visible drusen or significant optic disc elevation during childhood, which gradually acquires a yellow or straw colour; the buried drusen gradually appears a scalloped at the margin of the disc and produce subtle excrescences on the disc surface that tend to be nasally. Such changes are usually found during the second decade of life.

Finally optic nerve head hypoplasia is a more common optic disk anomaly encountered in ophthalmology practice,⁶ which ophthalmoscopically appears as an abnormally small optic nerve head. It may appear grey or pale in colour and is often surrounded by a yellowish mottled peripapillary halo, bordered by a ring of increased or decreased pigmentation (double-ring sign). Histopathologically, optic nerve hypoplasia is characterized by a smaller than normal number of optic nerve head axons with normal mesodermal elements.

Case Report

Female, 23 aged with bilateral miodesopsias with no familial or personal clinical history of interest. On ophthalmologic exploration visual acuity was:

- Right eye 0,6 = 175 x -0,75 -4,75
- Left eye 1 = neutral

On visual field exploration we found:

- Right eye generalized constriction of the visual field, with a denser defect in temporal periphery (Figure 1).
- Left eye generalized constriction of the visual field (Figure 2).

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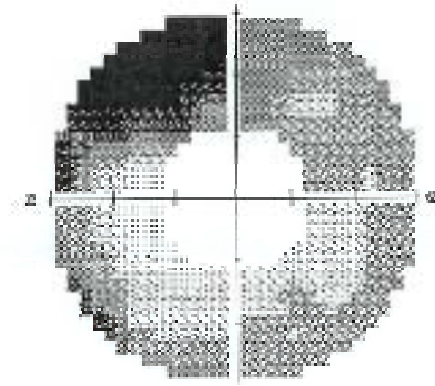


Figure 1: Right eye: generalized constriction of the visual field, with a more dense defect in temporal periphery.

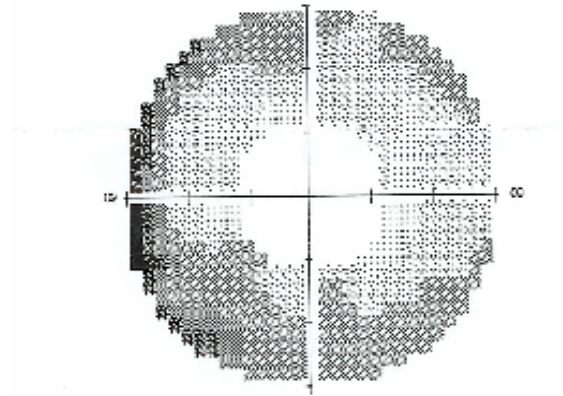


Figure 2: Left eye: generalized constriction of the visual field.



Figure 3: Retinography centered in an inferotemporal retinoschisis, a pseudopapilledema for occult congenital drusen buried within the substance of optic nerve head, a cilio-retinal artery is visible, and incipient bifurcation of major vessels in numerous and sinuous branches.

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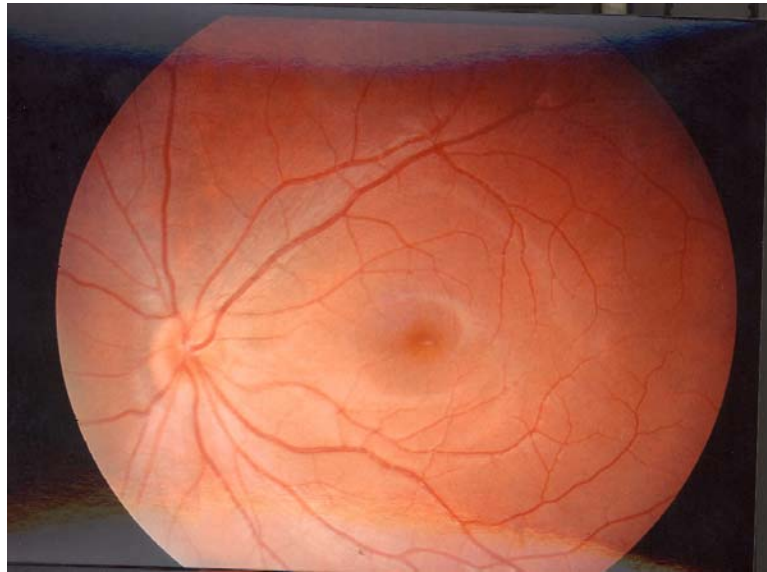


Figure 4: Left eye: we observed optic nerve head hypoplasia (Table 1) with a normal exploration of the retina and macula.

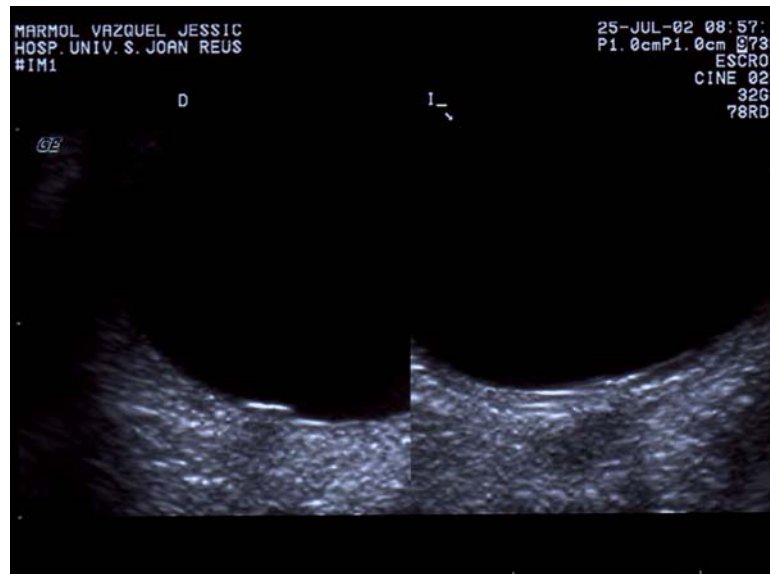


Figure 5: B-scan ultrasonography in the right eye demonstrated the presence of nonvisible drusen as a dense high density signal from the region of the optic disc.

Measure	Reference values	Right eye	Left eye
Disk-to-macula/disk diameter ratio (Zeki <i>et al</i>)	Normal value < 2,9 mm	3 mm	3.9 mm
Horizontal disk diameter (Romano)	Optic nerve hypoplasia if < 3,4 mm	2 mm	1.8 mm
Mean optic disk area (Jonas <i>et al</i>)	Microdisc if < 1,4 mm ²	1.32 mm ²	1.1 mm ²

Table 1: Optic nerve head measures.

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The anterior segment was normal in both eyes, with no inflammatory or degenerative signs, and with no malformations.

On fundus examination we observed:

- *Right eye:* Inferior temporal retinoschisis appearance of anomalously greyish elevated optic nerve head in the nasal region without obscuration of vessels, with a cilio-retinal artery, and incipient bifurcation of major vessels in numerous and sinuous branches (Figure 3). The macula was normal without crystallic stellate maculopathy or foveal schisis. In the peripheral fundus, vitreous veil-like membranes and folds may be seen, denser in the inferonasal vitreous.
- *Left eye:* we observed optic nerve head hypoplasia (Figure 4) the values of which are described in Table 1, where we can compare those with the values described by others authors⁷⁻⁹ as diagnostic of hypoplasia.

The fluorescein angiography presented:

- Right eye peripheral vitreo-retinopathy associated to infero-temporal retinoschisis
- Left eye with no angiographic lesions.

B-scan ultrasonography in the right eye demonstrated the presence of nonvisible drusen as a dense high density signal from the region of the optic disc (Figure 5).

Electrophysiological studies given in the electroretinogram: in the right eye, scotopic and photopic responses present a normal a-wave and a diminished b-wave. The oscillatory potential is generated by reduced photoreceptors, and the implicit times are delayed; in the left eye normal scotopic and photopic responses; in the electrooculogram the responses of both eyes were normal (Right eye: Arden index = 241; left eye: Arden index = 239).

Familial members studied were normal in the fundus examination, but the subject's mother presented vitreous veils in right eye, without retinoschisis or macular degeneration.

Discussion

In the present report we describe a case of a female with a complex bilateral multiple malformation: right eye retinoschisis without macular affection, and congenital occult drusen, and contralateral optic disc hypoplasia.

Retinoschisis in the right eye, with vitreous veil-like membranes, and with a b-wave and oscillatory potential diminished in the electroretinogram can be similar to juvenile retinoschisis but we did not observe any macular lesions (crystallic stellate maculopathy or foveal schisis) which makes diagnosis doubtful.²⁻⁵ The differential diagnosis of retinoschisis also includes Goldmann-Favre retinal degeneration,^{10,11} but this disorder affects both the central and the peripheral retina with retinoschisis similar to juvenile hereditary retinoschisis and pigmentary chorioretinal degeneration that clinically resembles retinitis pigmentosa, together with night-blindness in the first decade of life. The electroretinogram is profoundly abnormal at an early

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age, with an early affect on rod responses and a later affect on cone responses. The electrooculogram may also be markedly abnormal but is not as characteristic for this disorder as the ERG.

Generalized constriction and denser defect in temporal periphery of the visual field in the right eye may be due to optic disc drusen, these may present visual field defects that fall into three general categories: arcuate, quadrant or sector defects, enlargement of the blind spot, and concentric visual field constriction. Generalized constriction in the left eye may be caused by optic nerve head hypoplasia.

The present case was diagnosed as retinoschisis that may be a sporadic case, but a hereditary form cannot be excluded because unilateral presentation in autosomal forms of retinoschisis have been described previously by other authors: Yassur² in dominant forms, and Shimazaki³ in recessive forms. Both authors have also described unilateral cases of retinoschisis in female patients.

In the bibliography we have not been able to find any previous description of the association of retinoschisis and optic disc drusen, but there are descriptions of optic disk pit^{4,5} and papillary coloboma.⁶ Despite optic disc hypoplasia being frequent in the population no one reference to associate of it with retinoschisis or optic disc drusen in the contralateral eye has been described.

The authors think we are in front a new description of multiple bilateral malformation that affects the optic disc and the retina.

Reference

1. George ND, Yates JR, Moore AT. X linked retinoschisis. *Br J Ophthalmol* 1995; 79: 697-702.
2. Yassur Y, Nissenkorn I, Bern-Sira I, Kaffe S, Goodman RM. Autosomal dominant inheritance of retinoschisis. *Am J Ophthalmol* 1982; 94: 338-343.
3. Shimazaki J, Matsushashi M. Familial retinoschisis in female patients. *Documenta Ophthalmologica* 1987; 65: 393-400.
4. Alexandrescu M, Thurn G. Foveal optic disk with central retinoschisis. *Ber Zusammenkunft Dtsch Ophthalmol Ges* 1975; 73: 149-152.
5. Morales J, Teus MA, Pérez P, Bermúdez L, Arranz E. Fosea papilar congénita asociada a retinosquisis foveolar. *Arch Soc Esp Oftalmol* 2001; 76: 327-330.
6. Hotta K, Hirakata A, Hida T. Retinoschisis associated with disc coloboma. *Br J Ophthalmol* 1999; 83:123-124.
7. Jonas JB, Gusek GC, Guggenmoos-Holzmann I, Naumann GOH. Variability of the real dimensions of normal human discs. *Graefes Arch Clin Exp Ophthalmol* 1988; 226: 332-336.
8. Romano PE. Simple photogrammetric diagnosis of optic nerve hypoplasia. *Arch Ophthalmol* 1989; 107: 824-826.
9. Zeki SM, Dudgeon J, Dutton GN. Reappraisal of the ratio of disc to macula/disc diameter in optic nerve hypoplasia. *Br J Ophthalmol* 1991; 75: 538-541.
10. Nasr YG, Cherfan GM, Michels RG, Wilkinson CP. Goldmann Favre maculopathy. *Retina* 1990; 10: 178-180.
11. Käheimo K, Tuppurainen K, Män T, Järvi M. Clinical features of Goldmann Favre syndrome. *Acta Ophthalmol Scand* 1999; 39: 459-461.