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Imaging of Drusen of the Optic Disc: *About 1 Case Report*

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Abstract: *Optic disc drusen are acellular deposits situated in the optic disc nerve head found in 2.4% of the population. Although benign, they most often involve loss of visual field and may lead to cecity. We herein describe the case of a 12 year old boy with a calcified optic disc drusen and highlight the different radiological aspects seen in this pathology.*

Keywords: Disk, Drusen, Imaging, Optic.

Introduction :

Optic disc drusen are benign congenital anomalies of the optic nerve characterized by acellular, often calcified nodules within the substance of the optic nerve head. (1) Yet, if left untreated, it could evolve to cecity due to various complications. We describe a case of a symptomatic visual field constriction in one eye of a 12 year old male with an optic disc drusen. The patient consulted in the ophthalmology department of the Universal hospital center "HASSAN II" of Fes for a slowly progressive loss of vision and the peripheral visual field of the left eye. The diagnosis was at first suspected by fundoscopy, and was later confirmed by B-scan ultrasonography and CT-scan. In this article, we are going to discuss different imaging aspects of optic disk drusen based on 1 case report.

Case report:

A 12 year old boy was admitted to the outpatient ophthalmology department In the Universal hospital center "HASSAN II" of Fes, due to a visual field constriction in the left eye, associated with a loss of vision, increasing since the age of 10.

There was no similar case in the family, no medical history and the patient was otherwise in good health.

On physical examination, our patient had a visual acuity of 12/10 in the right eye, and 0,5/10 in the left eye. The eye tone was measured to 14 in the right eye and 13 in the left eye.

Our patient had, however, a normal psychomotor development, as well as a good weight and height growth.

A fundoscopy exam showed a papilledema, the nerve fiber layer was not swollen and there was no hemorrhage. (fig. 1)

The size of papilledema was calculated to 3,8 x 4,4 mm (versus normal size 1,7 x 1,9 mm).

There was an ectasia of superficial vessels of the optic disk and obscurations of the retinal vessels as they cross the border of the optic disk.

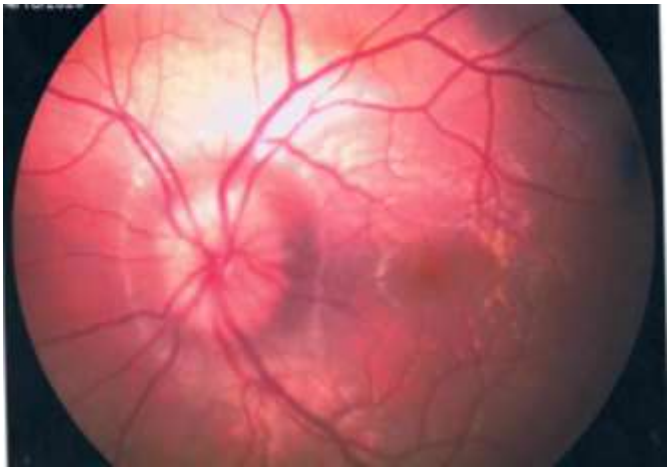

Fig. 1 : Fundus photograph of the left eye of our patient showing a pseudo-papilledema.

Otherwise, the right eye was clear of any abnormality. (Fig.2)

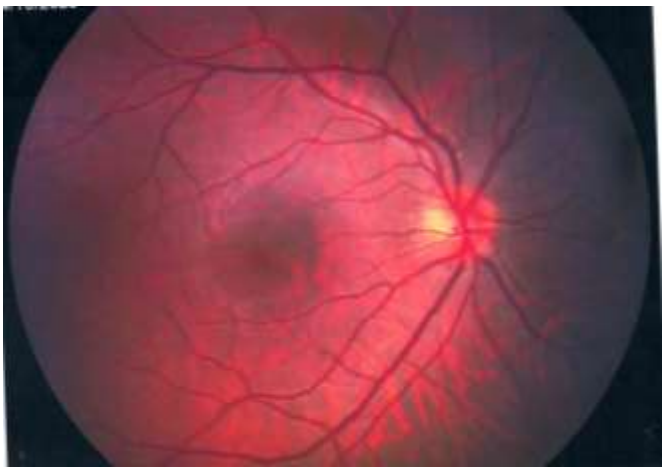

Fig. 2 : Fundus photograph of the right eye of our patient with a normal papilla.

He was then referred to the radiology department, for a CT scan because they suspected an intracranial hypertension due to the visual loss and the papilledema that was viewed in the fundoscopy.

Imaging with computerized tomography was performed at first which was done in 5 mm sections with thin reconstructions (2,5mm), showed a calcification in the emergence of the optic nerve in the left eye. (Fig. 3)

Fig. 3 : Computerized tomography scan of our patient showing an hyperdense signal in the optic nerve head of the left eye consistent with calcified optic disk drusen.

The hypothesis of a retinoblastoma was discussed at first, but was quickly rejected due to the age of our patient, and the absence of tissular component.

A B-scan echography detected calcium deposits in the optic nerve head and was viewed as hyperechoic round structures with acoustic shadowing. (Fig.4)

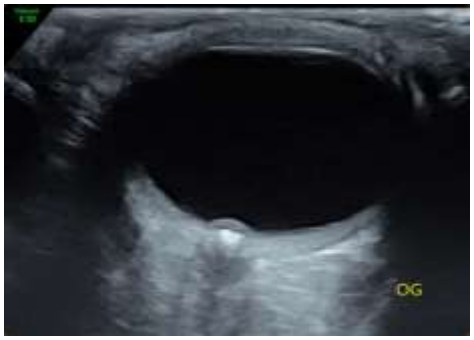

Fig. 4 : B-scan ultrasound of the left eye of our patient showing increased reflectivity from the optic nerve head consistent with a calcified optic disk drusen.

The right eye was normal in both B-scan echography and computer tomography. (Fig.3, Fig.5)

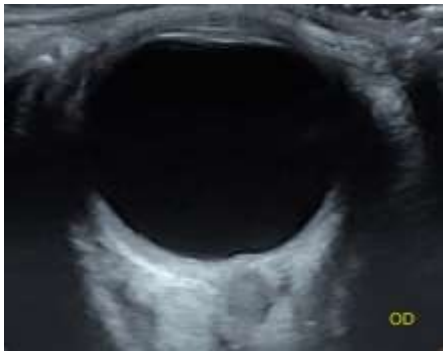

Fig. 5 : B-scan ultrasound of the right eye of our patient confirming a normal papilla.

Discussion:

A. Pathogenesis :

Optic disk drusen are benign congenital anomalies of the optic nerve characterized by acellular, often calcified nodules within the substance of the optic nerve head. (1)

These nodules are formed by calcified hyaline deposits accumulated on the inner side of the lamina cribriformis (from the vitreous body) (2)

Although the mechanism of drusen formation has not been fully determined, they are thought to result from disturbance of the axonal transport and deposition of debris in eyes with a small scleral canal and a congenitally abnormal disc vasculature, allowing transudation of plasma proteins (1).

B. Epidemiology :

Their prevalence ranges from 3.4 to 24 per 1,000 according to clinical studies and 1-2.4% in histological studies. (3)

The prevalence of optic disk drusen is higher among women and whites (5) and involvement is bilateral in 75% of the time. (4)

Ophthalmic and systemic diseases commonly associated with drusen are retinitis pigmentosa, angioid streaks, Usher syndrome, Noonan syndrome and Alagille syndrome (3)

C. Symptoms :

Initially optic disk drusen seem not to disable visual functions significantly, because in most cases visual acuity is well preserved. Visual fields, however, are often abnormal and can further deteriorate without the patient being aware of these defects. (3)

Transient amaurosis or even permanent monocular blindness was reported in literature, in the absence of signs of vascular complications. (3)

Optic disk drusen rarely cause the decline of visual acuity alone. In such patients with normal visual fields, another reason for the decreasing of the visual acuity should be sought. For that matter, it is crucial to not overlook a compression by a mass lesion in patients with severe visual field and visual acuity loss. (3)

An important differential diagnosis is glaucoma, as slowly progressive, asymptomatic visual field defects occur commonly in optic disc drusen while cupping is not present. (1)

Another concomitant entity to consider in light of the visual field loss is pseudotumor cerebri (idiopathic intracranial hypertension). (8)

D. Diagnostic imaging:

1- B-scan ultrasonography :

B-scan ultrasonography is considered the gold standard imaging technique to identify optic disk drusen because of its ability to detect calcium deposits. (5, 10)

On the other hand, it provide a poor resolution of the optic disk drusen and does not offer any information concerning the neuroaxonal integrity of the optic nerve and retinal structures. (10)

The echogenic focus is usually spotted at the junction of the retina and the optic nerve and tend to project posteriorly rather that protrude into the vitreous body. (7) It appears hyperechoic with posterior shadowing as a result of the calcifications.(7)

Lesions are most commonly ovoid, with their long axis perpendicular to the long axis of the optic nerve. Large lesions are more rounded than smaller ones. (7)

The use of high gain facilitates to detect optic disk drusen. (8)

The Color Doppler Imaging can be helpful in establishing the risk profile for ocular vascular accidents in patients with optic disk drusen. It also could be of a great value in considering long term antithrombotic treatment in patients with optic disk drusen.(1) It was also reported that the extent of visual field defect was proportional to blood flow velocity in the central retinal artery. However, this could be as a result of a secondary phenomenon reflecting inner retinal atrophy. (11)

Some authors reported viewing by the Color Doppler imaging of the vascular supply of the optic nerves attenuated flow velocities of the small retrobulbar vessels.(1)

However, calcified mass in the region of the optic disk could be viewed in patients who present other conditions that optic disk drusen.

The main differential diagnosis is the retinoblastoma, which appears as a solid, partly or entirely calcified, smoothly marginated mass in the posterior aspect of the globe that protrudes into the vitreous body. The tumor is not essentially located at the optic disk, nonetheless, it may invade it. (7)

It can also be difficult to distinguish the optic disk drusen from the astrocystic hamartoma of the retina and optic disk, especially if it occurs at the disk and is small. It appears as a calcified mass that does not usually extend into the vitreous body. However, these tumors are most commonly unilateral and could be associated with retinal detachment. (7)

It is however more difficult to detect small or buried optic disk drusen that are not yet calcified.(6)

The benefit of this method is that the whole area of the optic disk is visualized by a sweeping movement of the ultrasound probe. (6)

2- Orbital computed tomography :

The diagnosis of the optic disk drusen by B-scan ultrasonography was considered a more reliable technique than CT-scans. (6, 10)

Neuroimaging by CT-scan is essencially used when the clinical presentation requires further investigation to exclude an intracranial lesion. (6)

The optic disk drusen appears as hyperdense spots on CT scans. (3)

When it comes to deep buried drusen, this technique was stated to be superior to ophthalmoscopy,(3) but not as sensitive as echography since the slice thickness of the CT usually used is up to 1.5 mm, which could not be thin enough to spot smaller drusen. (5)

Hence, using this technique might miss a considerable number of optic disk drusen. (3)

Moreover, the great cost, the more complex handling make CT unfitting for standard diagnosis in patients with presumed optic disk drusen (3), especially when it comes to children regarding the excess of radiation (5).

3- Magnetic resonance imaging MRI :

MRI is undeniably more sensitive for diagnosing intracranial abnormalities, which should be ruled out in case of a clinical presentation of increased intracranial pressure. (6) It is also indispensable in cases of disk oedema. (11) However, it is not reliable for identifying optic disk drusen considering that drusen are formed by calcified bodies. (9)

Also, in most cases that were reported, it didn't reveal any changes in the region of the optic nerve head consistent with the diagnosis of drusen. (6)

The optic disk drusen could appear as an asignal in all sequences related to the calcium deposits in optic nerve head. (Fig.6) (13)

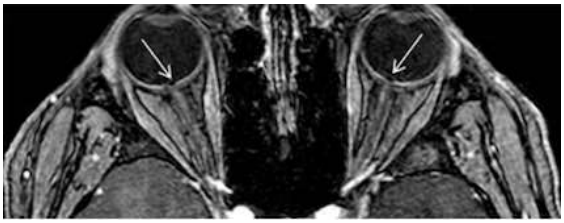

Fig.6 : MRI in 3D T1 sequence injected in echo gradient, centered on the anterior visual pathways of a young patient with a clinical presentation of a right NOIA, but in which the final diagnosis was a bilateral optic disk drusen. (white arrows) (13)

E. Diagnostic by non-imaging techniques:

4- Fluorescein angiography :

Fluorescein angiography (FA) was essential to differentiate between buried optic disk drusen and true disk oedema. (11)

The fluorescein angiography findings comparing eyes diagnosed with pseudo-papilledema to those of eyes with papilledema described the "increased vascularity" seen more often in papilledema. Other studies have differentiated optic disc drusen by noting "disc staining" from optic disc edema characterized by "disc leakage". (14)

However, fluorescein angiogram criteria to distinguish optic disk drusen and true disk swelling have not been published. (11)

It can show a pseudoedema caracterised with a simple impregnation without diffusion. In the case where the drusen is not visible in funduscopy, it is necessary to complete the investigation with the optical coherence tomography (4).

The optic disc drusen constitute one of the four causes of pseudoedema with myeline fibers, small papilla in hypermetropia and pseudoedema with hyperemia found in Leber's hereditary optic neuropathy. (4)

5- Optic disk autofluorescence:

In visible optic disk drusen, the calcium salt deposits or mitochondrial pophyrins will produce bright hyperfluorescent foci in the disk. (11) (Fig.7)

Bright excitation flashes used in this technique can recognize most of the visible drusen. Buried drusen, on the other hand, are only apparent in half of the cases that were diagnosed by ultrasonography, and this is presumptively due the thickness of overlying neural tissue. (11)

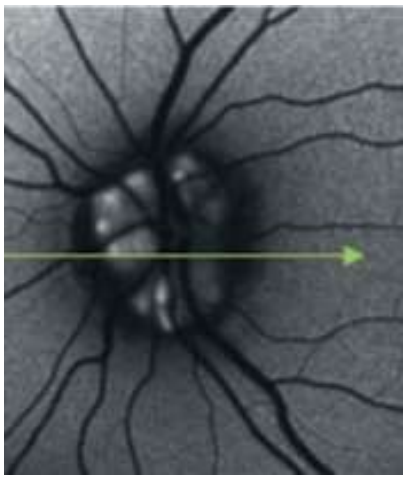

Fig.7 : Visible optic disk drusen using autofluorescence technique (11)

6- Optical coherence tomography :

Recent studies showed that OCT with EDI was more sensitive than ultrasonography for the detection of optic disk drusen. (11)

The swept-source optical coherence tomography (SS-OCT) with enhanced depth imaging (EDI) described the optic disk drusen as irregular or spheroidal hypo-reflective structures (demonstrating a uniform internal structure), with hyper-reflective foci or borders or bands in the surface of the optic disk drusen. (11) (Fig.8)

It was reported that similar hyper-reflective bands without a hypo-reflective core suggested small drusen, even though this has not been proven with corroborating data. (11)

This method helps to distinguish between buried optic disk drusen and optic disk oedema, although the description of how the two can be differentiated have varied. (11)

Nonetheless, the retinal nerve fiber layer thickness is most widely employed to identify optic nerve head fiber from disk oedema. (11)

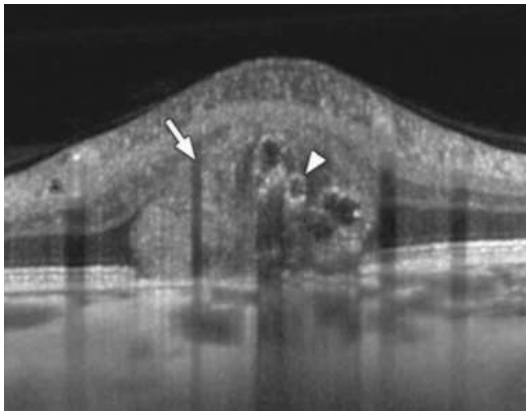

Fig. 8 : Enhanced depth imaging spectral domain optical coherence tomography image with evidence of confluent optic nerve head drusen (arrowed) with peripapillary subretinal optic disk drusen (arrow) (12)

7- New technologies :

Latest techniques are available. Tomography of the nerve head is proving to be helpful. GDx ® (scanning Laser Polarimetry) and Hedelberg Retinal Tomography (HRT) are actually accessible. (8)

These technologies are used to search for nerve fiber layer loss which could be observed in the optic disk drusen. (8)

F. Complications:

The sudden vision loss in the optic disk drusen is most frequently correlated to a nonarteritic anterior ischemic optic neuropathy (NAION). In opposition to the typical NAION patients, optic disk drusen patients are younger and have a better visual prognostic. (5)

Other uncommon vascular complications that have been described in the literature involve subretinal neovascularization, central retinal artery and vein occlusion, and choroidal neovascular membranes (5, 8)

The optic disk drusen may also cause hemorrhage in the retina and disc margin. Optic disk hemorrhages are specifically more frequent in children. (11)

G. Patient management and treatment :

Even though no beneficial treatment has been recognized, frequent examinations should be done so as to identify or rule out treatable concomitant disorders such as elevated intraocular pressure or subretinal neovascularization. (3)

Therefore, regular tonometry and visual field examination is crucial given that the predamaged optic nerve fibers are more sensitive to elevated or even normal intraocular pressure as opposed to undamaged nerve fibers. (3)

In cases with progressive visual field defects, topical ocular hypotensive therapy may be initiated (5), even though there has been no studies to appreciate the benefit of this therapy.

Surgical treatment for visual field defects with optic nerve sheath fenestration or radial optic neurotomy is controversial and not considered conventional, but success registered by a few authors. (5)

Beneficial treatment complications with choroidal neovascular membranes has been reported : surgery, laser photocoagulation, photodynamic therapy and most recently intravitreal anti-vascular endothelial growth factor agents.(5)

Conclusion:

In summary, buried optic nerve head drusen can be identified most accurately by B-scan ultrasonography which is considered the most sensitive technique in comparison with computed tomography, and therefore should be the method of choice when optic disk drusen is suspected as a non-invasive, affordable and easy exam.

Calcified optic disk drusen are more easily seen in computed tomography, but this method is essentially used to exclude an intracranial lesion, as well as for the MRI. Those two techniques are however not considered as a reliable technique for identifying the optic disk drusen.

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