

Clinical Picture and Diagnosis in a Patient with Optic Disc Drusen: A Case Report

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- ✓ Background: Drusen are deposits of a hyaline-like calcific material within the substance of the optic nerve head. Clinically, drusens are present in nearly 0.3% of the population and are frequently bilateral and familial. In children, drusen lie deep beneath the surface of the disc tissue and cannot be identified ophthalmoscopically with ease. In this setting they may be confused with papilloedema.

Case: A 9 year old male patient complained of headache and poor vision. The snellen visual acuity was 1.0 with correction. There was a slight elevation in the head of the optic discs and the borders were slightly blurred . Hyperdense images were found in optic nerve heads in B-scan ultrasonography (B-USG) and computerised tomography (CT). A sector defect was detected in the right eye using static perimetry.

Conclusion: This case shows that optic disc drusen should be considered in the differential diagnosis of papilloedema especially in children. The diagnosis should be confirmed by computerised tomography and ultrasonography. Especially B-scan ultrasonography is an easy method that could be used as diagnostic tool. It is non-invasive, inexpensive and comparable with other diagnostic tools.

Key words: Optic disc drusen, B-scan ultrasonography, computerised tomography

- ✓ **Optik Disk Druzeni Olan Bir Hastada Klinik Görünüm ve Tanı: Bir Vaka Takdimi**

Giriş: Druzen, optik disk başında hyalin benzeri madde birikimleridir. Toplumun yaklaşık %0.3'ünde görülürler. Çoğunlukla bilateral ve familyaldirler. Çocuklarda özellikle optik disk'te derinde yerleşimli druzenler oftalmoskopik olarak kolay ayırt edilemezler. Bu yerleşimleri ile kolaylıkla papilödem ile karıştırılabilirler.

Olgu sunumu: 9 yaşında erkek hasta baş ağrısı ve görme bulanıklığı şikayeti ile kliniğimize müracaat etti. Hipermetropisi olan hastanın görme keskinliği tashihle iki gözde de 10/10 olarak bulundu. Her iki gözde optik disk başında hafif kabarıklık ve sınırlarında siliklik vardı. B-scan ultrasonografide ve orbital tomografide optik disk başında hiperdens görünüm tespit edildi. Periferik görme alanı muayenesinde sağ gözde sektör şeklinde defekt vardı.

Sonuç: Bu vaka göstermektedir ki özellikle çocuklarda papilödemden ayırıcı tanısında optik disk druzeni akılda tutulmalıdır. Optik disk druzenin teşhisi B-scan ultrasonografi ve orbital tomografi ile doğrulanmalıdır. Özellikle ultrasonografi kolay uygulanabilir, non-invaziv, ucuz ve güvenilir olmasından dolayı bu hastalarda önerilebilir.

Anahtar kelimeler: Optik disk druzeni, B-scan ultrasonografi, komputeze tomografi

INTRODUCTION

Drusen are deposits of a hyaline-like calcific material within the substance of the optic nerve anterior to the lamina cribrosa.

Their incidence is 0.3% to 1% clinically, and 2% ophthalmoscopically. They are bilateral in 75% to 80% of cases. Histopathologically, drusen appear as

concentric hyaline-like laminations with calcification. They stain positively for amino acids, acid mucopolysaccharides, calcium and hemosiderin, and negatively for amyloid. The aetiology of drusen is unknown^(1,2). The occurrence of drusen may be inherited as an autosomal dominant characteristic with incomplete penetrance. Superficial drusen appear as irregular, glistening, yellow globules that may be isolated or clustered. Deep drusen are not directly visible, thus causing pseudo-papilledema⁽³⁾. In children, drusen lie deep beneath the surface of the optic disc tissue and cannot be identified ophthalmoscopically with ease. In this setting they may be unnecessarily investigated for the possibility of a brain tumour. Buried drusen often become visible with advancing age⁽¹⁻⁴⁾. Optic disc drusens can be associated with progressive optic

atrophy, papillary haemorrhage and vascular shunt, central retinal vein occlusion, high hyperopia^(2,4-8).

We describe a patient with bilateral optic disc drusen and diagnosis was confirmed by radiographic methods in this study.

CASE REPORT

A 9 year old male patient complaining of headache and poor vision applied to our clinic. On ophthalmological examination the visual acuity was 10/10 with optical correction. He had hyperopia (Right eye: +1.0 sph. +0.50 cyl 90°. Left eye: +1.0 sph +0.75 cyl 90°). Slit-lamp examination and intraocular pressures were normal. There was a slight elevation in the head of the bilateral optic disc and the border was not so clear. Similar images were detected in both optic discs. The right eye fundus image is shown in Figure 1. B-scan ultrasonography

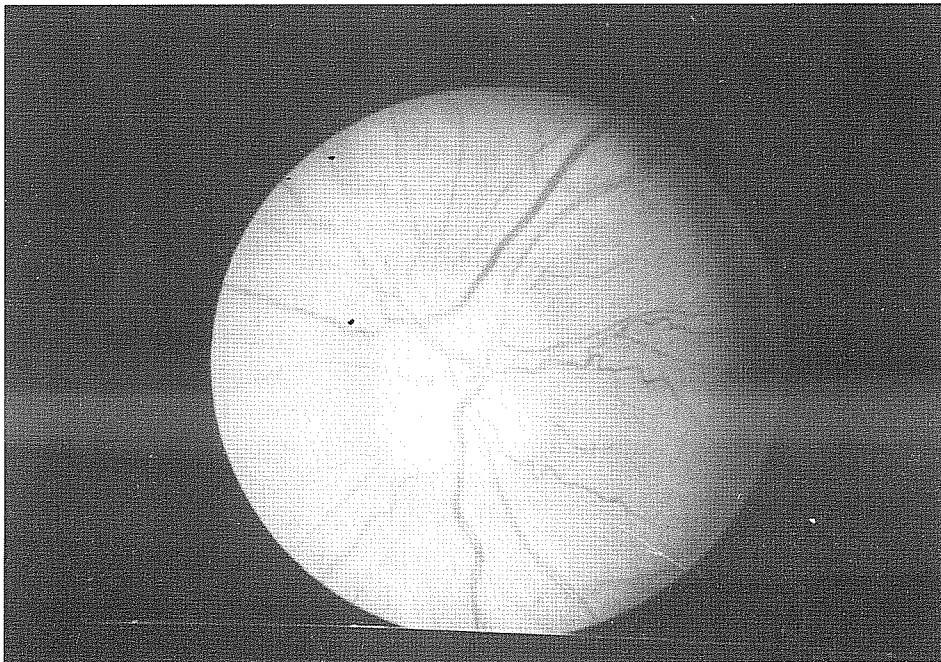


Figure 1. The blurred image of the disc in the right eye. There is some tortuosity of the vessels and the entire disc area is swollen.

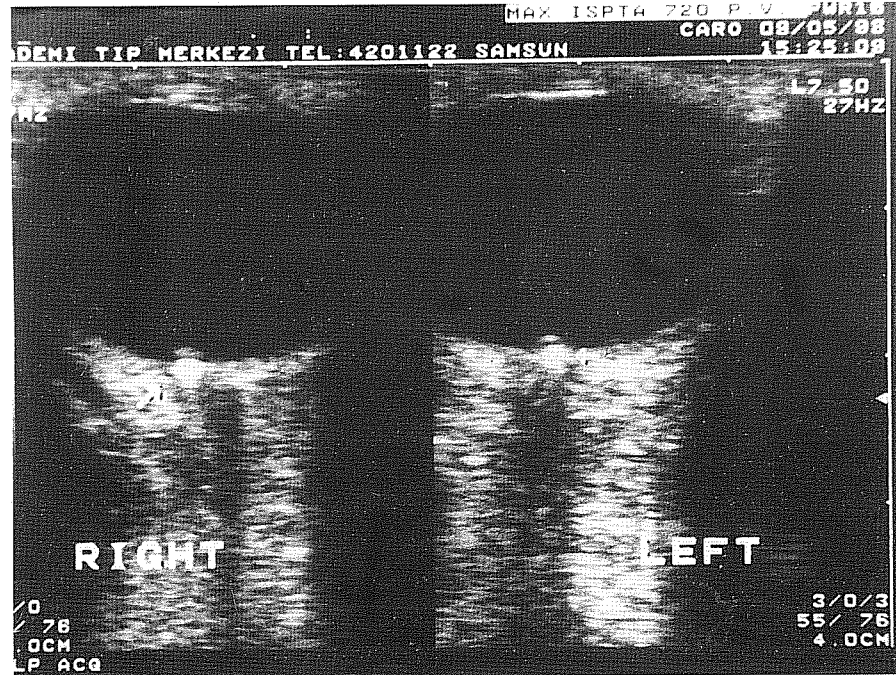


Figure 2. B-Scan ultrasonography. Hyperdense image and smooth elevation are evident in both optic discs.

revealed a 0.3 mm smooth elevation and calcific appearance in the optic nerve head (Figure-2). The computerised tomography showed a hyperdense image at the level of optic disc in both eyes (Figure-3). The visual fields showed a sector defect in the right eye. In the FFA, fluorescein leakage was not seen in the optic disc, but staining was observed in the optic disc in the late period. Extensive systemic evaluations, including complete blood count, platelet count, bleeding time, clotting factors were unremarkable. Ophthalmological examination was normal in his parents and brother.

DISCUSSION

Drusen is generally asymptomatic. Although the vast majority of eyes with drusen remain asymptomatic during the patient's lifetime, drusen can on occasion cause visual impairment⁽⁹⁻¹¹⁾. Visual loss may

occur from compression of nerve fibers, subretinal haemorrhage, or subretinal neovascularization⁽³⁾. Decreased visual acuity due to drusen in only one eye in 40 cases was reported by Hoover et al.⁽¹¹⁾. Of 307 eyes with optic disc drusen examined by Mustonen, decrease in visual acuity was detected in only four (1.3%)⁽¹²⁾. Loss of visual acuity from optic disc drusen is so rare that patients with drusen and visual acuity loss should be evaluated for a possible compressive lesion. Although visual acuity is rarely directly affected by superficial drusen of the optic disc, visual field defects related to the drusen are common and may be significant^(3,13,14). In several studies, about 75% of patients with drusen of the optic disc have been found to have associated visual field defects^(3,11,14,15). Visual field defects in patients with buried drusen are much less common and less severe than those present

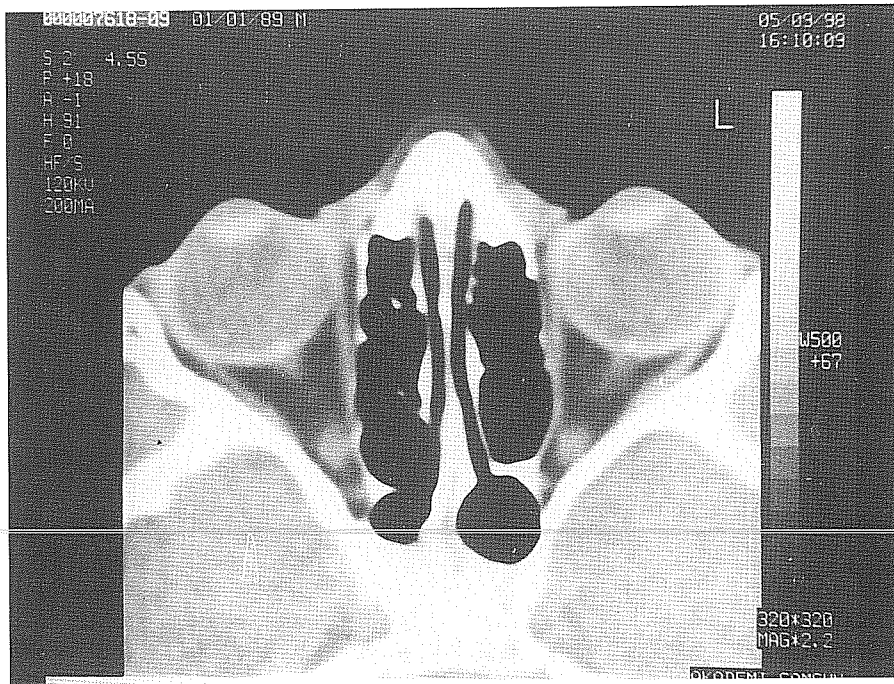


Figure 3. Computerised tomographic scan of the skull showing calcific appearance at the level of both optic discs.

in patients with superficial drusen. Visual field defects include enlargement of the blind spot, nerve fiber bundle defect and occasionally, irregular constriction of the peripheral field. Severe visual field loss is possible^(3,6,14). Auw-Haedrich et al.⁽¹⁶⁾ reported a case who has optic disc drusen with visual field defect. In our case, although visual field defect was present in both eyes, the visual acuity was normal.

Drusen of the optic disc are usually easily identified during an ophthalmoscopic examination; however, in some patients especially in children, they may be buried deep within nerve tissue. Such drusen become more visible with time. In the setting they may be confused with early papilloedema⁽¹⁾. In contrast to papilloedema, optic disc drusen includes the followings: optic cup is absent, spontaneous venous pulsation is present in 80% of cases, the disc

has a pink or yellow colour, the peripapillary nerve fibre striations are not obscured, fluorescein does not leak from disc vessels, and vessels arise from central apex of disc⁽¹⁻³⁾. Nevertheless, special techniques may need to be used to identify buried optic disc drusen. The patients with elevated optic discs that are thought to be anomalous but in which no drusen can be observed, both ultrasonography and computed tomographic scanning may be extremely useful in identifying buried drusen⁽¹⁷⁻²⁴⁾. Computed tomographic scanning is recommended in cases of drusen with deep localization or in cases associated with other pathologies of the posterior pole. Computerized tomography shows hyperdense image at the level of optic disc^(18,19,22,24). One of the B-scan ultrasonographic characteristics of drusen of the optic disc are the presence of strong reflections at the low energy levels^(17,20-23).

Bec and al.⁽¹⁹⁾ reported that CT scan can show small and buried drusen, which are sometimes difficult to diagnose by the ophthalmoscopic examination. Noel et al reported that drusen of the optic disc B-scan ultrasonography with a lowered sensitivity was found to be useful in the uncooperative child⁽²²⁾. FFA can be useful at the diagnosis of the optic disc drusen^(22,23).

The case we presented here, shows typical aspects of optic disc drusen detected by CT and B-scan ultrasonography. We conclude that B-USG, is a non-invasive and inexpensive technique, that can be used as an imaging method of choice in identifying the presence of disc drusen.

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