Norcia:

“...”

Beres:

“A fallacious similarity between the elevation of the optic disc caused by buried drusen and optic disc edema caused by increased intracranial pressure presents a problem of differential diagnosis, expressly in younger children”

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A New Autosomal Recessive Syndrome Consisting of Posterior Microphthalmos, Retinitis Pigmentosa, Foveoschisis, and Optic Disc Drusen Is Caused by a MFRP Gene Mutation

Raul Ayala-Ramirez, Federico Graue-Wiechers, Violeta Robredo, Monica Amato-Almanza, Illiana Horta-Diez, Juan Carlos Zenteno

Affiliations + expand
PMID: 17167404
Ocular Ultrasound in Alagille Syndrome
A New Sign

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Alan C. Bird, MD, FRCS, FRCOphth,1 Alastair J. Baker, MRCP,4
Alex P. Mouat, FRCP,4 Georgina Mardi-Vergani, MD, PhD, FRCP,4
Wagih A. Aclimandos, FRCS, FRCOphth4

Background: Alagille syndrome (AS) is one of six forms of familial intrahepatic cholestasis, all of which present with neonatal jaundice and paucity of intrahepatic bile ducts. Differentiation of these individual syndromes is crucial as their treatments and prognoses vary. It is the ophthalmic features, posterior embryotoxon in particular, that distinguish AS.

Methods: The authors performed full ocular examination, including A- and B-scan ultrasound, retinal, and, where possible, fluorescein angiography in 20 unrelated children with AS and 8 with non-AS-related cholestasis.

Results: There was ultrasound evidence of optic disc drusen in at least one eye in 95% and bilateral disc drusen in 80% of patients with AS but in none of the patients who were non-AS at the time of examination. Independent review of hard-copy scans suggested drusen in at least one eye in 90% of the cases and bilateral drusen in 50%, although this latter figure rose to 65% on review of the angiograms. This is markedly higher than the incidence in the normal population (0.3%–2%). Axial lengths were shorter than expected for the older age group (older than 10 years of age), but this was not associated with gross ametropia.

Conclusion: This strong association of AS and optic disc drusen has not been reported previously and represents not only the first significant association between a systemic condition and disc drusen but also a possibly useful tool in the diagnosis of AS, especially in young children. Ophthalmology 1997;104:79–85
A study of optic nerve head drusen in 38 pseudoxanthoma elasticum (PXE) patients (64 eyes). Location of optic nerve head drusen in PXE

Étude des drusen du nerf optique dans le pseudoxanthome élastique chez 38 patients (PXE). Localisation des drusen du nerf optique dans le PXE

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Keywords
Pseudoxanthoma elasticum; Calcification; R Motor neuron; Nerve plexus; epithelium atrophy; Optic nerve head drusen

Summary
Purpose. — To investigate the prevalence and location of optic nerve head drusen and their potential association with other PXE-related ophthalmic abnormalities.

Methods. — Thirty-eight of the 55 patients (37 male and 8 female aged 60 ± 17 years) included in this retrospective study had optic nerve head drusen. All of the patients underwent a comprehensive ophthalmic examination, including color images using red-free, blue and red filters, autofluorescence imaging and late-gain ICG frames. Comparative analysis of both groups (optic nerve head drusen vs. no drusen) was conducted using statistical software.

Results. — The prevalence of optic nerve head drusen in our cohort was 67.3%. In this study, the existence of a significant link between optic nerve head drusen and other fundus abnormalities was detected. They were more frequently located in the macular sector than in the temporal sector of the optic disc. The prevalence of optic disc drusen was 9.7%. The mean visual field defect was observed in 26% of patients who were uncorrected for macular disorders.

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Echographic Diagnosis of Drusen of the Optic Nerve Head in Patients with Angioid Streaks

Key Words
Angioid streaks
Drusen of the optic nerve head
Pseudoxanthoma elasticum
Echography
Fluorescein angiography

Abstract
A total of 58 patients (116 eyes) with angioid streaks, referred to this department over 5 years (1990-1995), underwent a thorough ophthalmic examination, retinal fluorescein angiography, dermatological visit with skin biopsy, and a series of other examinations (blood and biochemical tests, skull X-ray, sonar, echography of the hip, abdominal ultrasonography) and eye ultrasonography. Fifty patients (100 eyes, 86.2%) had pseudoxanthoma elasticum (PXE). In the whole material, 24.1% of patients had angioid streaks compared with 21.0% in the group with PXE. Both these findings are higher than those published elsewhere. Drusen of the optic nerve head were also found in 25% of the 100 eyes of patients with angioid streaks but no PXE. We believe we found a higher prevalence of drusen of the optic nerve head in these patients with angioid streaks because we systematically employed ultrasound.

Introduction
The association of drusen of the optic nerve head (ONH) and angioid streaks has only been described in patients with pseudoxanthoma elasticum (PXE) [1-3]. More than 90% of these patients present streaks [4-5], but the prevalence of drusen is reported as ranging from only 4% [6] to 60% [7]. However, angioid streaks with drusen have recently been described in a patient with Weissenbacher-Vivanti heredity [8], and it is conceivable that the two clinical signs may occur together in other diseases too [9].

To our knowledge, echography has never been used systematically to check for drusen of the ONH in patients with angioid streaks, although we believe this diagnostic technique lends itself better to identifying small drusen. In the present study ocular echography was employed to check for the prevalence of ONH drusen together with angioid streaks.

Patients and Methods
Fifty-eight patients (116 eyes) with angioid streaks, who were referred to our department over 5 years (1990-1995), underwent a thorough ophthalmic examination, retinal fluorescein angiography, dermatological visit with skin biopsy, a series of other examinations (blood and biochemical tests, skull X-ray, sonar, echography of the hip, abdominal ultrasonography) and eye ultrasonography. Fifty patients (100 eyes, 86.2%) had PXE. The mean age was 33 years (range 16-74, SD 14 years). All examinations of the ONH were checked before fluorescein angiography, using the echographic scanning filter from Anima. An Optimark (Optronics Medical) echography was employed in thebilt scan mode. A skin biopsy for histological testing (immunohistochemistry and electron microscopy) was taken from the angioid streak area, whereas there were no further alterations attributable to PXE. The person responsible for the echographic test is the author of the present study.
Frequency of Optic Disc or Parapapillary Nerve Fiber Layer Drusen in Retinitis Pigmentosa

Sandee Grover, MD, Gerald A. Fishman, MD, Jeremiah Brown, Jr., MD

**Purpose:** To determine the frequency and characteristics of optic disc and parapapillary nerve fiber layer drusen in patients with retinitis pigmentosa and to attempt to document any differences in the frequency within different genetic subtypes of retinitis pigmentosa.

**Methods:** This retrospective case series reviewed 117 patients with autosomal-dominant, 84 with autosomal-recessive, and 61 with X-linked recessive forms of retinitis pigmentosa. Color fundus photographs were reviewed independently by three investigators. The presence of optic disc or parapapillary nerve fiber layer drusen was documented only when all three observers concurred as to their presence. The number (isolated or multiple), site (disc, parapapillary, or both), and bilaterality (unilateral or bilateral) were noted in each patient.

**Results:** Optic disc or parapapillary nerve fiber layer drusen or both were observed in 11 patients (9.4%) with autosomal-dominant, 8 patients (7.1%) with autosomal-recessive, and 7 patients (11.5%) with X-linked recessive types of retinitis pigmentosa. The differences in the observed frequencies were not statistically significant ($P = 0.67$). Overall, the frequency of optic nerve head or parapapillary drusen or both in 262 patients with retinitis pigmentosa was 9.2%.

**Conclusion:** In a large population of patients with retinitis pigmentosa, our findings suggest that approximately 10% are likely to show optic nerve fiber layer drusen involving the optic disc or parapapillary regions or both. The frequency does not vary significantly between different genetic subtypes. Ophthalmology 1997; 104:280–289

- Müller (1858) The first to give a histologic description of ODD found together with RP in both eyes of an almost blind man, aged 75
- Nieden (1878) First ophthalmological description of ODD seen in a 14-year-old girl with RP
- Ancke (1885) Described ODD in 3 members of a sibship of 5 and in 2 members of a sibship of 5. The ODD were in all 5 cases associated with RP

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From the University Eye Clinic, Aarhus
(Chief: Professor Viggo A. Jensen, M.D.)

**DRUSEN OF THE OPTIC DISK.**
AN IRREGULARLY DOMINANT HEREDITARY AFFECTION

BY
S. E. Lorentzen

- Braun (1935): 2 families, 3 relatives affected out of 22 examined in one family and none affected in 26 relatives in the other family
Leimgruber (1936): 8 families, 4 families with 13 relatives affected out of 72 examined, although in the other 4 families no relatives were affected

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Müller’s drawing attending the first histologic description of optic disc drusen (1858)
De Wecker & Jäger’s (1870) drawing of optic disc drusen after Iwanoff’s histological specimen
Stood’s (1883) illustration of the ophthalmoscopic image of optic disc drusen
de Schweinitz’ (1892) photomicrograph of optic disc drusen (x about 200)
Drusen of the optic disc—
A histopathological study

A. H. FRIEDMAN, P. HENKIND, AND S. GARTNER
From the Department of Ophthalmology, Albert Einstein College of Medicine/Montefiore Hospital and Medical Center, New York.
Pathology and Pathogenesis of Drusen of the Optic Nervehead

MARK O. M. TSO, MD

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Young Adults with Anterior Ischemic Optic Neuropathy: A Multicenter Optic Disc Drusen Study


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Mahajan:


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