Available online at www.sciencedirect.com
ScienceDirect
journal homepage: www.elsevier.com/locate/survophthal

Major review

# Optic disk drusen in children

*Melinda Y. Chang, MD*[a,b], *Stacy L. Pineles, MD*[a,b,*]

[a] *Department of Ophthalmology, Stein Eye Institute, Los Angeles, California, USA*
[b] *University of California, Stein Eye Institute, Los Angeles, California, USA*

ARTICLE INFO

*Article history:*
Received 15 July 2015
Received in revised form 18 March 2016
Accepted 21 March 2016
Available online xxx

*Keywords:*
optic disk drusen
children
pediatric
pseudopapilledema
choroidal neovascular membrane

ABSTRACT

Optic disk drusen occur in 0.4% of children and consist of acellular intracellular and extracellular deposits that often become calcified over time. They are typically buried early in life and generally become superficial, and therefore visible, later in childhood, at the average age of 12 years. Their main clinical significance lies in the ability of optic disk drusen, particularly when buried, to simulate true optic disk edema. Misdiagnosing drusen as true disk edema may lead to an invasive and unnecessary workup for elevated intracranial pressure. Ancillary testing, including ultrasonography, fluorescein angiography, fundus autofluorescence, and optical coherence tomography, may aid in the correct diagnosis of optic disk drusen. Complications of optic disk drusen in children include visual field defects, hemorrhages, choroidal neovascular membrane, nonarteritic anterior ischemic optic neuropathy, and retinal vascular occlusions. Treatment options for these complications include ocular hypotensive agents for visual field defects and intravitreal anti–vascular endothelial growth factor agents for choroidal neovascular membranes. In most cases, however, children with optic disk drusen can be managed by observation with serial examinations and visual field testing once true optic disk edema has been excluded.


## 1. Introduction

Optic disk drusen are acellular deposits located both intracellularly and extracellularly first described by Müller in 1858.[130] The main clinical significance of optic disk drusen in children is that they can simulate true optic disk edema (Fig. 1).[52,81,127,189,213] Misdiagnosing drusen as true disk edema may lead to an extensive, invasive, and unnecessary workup for elevated intracranial pressure, including neuroimaging and lumbar puncture.[115] Optic disk drusen are typically buried in the optic disk early in life and become more superficial later.[7,57,80,197] In children, therefore, drusen are more likely to be buried and may be more difficult to detect.[43]

### 1.1. *Pathogenesis*

The pathogenesis of optic disk drusen is unknown. The 3 classical theories on the formation of optic disk drusen postulate that they are caused by a disturbance in axonal metabolism with slowed axoplasmic flow[196,204]; congenitally dysplastic disks with a propensity for drusen formation[139,174]; or a small scleral canal that physically compresses the optic nerve, causing ganglion cell death, with extrusion and calcification of mitochondria.[132] The latter theory has been called into question by a study that showed that the scleral canal in patients with optic disk drusen was not smaller than controls when measured by optical coherence tomography (OCT).[51]

* Corresponding author: Stacy L. Pineles, MD, Department of Ophthalmology, Stein Eye Institute, University of California, 100 Stein Plaza, Los Angeles, CA, USA.
E-mail address: Pineles@jsei.ucla.edu (S.L. Pineles).
0039-6257/$ – see front matter 
http://dx.doi.org/10.1016/j.survophthal.2016.03.007

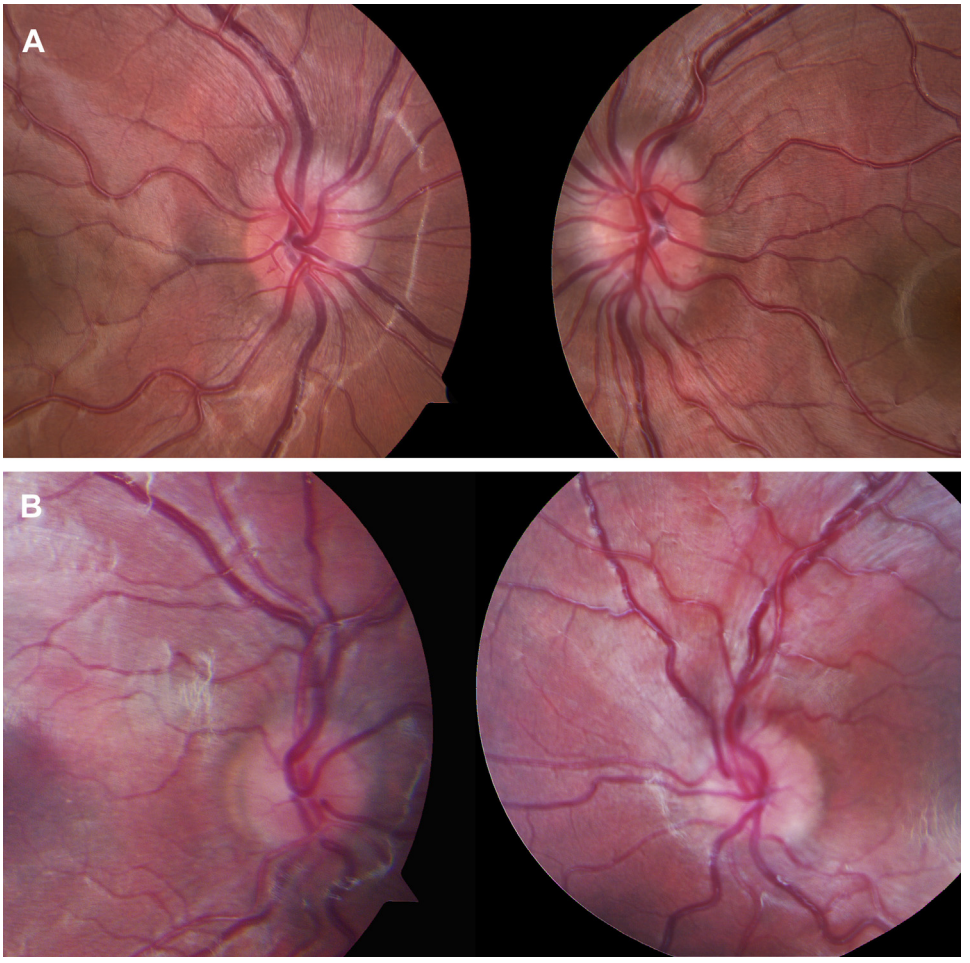


**Fig. 1 – Comparison of optic disk in children with optic disk drusen and papilledema. *A:* Optic disk photos of a 10-year-old boy with bilateral buried optic disk drusen. The disk margins are blurred, but there are no hemorrhages, exudates, or vessel obscuration. *B:* Optic disk photos of a 5-year-old girl with mild papilledema due to increased intracranial pressure secondary to the use of exogenous growth hormone. Disk margins are blurred with mild obscuration of vessels, but no hemorrhages or exudates.**

### 1.2. *Demographics*

The prevalence of optic disk drusen in children is about 0.4%.[45] In adults, studies have found a prevalence of 0.5%–2.4%.[7,56] The lower prevalence of optic disk drusen reported in children is likely due to the difficulty in detecting buried drusen. In children and adults, optic disk drusen are more common in women and whites and are bilateral in over two-thirds of cases.[7,19,97,171,203]

### 1.3. *Inheritance*

Optic disk drusen are frequently familial. Family members of patients with optic disk drusen have up to 10 times the risk of harboring optic disk drusen compared to the general population, and they have an increased risk of optic disk dysplasia and anomalous retinal vasculature.[4,119] Optic disk drusen can also be inherited as part of a genetic syndrome with other ocular or systemic manifestations.

## 2. Association of optic disk drusen with other ocular or systemic disorders

Optic disk drusen have been reported in association with many ocular (Table 1) and systemic (Table 2) disorders; however, there are only a few disorders in which optic disk drusen have been demonstrated to occur more frequently than in the general population.

### 2.1. *Retinitis pigmentosa*

The association of retinitis pigmentosa (RP) with optic disk drusen has been known since the first case of optic disk drusen was published by Müller in 1858.[130] The frequency of optic disk drusen in children with RP is not known. The largest study of optic disk drusen in patients with RP included both adults and children and found that 9.2% of patients with RP had optic disk drusen. The frequency of optic disk drusen in children was not reported separately, however, and because