Focal Choroidal Excavation at Pediatric Age

Jorge Vasco Costa[1]; Marina João[1]; Keissy Sousa[1]; Gil Calvão-Santos[1]; Luís Mendonça[1]; Nuno Lourenço Gomes[1]
[1] Ophthalmology Department, Hospital de Braga, Braga, Portugal

ABSTRACT

Purpose: To present a case of unilateral focal choroidal excavation (FCE) in a 4-year-old patient.

Methods: Case report

Results: A 4-year-old patient presented with diminished visual acuity of the left eye with no other concomitant symptoms. Fundoscopic examination revealed a whitish-yellow lesion in the fovea, and spectral domain optical coherence tomography (SD-OCT) revealed a local choroidal excavation with depression of the retinal structures. No subretinal fluid was observed. Extensive blood and serum analysis were performed, with no positive results. The diagnosis of unilateral FCE was made.

Conclusion: FCE is a local idiopathic choroidopathy, usually diagnosed through SD-OCT. Its etiology is unknown. Some authors hypothesize that it is a congenital malformation, while other see it as a secondary manifestations of other conditions. To our knowledge, this is the first report of this disease in a patient this young, and we theorize that this condition presents as both a primary defect and secondary to other chorioretinal diseases.

Keywords: Focal choroidal excavation; macular dystrophy; spectral domain optical coherence tomography; chorioretina; pediatrics

RESUMO

Objetivos: Apresentar um caso clínico de uma escavação coroideia focal (ECF) numa cirança de 4 anos.

Materiais e métodos: Caso clínico

Resultados: Uma criança de 4 anos apresenta-se com perda da acuidade visual do olho esquerdo sem outros sintomas concomitantes. A fundoscopia revelou uma lesão amarela-esbranquiçada na fóvea e a tomografia de coerência óptica de domínio espectral (SD-OCT) mostrou uma escavação coroideia local com depressão das camadas retinianas. Não foi identificado líquido subretiniano. Foi realizado um extenso estudo analítico e imagiológico, sem resultados positivos. Foi realizado o diagnóstico de ECF unilateral.

Conclusões: A ECF é uma coroidopatia idiopática local, habitualmente diagnosticada através de SD-OCT. A sua etiologia é desconhecida. Alguns autores admitem a hipótese de ser uma malformação congénita, enquanto outros a veem como uma manifestação secundária a outras patologias. De acordo com o nosso conhecimento, este é o primeiro caso clínico reportado desta patologia idiopática numa criança tão jovem, pelo que especulamos que esta entidade se poderá apresentar de forma primária ou secundária a outras doenças corioretiniana

Palavras Chave: Excavação coroideia focal; distrofia macular; tomografia óptica de domínio espectral; coriorretina; pediátrica

INTRODUCTION

Focal choroidal excavation (FCE) is a rare ocular finding characterized by a submacular excavation of the choroid detected on optical coherence tomography (OCT), usually in asymptomatic patients.[1] It is divided in two types, conforming FCE [no separation between the photoreceptor tips and the retinal pigment epithelium (EPR)] and nonconforming FCE (separation between the two structures). The former has better prognosis.[2] Its etiology is still unknown and it has been subject of great debate. Initially, some authors considered it to be a congenital posterior segment malformation.[2] However, there has been an increasingly report of cases associating FCE to macular diseases,[4-6] inflammatory diseases,[7,8] and to the spectrum of the pachychoroid manifestations.[9]

CASE REPORT

A 4-year-old child is referred to an Ophthalmology appointment following a diminished visual acuity of the left eye (OS) detected during a screening test in school. She was a premature child of thirty-two weeks; however, she had no other past relevant ocular or medical history. There was no family history of ocular or hereditary diseases.

On ophthalmic examination, the best corrected visual was 20/25 in the right eye and 20/50 in the OS. The biomicroscopy was normal. The fundoscopy revealed a whitish-yellow lesion in the fovea (Figure 1). Spectral-domain OCT (SD-OCT) was performed and revealed a local choroidal excavation with retinal, retinal pigment epithelium (RPE) and Bruch membrane depression (Figure 2). A complete bloodwork as well as multiple serologies related to infectious agents were executed, with no relevant results. The diagnosis of conforming FCE was made. No other retinal conditions such as choroidal neovascularization (CNV) or subretinal fluid were found.

Two years after the diagnosis, there was no clinical or imagiologic progression of the lesion.

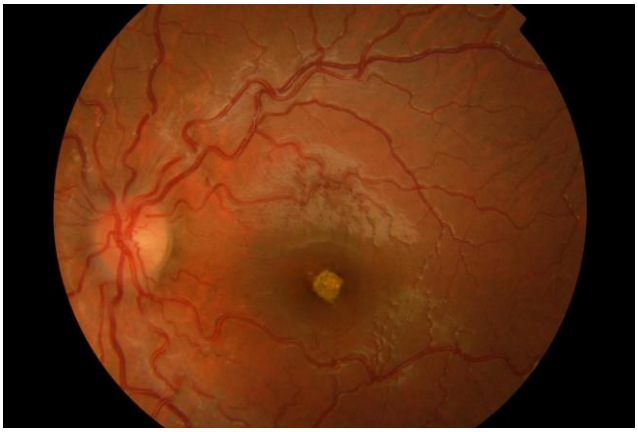

Figure 1 – Color retinography of the left eye on presentation revealing a whitish-yellow lesion in the fovea

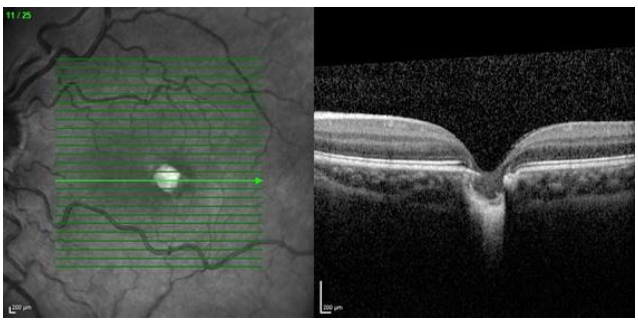

Figure 2 – Spectral-domain optical coherence tomography scan through the fovea reveals a conforming focal choroidal excavation

DISCUSSION

FCE is a somewhat recent clinical entity that has been the subject of investigation because of its unclear etiology. It's diagnosed through OCT and is characterized by one or more focal areas of choroidal depression without staphyloma or scleral ectasias or malformations. Most cases are diagnosed in their fourth to fifth decades and no gender predilection has been found. Most patients have moderate myopia. Fundoscopy usually appears normal or show non-specific pigmentary alterations. However, it can present as a yellow-whitish single or multiple lesions.[9] Considering only the fundoscopy (and retinography), the lesion described could also be associated with a torpedo maculopathy. However, this rare, benign entity presents typically as a hypopigmented oval lesion temporal to the fovea, elongated in horizontal axis, and with its tip pointing to the fovea. Adding to that, the OCT usually show an attenuation of the outer retinal structures with or without outer retinal cavitation always temporal to the fovea.[10]

Margolis et al,[2] the first to establish the definition of FCE, theorized that the this disease could be a congenital malformation that could progress in time, following phenomena of ischemia to the retina with subsequent atrophic changes and visual alterations. Since it's etiology remains a mystery, there have been various recent case reports and studies that have reported and established an association with other conditions, including central serous chorioretinopathy, CNV, polypoidal choroidal vasculopathy and various macular dystrophies, which led to the conclusion that FCE might be a secondary ocular change.[4-6] The multiplicity of published works also indicates that this is a more common entity that initially believed.[6-8]

To our knowledge this is the first time that FCE is reported in a patient this young and, associated with the lack of history of any ocular disease, we hypothesized and believe that FCE can, in fact, be a primary ophthalmic defect due to an embryonic development disorder because of a failure of the choroid fissure to fuse or an erroneous differentiation of the chorioretina. However the unusual fundus findings, with some characteristics of a torpedo maculopathy could also mean that this FCE can be secondary to this persistent defect in the development of the RPE. Our case report seems to reinforce the idea that the FCE is not a purely primary or secondary finding and that its etiology might have different explanations according to the presence of other concomitant ocular pathologies. It may have a congenital origin, where a small embryonic defect of the choroid may lead to outward migration of the retina. On the other hand, it can be acquired, maybe due to mechanical changes and, ultimately, imbalance of the RPE/Bruch membrane complex caused by other retinal diseases. Either way, periodic monitoring of all cases is of importance to detect and timely treat any of the known complications associated with FCE.

REFERENCES

1. Castro Navarro V, Montero Hernández J, Navarro Palop C, et al. Focal choroidal excavation: Clinical findings and complications. Arch Soc Esp Oftalmol. 2016; 91(1):34-9.
2. Margolis R, Mukkamala SK, Jampol LM, et al. The expanded spectrum of focal choroidal excavation. Arch Ophthalmol. 2011; 129(10):1320-5.
3. Lee CS, Woo SJ, Kim YK, et al. Clinical and Spectral-Domain Optical Coherence Tomography Findings in Patients with Focal Choroidal Excavation. Ophthalmology. 2014; 121(5):1029-35.
4. Chung CY, Li SH, Li KKW. Focal choroidal excavation-morphological features and clinical correlation. Eye (Lond). 2017; 31(9):1373-1379.
5. Liu GH, Lin B, Sun XQ, et al. Focal choroidal excavation: a preliminary interpretation based on clinic and review. Int J Ophthalmol. 2015; 18;8(3):513-21.
6. Battaglia Parodi M, Casalino G, Iacono P, et al. The expanding clinical spectrum of choroidal excavation in macular dystrophies. Retina. 2018; 38(10):2030-2034.
7. Matsubara H, Uchiyama E, Suzuki K, et al. A Case of Focal Choroidal Excavation Development Associated with Multiple Evanescent White Dot Syndrome. Case Rep Ophthalmol. 2018; 9(2):388–394.
8. Parodi MB, Romano F, Montagna M, Bandello F. Large choroidal excavation in a patient with rubella retinopathy. Eur J Ophthalmol. 2018; 28(2):251-252.
9. Cheung CMG, Lee WK, Koizumi H, et al. Pachychoroid disease. Eye (Lond). 2019; 33(1):14-33.
10. Shirley K, O'neill M, Gamble R, et a. Torpedo maculopathy: disease spectrum and associated choroidal neovascularisation in a paediatric population. Eye (Lond). 2018 Aug; 32(8): 1315–1320.

CONTACT

Jorge Vasco Costa
Rua Poetisa Natália Correia, 50
4435-398, Rio Tinto, Portugal
E-mail: jc09094@gmail.com

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