

Optic nerve Drusen: Find the differences

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RESUMO

Introdução: Os drusens do disco ótico são anomalias congénitas do desenvolvimento da cabeça do nervo ótico, correspondendo a depósitos hialinos calcificados, localizados anteriormente à lâmina crívosa. O seu diagnóstico é maioritariamente acidental, em doentes normalmente assintomáticos.

Material e Métodos: Os autores apresentam 5 casos clínicos de doentes com idades de apresentação compreendidas entre 6 e 12 anos, observados na Consulta de Oftalmologia Pediátrica e Estrabismo, à qual foram referenciados por diferentes motivos.

Resultados: Nos casos clínicos apresentados os motivos de consulta foram diminuição da acuidade visual, estrabismo divergente, cefaleias com suspeita de papiledema e rotina. O exame oftalmológico e os meios complementares de diagnóstico realizados, nomeadamente retinografia, ecografia ocular, tomografia de coerência ótica e campos visuais, contribuíram para o diagnóstico de drusens do nervo ótico. Foram ainda encontrados erros refrativos em 4 dos casos descritos.

Conclusão: Salienta-se a importância de uma história clínica e observação detalhadas para o diagnóstico diferencial e despiste de patologias oftalmológicas concomitantes, em doentes com drusens do disco ótico e seus familiares.

Palavras-chave

Drusen disco ótico; papiledema; ecografia; tomografia de coerência ótica; diagnóstico diferencial.

ABSTRACT

Introduction: Optical disc drusen are congenital developmental anomalies of the optic nerve head, corresponding to calcified hyaline deposits, located anteriorly to the lamina cribrosa. Patients are usually asymptomatic and the diagnosis is mostly accidental.

Material and Methods: The authors present 5 cases of patients aged between 6 and 12 years referred to Pediatric Ophthalmology and Strabismus outpatient clinic for different reasons.

Results: Patients were referred for decreased visual acuity, exotropia, headache with suspected papilledema and routine. The ophthalmological examination and diagnostic exams performed, including fundoscopy, ocular ultrasound, optical coherence tomography and visual fields, all contributed to the diagnosis of optic disc drusen. Refractive errors were also found in 4 of the cases described.

Conclusion: We emphasize the importance of a detailed clinical history and observation for the differential diagnosis and screening of concomitant ophthalmic pathologies in patients with optic disc drusen and their relatives.

Keywords

Agnosia; Alzheimer disease; dementia; visual cortex.

INTRODUCTION

The word drusen whose german origin dates back to the sixteenth century, used to denominate crystallized spaces within rocks. The initial description of optic disk drusen was made by Müller in 1858¹.

Optic disc drusen (ODD) are congenital developmental abnormalities of the optic nerve head and consist of calcified hyaline deposits located above the cribriform plate².

There are three pathophysiological theories: Tso, change of axonal metabolism leads to formation of mitochondrial calcium deposits and axonal destruction and their extrusion; Seitz, who believes that the disruption of axonal transport leads to axoplasmic stasis and disintegration of nerve fibers; to AntCliff Spalton ODDs result of optic nerve hereditary dysplasia and vascularization³.

Their prevalence is between 0.3 to 2.4%⁴ and are bilateral in 70% of the cases⁵.

It is most common in Caucasians and there is a slight predominance of females (61% to 71%)¹. The age of presentation is extremely variable but most cases are diagnosed between the 2nd and 3rd decade of life in a routine eye examination⁴.

ODDs can be divided in two anatomical subgroups according to Roh classification⁴:

1. Hidden or deep papillary drusen: usually appearing as elevated papilla with poorly defined edges, without visible nodular images. The presence of hidden or buried drusen is confirmed with echography.
2. Visible or superficial papillary drusen: defined as yellowish nodular images having variable size, in a generally elevated papilla with poorly defined edges.

The visible drusen can also be classified on the basis of their abundance as scarce (between 1 and 5), numerous (between 5 and 10) and very abundant (when the number exceeds 10 and occupies the entire papilla)⁴.

The visual acuity (VA) is usually preserved². Lorentzen⁶ describes a case of decreased VA in 91 eyes, Rosenberg⁷ one case in 151 eyes and Mustonen⁸ 4 cases in 307 eyes.

However, these patients may present visual fields defects that can worsen over time. The described visual field defects in adults involve the inferior nasal quadrant,

enlargement of the blind spot (23 to 60 %) and concentric narrowing. The arcuate defects appear to be the main cause of visual field loss in ODD¹.

The diagnosis is mostly accidental, usually in asymptomatic patients².

Numerous pathologies are described in association with ODD, the most common are pigmentary retinopathy and angioid streaks with or without pseudoxanthoma elasticum¹.

The ODDs may mimic papilledema, therefore, the differential diagnosis is essential between these two entities, avoiding unnecessary invasive examinations².

Several additional tests may contribute to ODD diagnosis, the most used being ocular ultrasound and more recently, the auto-fluorescence. Other tests that can be performed are computed tomography of the orbits and optical coherence tomography.

The authors present 5 cases of patients aged between 6 and 12 years referred to Pediatric Ophthalmology and Strabismus outpatient clinic for different reasons: decreased VA, divergent strabismus, headache with suspected papilledema and routine examination.

CASE REPORTS

Case 1

6 year-old girl, mother with congenital glaucoma. Referred to Pediatric Ophthalmology outpatient clinic for decreased VA in the left eye (LE) in preschool screening.

Her best corrected visual acuity (BCVA) was 10/10 in both eyes.

Right eye (RE) fundoscopy showed irregular elevation of the optic disc and LE an optic disc with normal contours (Fig. 1).

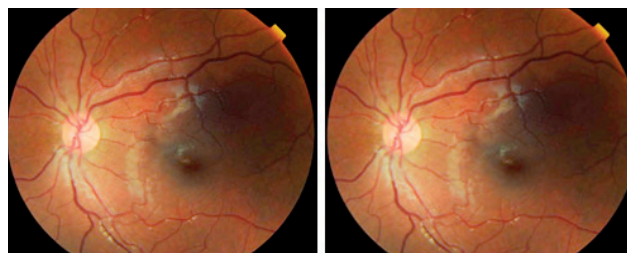


Fig. 1 | RE and LE Retinography

OCT revealed an elevation of the optic nerve head with irregular inner contour and an abrupt end with a hyporeflective space in the RE (Fig. 2).

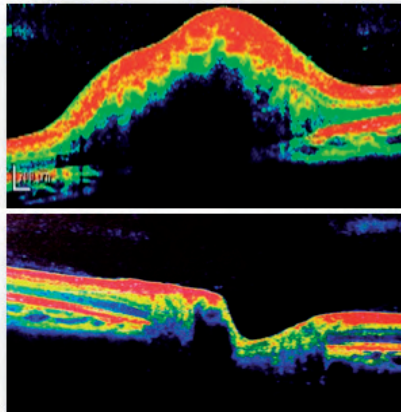


Fig. 2 | RE and LE OCT

Computerized perimetry showed a low reliability index (RF 40.9 RE and 28 LE) and catch trials 3/11 (+); 6/11 (-) in RE and 3/12 (+); 4/13 (-) in the LE (Fig. 3).

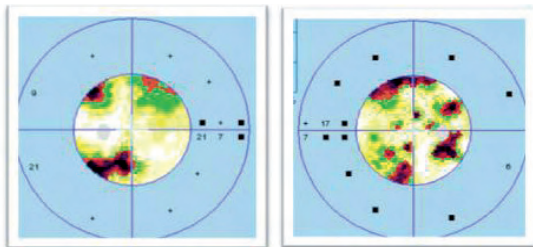


Fig. 3 | RLE Visual Fields

After one year of follow-up, she presented BCVA of 10/10 in RLE.

Case 2

9 year-old girl with delayed psychomotor development, fronto-occipital cranial angiomas and facial dysmorphism. Maternal uncle with ODD.

She was referred from neurology clinic with headache and a diagnosis of papilledema already medicated with acetazolamide. She had performed CT and MRI-EC.

Showed hyperopia of +4.75 RLE under cycloplegics. The fundus examination revealed optic discs with blurred edges (Fig. 4).

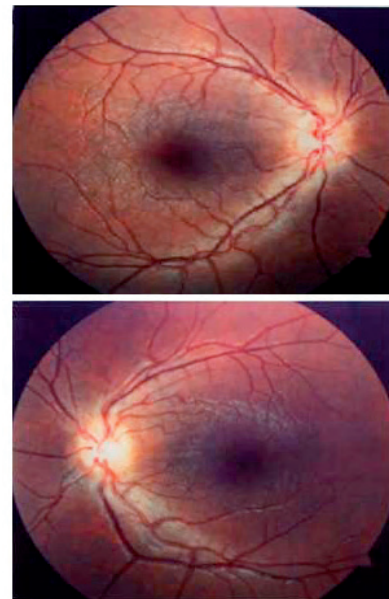


Fig. 4 | RRE and LE Retinography

The ultrasound revealed bilateral ocular papillary calcification compatible with ODD (Fig. 5).

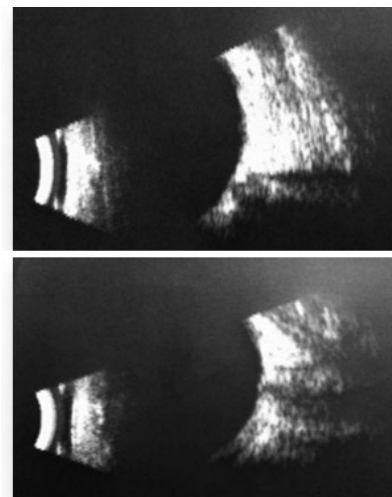


Fig. 5 | RE and LE ultrasound

The optical coherence tomography demonstrated elevation of the optic nerve head with irregular inner contour in RLE (Fig. 6).

Visual field analysis revealed a temporal visual field defect in RE and a inferior temporal defect in LE, however the RF was 22.7 in RE and 19.2 in LE and catch trials of 3/11 (+), 2/11 (-) in RE and 4/ 13 (+) and 1/13 (-) in the LE (Fig. 7).

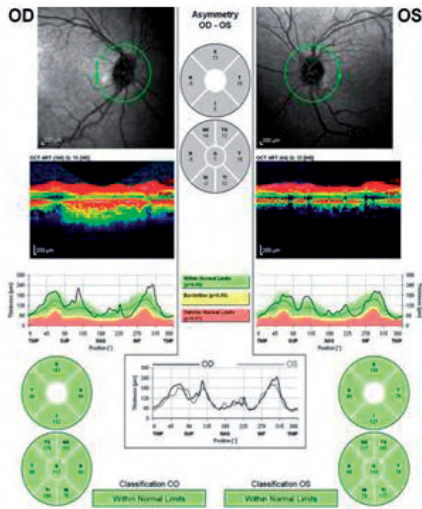


Fig. 6 | RE and LE OCT

After a year and a half of follow-up, BCVA showed: RE (+0.75 + 4.50 x 100 °) = 8/10; LE (+4.25 + 1.25 x 85 °) = 9/10.

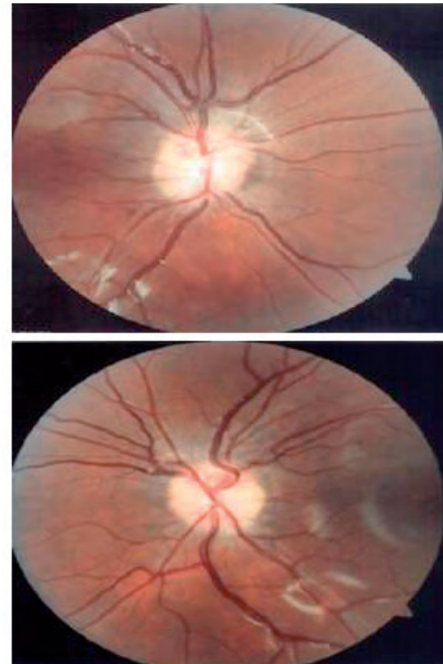


Fig. 8 | RE and LE Retinography

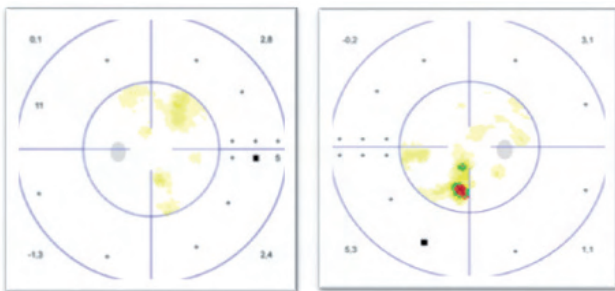


Fig. 7 | RE and LE Visual Field

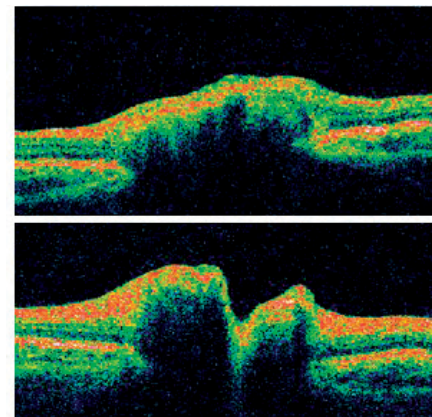


Fig. 9 | RE and LE OCT

Case 3

6 year-old girl, appeals to query for preschool screening. Father with strabismus and astigmatism and a maternal aunt with amblyopia.

At first outpatient clinic visit she had BCVA of (-1.00 -0.50 x 100 °) = 9/10 RE and (-0.50 -0.50 x 100 °) = 10/10 LE.

Fundus examination presented optic discs with ill-defined top edges, larger in RE and vascular tortuosity (Fig. 8).

B-scan revealed papillary drusen in RLE, most evident in the RE.

OCT demonstrated elevation of the optic nerve head with irregular inner contour in both eyes that contributed to the diagnosis of ODD (Fig. 9).

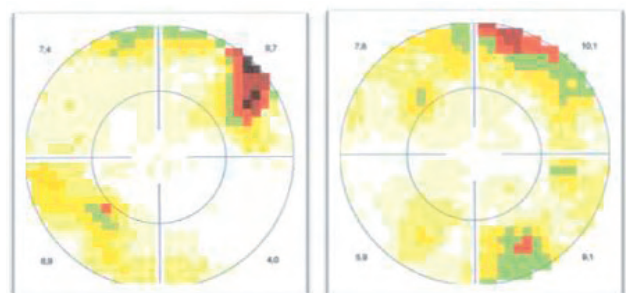


Fig. 10 | RE and LE Visual field

Perimetry showed superior temporal defect in RE and superior nasal defect in LE with reliability index of 4.1 and 4.3, respectively (Fig. 10).

After 13 years of follow-up, BCVA was: RE (-5.00 -1.75 x175 °) = 9/10; LE (-6.00 -1.25 x175 °) = 10/10.

Case 4

13 year-old girl, referred by decreased visual acuity.

At first examination she had BCVA: RE (-3.00 -1.75 x10 °) = 9/10; LE (-3.00 -1.25 x170 °) = 9/10

Fundus examination revealed raised optic disc edges in RLE (Fig. 11).

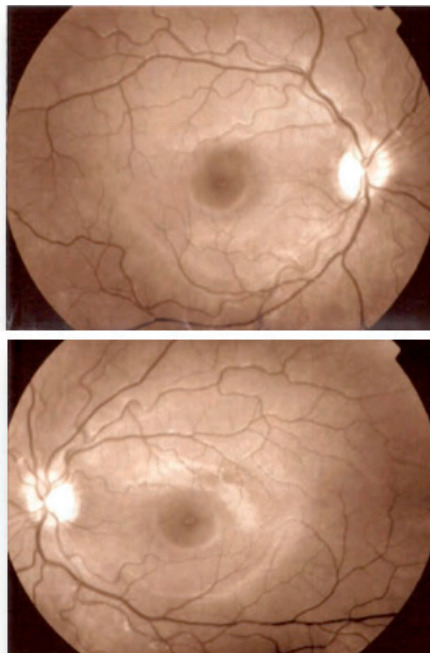


Fig. 11 | RE and LE Retinography

Ocular ultrasound showed bilateral papillary calcification.

OCT was compatible with the optical disc drusens RLE (Fig. 12).

Red-free fundus photography revealed optic disc autofluorescence, characteristic of ODD (Fig. 13).

She had performed many visual field analysis with changes attributed to poor cooperation of the patient, RF at the first examination was 4.7 in RE and 20.8 in the LE (Fig. 14).

After 8 years of follow-up, BCVA was: RE(-4.00 -2.25° x5) = 9/10; LE (-4.00 -2.00 x 150 °) = 9/10.

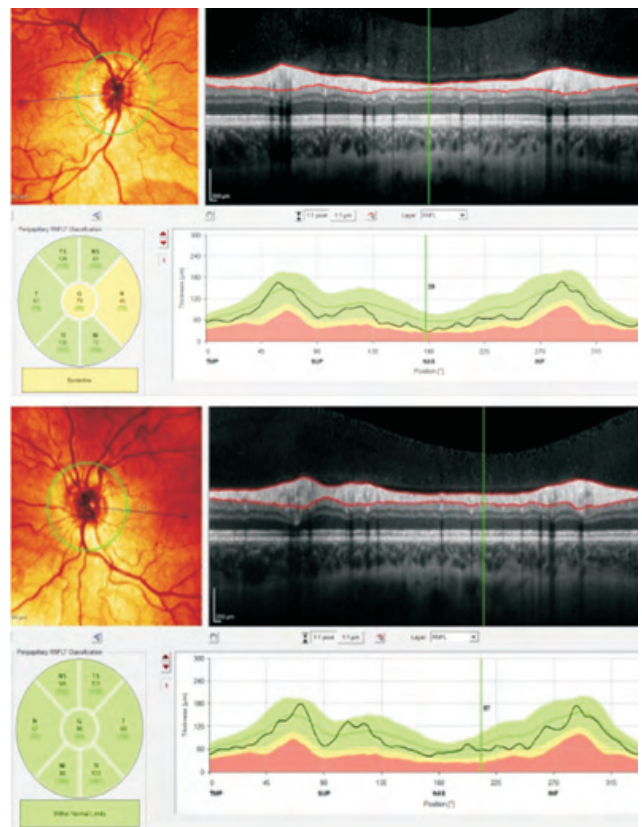


Fig. 12 | RE and LE OCT

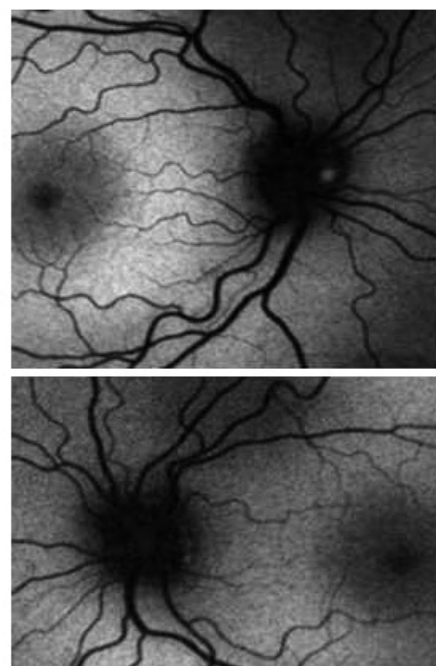


Fig. 13 | RE and LE Red-free fundus photography

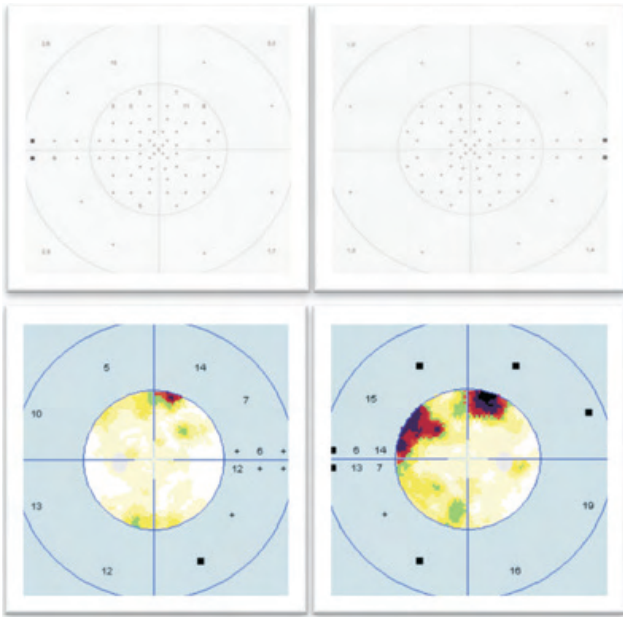


Fig. 14 | RE and LE Visual Field with 1 year of follow-up

Case 5

7 year-old boy with Charcot-Marie-Tooth syndrome type 1A (PMP22 gene), autoimmune thyroiditis unmedicated and delayed psychomotor development, which was referred by a divergent strabismus ophthalmologist

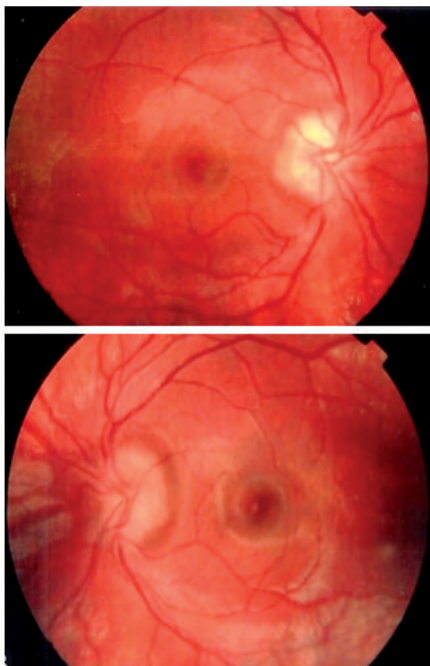


Fig. 15 | RE and LE Retinography

colleague. Family history: myopic mother and a maternal aunt with convergent strabismus.

Fundus examination showed raised edges of both optic discs, more exuberant in RE (Fig. 15).

Red-free fundus photography revealed optic disc autofluorescence in RE confirming the diagnosis (Fig. 16).

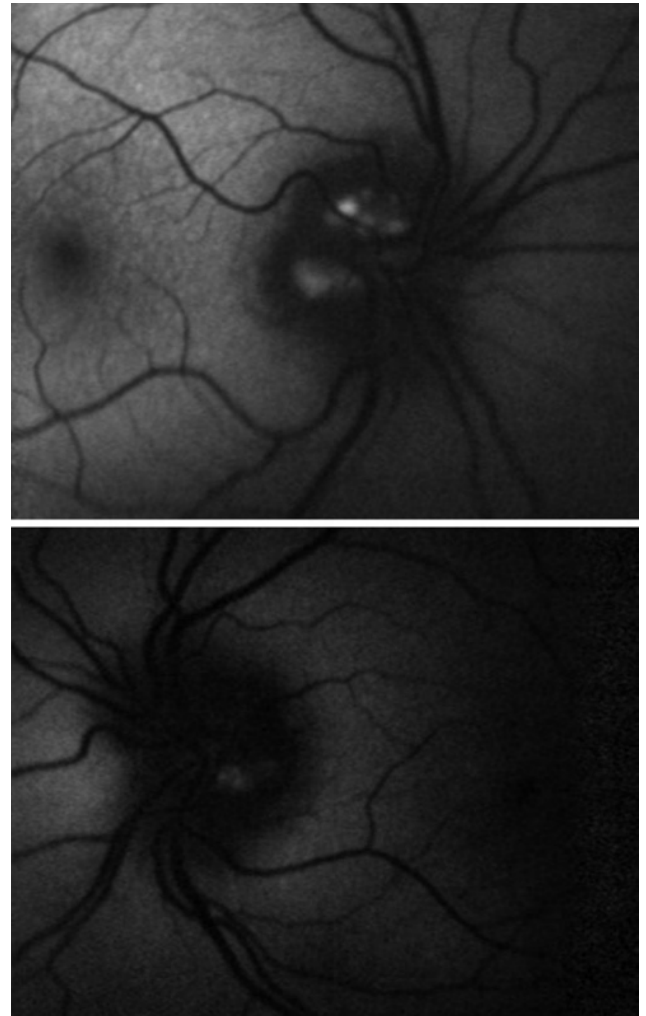


Fig. 16 | RE and LE Red-free fundus photography

DISCUSSION AND CONCLUSION

The ODDs are yellow and hyaline deposits derived from axonal calcified debris, present on the surface of the optic disc or buried within it.

ODD are inherited in an irregular dominant fashion. Thus, it may be useful to examine patient's relatives.

The ophthalmoscopic appearance depends on the location of ODD - superficial or deep. Diagnosing the superficial

Table 1 | Optic disc drusen vs papilledema.

	OPTIC DISC DRUSEN	PAPILLEDEMA
Optic disc appearance	<i>Lumpy bumpy</i> , ill defined optic disc	Hyperaemic, ill defined optic disc
Normal spontaneous retinal venous pulsation	Present	Absent
Peripapillary lesions	Abnormal retinal vasculature	Cotton wool spots, retinal hemorrhage, venous congestion
Visual acuity impairment	Very rare	Later stages
Visual fields defects	Rare Slow progression	Blind spot enlargement
Systemic symptoms	Absent	Headache, náusea, vomiting
Optic disc calcification	Detected with computed tomography and/or ocular ultrasound	Absent
Autofluorescence of the optic disc	Present	Absent

type is easier, but the deep type is more frequent in children².

Retinal vessels of eyes with ODD often exhibit pronounced tortuosity (case 3), venous dilation, abnormally early branching with trifurcations of temporal branches of the central retinal artery in the horizontal midline or vascular loops. The incidence of cilio retinal arteries in patients with DDO is higher than the normal population (15% vs 20-40%)¹.

Differential diagnosis is essencial, especially with papilledema.

Papilledema is associated with hyperemia of the optic disc and peripapillary retinal edema, complicating the visualization of retinal vessels at the disc margin. The presence of exudates, and peripapillary hemorrhages and absence of spontaneous venous pulse are in favor of papilledema. Frontal headache, with nausea and vomiting are symptoms often associated with papilledema (Table 1)².

Decrease VA in eyes with ODD is rare (16 to 28 %), but visual fields defects may occur in about 75% of the cases⁴, being less common in ODD deep type¹³.

In the presence of impaired VA we suspect other causes including accompanying diseases (pigmentary retinopathy, angioid streaks); other concomitant causes unrelated to ODDs; intrinsic to the ODDs; associated complications (peripapillary choroidal neovascularization, anterior ischemic optic neuropathy; central retinal artery or vein occlusion; macular and/or peripapillary serous detachment; central serous papillopathy)^{1,10}.

Several additional tests contribute to the differential diagnosis of ODD, including ocular ultrasound, computed tomography, fluorescein angiography and optical coherence tomography^{1,2,11}.

Due to its high calcium content, these formations are strongly echogenic on ultrasound B mode¹² and this method is one of the most sensitive methods for diagnosis, illustrated in case 1 and 2. In deep or not calcified ODDs, more frequent in younger patients, ultrasound represents a weak contribution.

Orbital and cranial computed tomography may be indicated, even allow to exclude intracranial space-occupying lesion and deep ODD².

Patel et al. showed that optical coherence tomography can differentiate between papilledema and ODD¹¹. Patients diagnosed with ODD by autofluorescence and ocular ultrasound, do not need to be subjected to more clinical and/or imaging research, in the absence of neurological signs².

Red-free fundus photography shows a typical image of autofluorescence of the disks¹. as observed in the case 4 and 5, thus contributing to the diagnosis of ODD.

Although no effective treatment exists for ODD and the periodicity of the follow-up is not consensual, regular observation of these patients is essential to exclude concomitant ocular pathologies and/or complications and should include VA evaluating, fundoscopy, intraocular pressure and visual field testing¹.

Being a major cause of pseudo-papilloedema in children¹, its early diagnosis is critical. A complete ophthalmologic examination and observation of family members, allows the diagnosis, screening for associated diseases and proper orientation.

Case 2 depicts the importance of timely diagnosis of this pathology, which would avoid the cascade of unnecessary interventions (cranial CT, MRI and sometimes Lumbar Puncture), as well as the anxiety of patients and their families.

A case of atypical edema and/or suspected ODD emphasizes the importance of a multidisciplinary clinical evaluation in particular for ophthalmology, neurology and radiology.

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