Congenital Choroidal Nevus in a Two-Month-Old Infant

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Received: May 26, 2019; Published: June 14, 2019

Abstract

We report the case of a 2-month-old male infant who was brought in for a routine visual examination. Fundus examination revealed generalized hypopigmentation of the fundus in both eyes and a small rounded pigmented choroidal lesion in his left eye of approximately two disc diameters near the temporal inferior vascular arcade. Ultrasonography showed a small slightly elevated lesion with high reflectivity. Based on the clinical findings, a diagnosis of congenital choroidal nevus was made and periodic follow-up was proposed. Choroidal nevi are uncommon before puberty; to our knowledge, there are no previous reports confirming that they may be present congenitally.

Keywords: Choroidal Nevus; Choroidal Nevi; Tumor; Congenital; Ultrasonography

Abbreviations

BMES: Blue Mountains Eye Study; NHANES: National Health and Nutrition Examination Survey; RPE: Retinal Pigment Epithelium; CNV: Choroidal Neovascularization; OCT: Optical Coherence Tomography; VEGF: Vascular Endothelial Growth Factor

Introduction

Choroidal nevus is the most common benign intraocular tumor occurring in about 5% of the adult population and predominantly in whites [1]. These lesions are common incidental findings in the ophthalmological examination and despite being benign tumors, they can produce symptoms like visual loss, visual fields defects and risk of transformation into melanoma [1].

These tumors are rare before puberty and there are no previous reports confirming that they may be present congenitally.

We present a case of a 2-month-old infant with clinical and ultrasonographic findings typical of choroidal nevus.

Case Report

A 2-month-old male infant presented at eye clinic for a routine visual examination. He was born at term by elective cesarean section due to cephalopelvic disproportion, his birth weight was 3100 g, no history of ocular or systemic disease.

He was able to follow objects with both eyes. Cycloplegic refractions were +4.50 and +3.50 diopters for right and left eye respectively. Eye movements were complete and equal in both eyes and examination of the anterior segment revealed hyperpigmented areas of the iris in both eyes (Figure 1).
Fundus examination by indirect ophthalmoscopy revealed generalized hypopigmentation of the fundus, with clear visibility of choroidal vessels in both eyes and a small rounded pigmented choroidal lesion in his left eye of approximately two disc diameters near the temporal inferior vascular arcade (Figure 2). An A and B scans ultrasonography was performed and revealed a small slightly elevated lesion with high reflectivity measuring 1mm in basal diameter and 1 mm in thickness (Figure 3).

**Figure 2:** Mobile phone indirect ophthalmoscopic fundus image (A) and composite photograph (B) of the left eye showing diffuse hypopigmentation, with clear visibility of choroidal vessels and a small rounded pigmented choroidal lesion of approximately two disc diameters near the temporal inferior vascular arcade (arrows). (Figure 2A: Courtesy of Dr. Hugo Valencia, Figure 2B: edited by MAILOR - Mexican Advanced Imaging Laboratory For Ocular Research).
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Based on the clinical findings, a diagnosis of choroidal nevus was made, images were obtained and periodic ultrasonographic and photographic follow-up was proposed.

Discussion

Shields., et al. reviewed 3422 cases of choroidal nevi and found that the mean age of diagnosis was 60 years (ranging from 1 to 97 years old); the majority of patients were over 50 years of age (74.9%), followed by those between 21 and 50 years old (23.2%), with only 1.8% of cases under 20 years of age [2]. Ma., et al. carried out a universal ocular screening of 481 6-weeks-age infants using wide-field digital imaging system and found 1 case of choroidal nevus [3].

The prevalence of choroidal nevus varies according the race, the type of study and the diagnostic method. Several criteria used in many studies results in different outcomes. The Blue Mountains Eye Study (BMES) found a prevalence of 6.5% in US white adults over 50 years [4] and it was 0.3% in an Indian based population [5]. The National Health and Nutrition Examination Survey (NHANES), which included all ethnic groups of adults over 40 years of age, found a prevalence of 4.7% (being higher in whites); in this study, two 45° ocular fundus photographs were examined, and the authors speculated a real prevalence of 20-25% if the entire retina was examined [1]. Clinically, choroidal nevus can be flat or minimally elevated with (melanotic, 77%) or without (amelanotic, 10%) pigmentation. It is most often located in the postequatorial region (91%) compared with the preequatorial area (9%) [6].

They are usually asymptomatic since most of the choroidal nevi are extrafoveal (94%) and only 6% are subfoveal [2]. Symptoms include loss of visual acuity, visual field defects and presence of floaters and flashes. It was estimated that the risk for vision loss at 15 years was 2% for the extrafoveal nevi and 26% for the subfoveal group [2,7].

The diameter of the lesion is usually between 1.25 [4] and 5.2 mm [2]; and the thickness is 2 mm on average. As the age increases, the average thickness and the probability of presenting multiple nevi per eye increases too. Overlying drusen also develop with time [2]. Detachment of the retinal pigment epithelium (RPE) and choroidal neovascularization (CNV) of a nevus are uncommon [2].

Choroidal nevi has the potential for transformation into melanoma, and the annual rate of malignant transformation was estimated to be 1 in 8845 [8]. Statistical risk factors for malignancy include tumor thickness of >2 mm at initial diagnosis, overlying orange pigment, drusen, and any changes in the size or shape of the lesion over time [2].

Figure 3: A) A-scan ultrasonography of the left eye showing a lesion with high reflectivity measuring 1mm in thickness (arrowhead). B) B-scan ultrasonography shows a slightly elevated small lesion (between arrowheads) with a basal diameter of 1 mm and absence of posterior acoustic shadow and calcifications. (Courtesy of Dr. Daniel Salas).

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subretinal fluid, acoustic hollowness, absence of halo, distance of the margin ≤3 mm from the optic disc, and the presence of symptoms [9]. Tumor growth into melanoma in 5 years was documented in more than 50% of patients with 3 or more risk factors [9].

Ultrasoundography can be used for baseline thickness measurements for elevated lesions (lesion with < 2 mm are considered a low risk choroidal nevus) [10]; likewise, the nevus is characterized by having a high internal reflectivity, unlike the melanoma that has a medium-low internal reflectivity [6]. Optical coherence tomography (OCT) can detect subtle subretinal fluid, cystoid retinal edema, and RPE alterations, but is not of value in diagnosis of the nevus itself [10].

Choroidal nevus typically requires no treatment. Baseline fundus photography and ultrasonography should be performed and the patient should be examined every 6 to 12 months to detect growth of the lesion [10]. If one or two risk factors are present follow up should be shortened. In cases with symptomatic secondary subretinal fluid or CNV, specific methods of laser photocoagulation, transpupillary thermotherapy, anti-vascular endothelial growth factor (anti-VEGF) therapy or photodynamic therapy have been used to achieve fluid resolution [6].

Conclusion

This case of a 2-month-old boy with a typical choroidal nevus, added to the case shown by Ma., et al. [3], confirms that this pathology can occur congenitally in exceptional cases and that visual screening in babies is important to rule out pathologies with potential damage to vision.

Conflict of Interest

There is no financial interest or any conflict of interest.

Bibliography


Volume 10 Issue 7 July 2019
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