

Optic disc drusen in children: a diagnostic challenge

Drusas de disco óptico em criança um desafio diagnóstico

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ABSTRACT

In children, optic disc drusen (ODD) are often mistaken for papilledema, this being the principal differential diagnosis. This report describes the case of an 11-year old patient with ODD, in which the condition was initially diagnosed as papilledema, and the patient referred for pulse therapy. Fundoscopic examination is important because it is the first examination conducted by the ophthalmologist that is capable of revealing some characteristics of ODD that will aid in the differentiation between this disease and papilledema. In cases of ODD, the optic disc presents blurred margins and elevation of the disc borders, with clearly defined vessels at the papilla border. The differential diagnosis of ODD in children is challenging and requires appropriate management and follow-up to avoid iatrogenesis.

Keywords: Optic disc drusen; Papilledema; Ultrasonography; Pseudopapilledema; Child

RESUMO

As drusas de disco óptico (DDO) em crianças são frequentemente confundidas com papiledema, sendo este o principal diagnóstico diferencial. Este artigo relata o caso de uma paciente de 11 anos com DDO, no qual o quadro foi inicialmente diagnosticado como papiledema, e a paciente encaminhada para pulsoterapia. O exame fundoscópico é importante por ser o primeiro exame realizado pelo oftalmologista que é capaz de revelar algumas características das DDO que auxiliarão na diferenciação do papiledema. Nos casos de DDO, o disco óptico apresenta margens mal definidas e bordas elevadas, com vasos na margem da papila bem definidos. O diagnóstico diferencial das DDO em crianças é desafiador e requer conduta e seguimento adequados para evitar iatrogenias.

Descritores: Drusas de disco óptico; Papiledema; Ultrassonografia; Pseudopapiledema; Criança

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INTRODUCTION

The principal differential diagnosis of optic disc drusen (ODD) is pseudopapilledema. The etiology of ODD is unknown; however, it may be inherited as an autosomal dominant trait with variable penetrance.⁽¹⁻³⁾ The term drusen comes from the German word for tumor, edema or intumescence.⁽³⁾

Prevalence ranges between 0.2-2% in adults and 0.37-1% in children, with the disease being more common in females (61-71%) and in Caucasians (85%). In 70-91.2% of cases, ODD is bilateral.⁽²⁾ The condition is visible in only 0.34% of cases.⁽⁴⁻⁶⁾

ODD may be granular or nodular, visible or buried in the retinal nerve fibers. This deeper location is more common in children and often mistaken for papilledema.⁽⁵⁾ There are normally no symptoms; however, visual field changes may be identifiable at funduscopy.

This paper describes a child with unilateral ODD and highlights the diagnostic challenge presented by cases in which ODD can mimic papilledema.

Case report

An 11-year old girl with normal eyesight presented with a white mark in the center of her left eye over the preceding 18 months. She had been diagnosed with papilledema at another healthcare facility and referred for pulse steroid therapy, which was contraindicated due to a suspected reaction to the influenza vaccine. Magnetic resonance images of the brain and orbits were normal. Uncorrected visual acuity was 20/20 in both eyes. Biomicroscopy was normal in both eyes. Funduscopy results were normal in the right eye; however, investigation of the left eye showed an elevated optic nerve head with blurred contours, and drusen in the upper region. Retinal blood vessels were clearly defined in the papilla border and visible along their entire trajectory.

Retinography of the left eye revealed autofluorescence of the drusen. Optical coherence tomography (OCT) of the optic nerve was normal in the right eye, but revealed a reduction in fibers in the temporal region of the left eye. Ultrasonography of the right eye was normal. In the left eye, ultrasonography detected a highly reflective lesion with a posterior acoustic shadow that remained even at a low-gain setting, being compatible with calcification and/or drusen of the optic papilla. Diagnosis of ODD was confirmed. Humphrey 30-2 visual field was normal for the right eye, with an increased blind spot in the left eye. The child's parents were submitted to retinal mapping. The condition was found in the mother alone, with drusen being present in her left eye.

DISCUSSION

ODD is a challenging diagnosis for ophthalmologists. Funduscopy is extremely important, since ODD can mimic papilledema, the principal differential diagnosis. Certain characteristics facilitate differentiation. In ODD, the optic disc head is elevated, and the contours are blurred, with clearly defined vessels at the edge of the papilla that are visible throughout its trajectory, with no veiling.⁽¹⁾ These characteristics were present here, and a hypothesis of ODD was made.

Supplementary tests can facilitate confirmation of diagnosis, with ultrasound being the most sensitive, since it enables areas of calcification to be visualized through the shadow cone behind the hyperechogenic image, as found in the present case.^(2,5)

Photographic evaluation without injection of contrast enables visualization of the autofluorescence of the optic disc that is characteristic of drusen. Fluorescein angiography is more sensitive than funduscopy but not as sensitive as ultrasound.^(2,5)

In up to 75% of cases, visual field abnormalities are present⁽⁵⁾ and consist of specific superior and inferior arcuate changes. Since this altitudinal defect can also be found in papilledema, it should be meticulously investigated to differentiate between these two conditions. The most common of these abnormalities is an increased blind spot (68%).⁽²⁾

Computed tomography (CT) of the brain and orbits showed hyperintense images (calcifications) adjacent to the optic nerve. Although CT is not the first choice method for diagnostic purposes, it is mandatory when papilledema is suspected in order to exclude the possibility of an intracranial tumor.^(2,5)

OCT can detect the disappearance of optic disc excavation and optic papilla protrusion. The peripapillary retinal nerve fiber layer (RNFL) is generally affected to a greater extent in the inferior nasal quadrant, which is the most common site of ODD. The larger the area of the optic disc occupied by the ODD, the greater the RNFL abnormalities. Reduction in the thickness of this layer may be associated with visual field abnormalities.^(2,5,7-9)

OCT is important when diagnosing papilledema/ODD, as it is capable of distinguishing differentiating characteristics: the RNFL, ganglion cell layer and external and internal plexiform layers are all thicker in papilledema. In ODD, the retinal pigment epithelium layer, the RNFL and internal plexiform layer are thin; however, the ganglion cell layer and external plexiform layer are thick.⁽⁵⁾ Nevertheless, a further reduction in the thickness of the RNFL may occur over time and visual field lesions may develop.⁽⁹⁾ Here, OCT showed hypo-reflective nodular lesions, albeit with hyper-reflective borders, in the prepapillary region, suggestive of ODD.

OCT of the RNFL showed a reduction in thickness in the temporal quadrant of the left eye. The growth of ODD results in direct compression of the RNFL, reducing thickness. Conversely, in papilledema and pseudopapilledema resulting from intracranial hypertension, there is a marked increase in RNFL thickness in an initial phase.⁽⁵⁾

Here, the fact that OCT showed a reduction in RNFL thickness in the left eye is a sign of axonal loss yet to be identified at campimetry.^(5,9)

ODD is a congenital condition that generally becomes apparent in the first or second decade of life, with cases having been reported, however, in patients of 7 to 73 years of age. Here, funduscopy in the patient's mother revealed ODD in her left eye, one more positive factor supporting a diagnosis of ODD.⁽³⁾

ODD is benign; however, it can be associated with complications, the majority of which are vascular in nature, including anterior ischemic optic neuropathy leading to ischemia, subretinal neovascularization, branch retinal vein occlusion or central retinal artery occlusion, and retinal hemorrhage.⁽²⁻⁵⁾

Transitory episodes of reduced visual acuity may occur. Since there is no cure and patients with ODD are generally

asymptomatic, they should be followed up periodically by an ophthalmologist.

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