

Iridochoroidal, Crystalline And Eyelid Coloboma: A Case Report

Z. Hammoumi; M. El Alami; Z. Laftimi; G. Daghouj; L. Elmaaloum; B. Allali;
A. El Kettani

Ophthalmology Department, Hospital 20 August 1953
Casablanca - Morocco

Abstract

Ocular coloboma is due to failure of the closure of the embryonic fissure. It can be can be sporadic, hereditary or associated with chromosomal abnormalities. We present a 12-year-old child diagnosed with unilateral iridochoroidal, crystalline and eyelid coloboma who was referred to our department for a full ocular examination. Ocular coloboma is a rare condition. It is generally located in the inferonasal quadrant. there are some physiopathological hypotheses, but but they are controversial. Treatment is most often conservative. Ocular coloboma is a unique congenital condition that can involve many ocular structures. Early screening helps prevent possible complications

Keywords: iridochoroidal- crsystalline- eyelid-coloboma -child

Date of Submission: 07-06-2024

Date of Acceptance: 17-06-2024

I. Introduction

The word “coloboma” defines a lack. It can involve the iris alone or several other structures such as the ciliary body, the lens or the chorioretina [1]. Ocular coloboma results from incomplete closure of the embryonic fissure of the neuroectodermal optic cup around weeks 5 to 8 of gestation [2]. The localization is always in the inferonasal territory [1]. The underlying etiology for each coloboma phenotype depends on the degree of lack of fusion of the choroidal fissure folds which may affect the cornea, the iris, the ciliary body, the lens, choroids and optic nerve [3].

II. Clinical Case

A 12-year-old child presented to our hospital for vision screening. The patient had been complaining about very poor visual acuity. A visual screening examination revealed an uncorrected visual acuity of 0.3 OD and 0.1 OS using the Snellen visual acuity chart. Autorefractometry revealed sphere/cylinder X axis of +1.75/-1.50 × 20 and +2.75/-3.50 × 110 for OD and OS, respectively. Auto-keratometry and aberrometry results were unremarkable. Ocular motility was full. Examination of the left eye showed nasal eyelid coloboma, a superior nasal iris coloboma, and a lens coloboma. The zonule absence at the superior nasal level of the lens was extending from 8 to 11 o'clock without associated dislocation. The lesion on the left retina was 0.3-disc diameters below the optic disc in the inferonasal quadrants with a height and width of 0.8 and 1.1. disc diameter, respectively. The right eye examination reveals no abnormality. There was no complication such as retinal detachment or choroidal neovascularization, which can complicate this type of lesion. Optical correction and amblyopia treatment were prescribed. No management was necessary but a regular visual examination was advised.



Figure 1. (A) Eyelid, iris and lens coloboma (B) Chorioretinal coloboma

III. Discussion

The prevalence of this condition is estimated to be between 2 and 14 per 100,000 births [4]. Unilateral cases represents 50% of the cases[5]. It may be sporadic or inherited and is often associated with systemic disorders. Typical colobomas are anatomically in the inferonasal quadrant and are depending on several factors such as the location, severity of failed closure. A continuous uveal coloboma from or “skip lesions” an occur where some areas are spared [6].

The genetic and environmental causes may contribute to the etiology [7] [8]. The role of Retinoid acid in optic cup development is particularly important with regard to coloboma. It is involved in the closure of the choroid fissure, which is the primum movens of the pathogenesis of coloboma [9]. Congenital hypothyroidism is also associated with an increased risk of congenital malformations [10]. The connection between congenital toxoplasmosis and coloboma has been suggested many times[s11].

Treatment of patients with coloboma is generally conservative. It focuses on optimizing the vision of the eye affected by the pathology through optical correction and amblyopia treatment. Treatment of eyelid coloboma is based on the prevention of exposure keratopathy and referral to oculoplastics for possible eyelid reconstruction. Treatment of lens coloboma is conservative and based on correcting refractive error. If significant cataract, lens extraction is an option to be considered with intraocular lens placement. Surgery may be complicated by zonular abnormalities . Chorioretinal coloboma needs regular monitoring for retinal tear or detachment [6].

References

- [1] Nezzar H, Chiambaretta F, Rigal D, Et Al. L’iris Et Sa Pathologie. Rapport Annuel Des Sociétés De Paris. Novembre Marseille: Lamy (2003). 65-80
- [2] Duvall J, Miller SL, Cheatle E, Tso MO. Histopathologic Study Of Ocular Changes In A Syndrome Of Multiple
- [3] Pederna E, Méndez C. Embriología En La Clínica. Casos Médicos. 2006;21:125–7
- [4] Shaheen P. Shah; Amy E. Taylor; Jane C. Sowden; Nicola K. Ragge; Isabelle Russell-Eggitt; Jugnoo S. Rahi; Clare E. Gilbert; For The Surveillance Of Eye Anomalies (SEA-UK) Special Interest Group Anophthalmos, Microphthalmos, And Typical Coloboma In The United Kingdom: A Prospective Study Of Incidence And Risk
- [5] Morrison D, Fitzpatrick D, Hanson I. National Study Of Microphthalmia, Anophthalmia, And Coloboma (MAC) In Scotland: Investigation Of Genetic Aetiology. J Med Genet. 2002;39:16–22.
- [6] Patel, H. R., & Bhaleeya, S. (2016). Coloboma. Manual Of Retinal Diseases, 69–73. Doi:10.1007/978-3-319-20460-4_16
- [7] Chang L, Blain D, Bertuzzi S, Brooks BP. Uveal Coloboma: Clinical And Basic Science Update. Curr Opin Ophthalmol. Oct 2006;17:447–70.
- [8] Kalaskar VK, Alur RP, Li LK, Et Al. High-Throughput Custom Capture Sequencing Identifies Novel Mutations In Coloboma-Associated Genes: Mutation In DNA-Binding Domain Of Retinoic Acid Receptor Beta Affects Nuclear Localization Causing Ocular Coloboma. Hum Mutat. 2020;41:678–95.
- [9] Lupo G, Gestri G, O’Brien M, Et Al. Retinoic Acid Receptor Signaling Regulates Choroid Fissure Closure Through Independent Mechanisms In The Ventral Optic Cup And Periocular Mesenchyme. Proc Natl Acad Sci U S A. 2011;108:8698–703
- [10] Olivieri A, Stazi MA, Mastroiacovo P. Study Group For Congenital Hypothyroidism. A Population-Based Study On The Frequency Of Additional Congenital Malformations In Infants With Congenital Hypothyroidism: Data From The Italian Registry For Congenital Hypothyroidism (1991-1998). J Clin Endocrinol Metab. 2002;87:557–62.
- [11] Orefice F. Toxoplasmosis.Foster CS, Vitale AT (Eds.). Diagnosis And Treatment Of Uveitis. New Delhi; London: Jaypee Highlights; 2013. P. 543–4.