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Ultrasonographic examination in the diagnosis of bilateral retinochoroid coloboma

Exame ultrassonográfico no diagnóstico de coloboma retinocoroidiano bilateral

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Abstract

We report here the case of E.R.S.S. female, 43 years old, diagnosed with bilateral coloboma of choroid and retina, in order to emphasize the importance of preoperative exams, even a good shot of light projection, which is often being relegated to a negligible level in ophthalmological practice.We emphasize also that the ultrasound examination prior to the surgical indication is of paramount importance, however,this should be performed by experienced professional and the correct interpretation must be thoroughly searched for interpretative errors not become inadequate surgical clinical conduct and consequential irreparable damage. Considering all aspects and complications already mentioned in this report, coloboma to conduct before a diagnosis of this malformation should be: search for association with other eye diseases and/or systemic (CHARGE: coloboma, congenital heart defect, atresia of posterior nasal apertures with multiple anomalies), performing and monitoring treatment in case of complications (E.g. retinal detachment, amblyopia and strabismus) and prevention is made through genetic counseling.

Keywords: *Coloboma/diagnostic imaging; Retina; Choroid; Ultrasonography .*

Resumo

Relatamos aqui o caso de E.R.S.S., feminino, 43 anos, diagnosticada com coloboma de retina e coroide bilateral, afim de enfatizar a importância dos exames pré-operatórios, até mesmo de uma boa tomada de projeção luminosa, a qual está sendo muitas vezes relegada a um patamar desprezível dentro da prática oftalmológica. Salientamos também que o exame ultrassonográfico prévio à indicação cirúrgica é de suma importância, no entanto, esse deve ser realizado por profissional experiente e sua correta interpretação deve ser exaustivamente procurada para que erros interpretativos não se transformem em conduta clínico cirúrgica inadequada e consequentes danos, muitas vezes, irreparáveis. Considerando-se todos os aspectos e complicações do coloboma já citados nesse relato, a conduta diante de um diagnóstico dessa malformação deve ser: pesquisar associação com outras doenças oculares e/ou sistêmicas (CHARGE: coloboma, cardiopatia congênita, atresia de coana com múltiplas anomalias), realizar tratamento e acompanhamento em caso de complicações (Ex. descolamento de retina, ambliopia e estrabismo) e prevenção é feita através de aconselhamento genético.

Descritores: Coloboma/diagnóstico por imagem; Retina, Coroide; Ultrassonografia

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Introduction

Retinochoroidal coloboma is secondary to incomplete closure of the embryonic optic fissure [1] at choroid [2] and papilla height, which starts at the eyeball equator and goes towards the anterior part of the eye until it reaches the iris, as well as towards the optic nerve in the posterior part of the eye (where it penetrates the hyoid artery).[3] Such incomplete closure can happen at different degrees, from formes frustes, such as a simple iris coloboma, to true colobomatous cysts, which leads to a single vestigial eyeball. This process is associated with hyperplasia caused by pigmented-margins epithelium reaction. [2]

True optic disc colobomas are inferior and can have different sizes. The atypical ones are located in other retinal regions and have unclear etiology. [3] They are divided based on the incomplete closure of the anterior (corneal, crystalline, iris and ciliary body coloboma) and posterior (choroidal, retinal and optic nerve coloboma) parts of the optic fissure. In addition, it is essential emphasizing that such malformations are overall bilateral, [4] but they can also be unilateral, sporadic or autosomal dominant. [3-5]

Patients' clinical picture depends on the involvement of the optic nerve, papilla and macula,[2] whose complications can comprise: 1. Higher risk of retinal (25-33% of cases, mainly in the macular region when there is optic nerve coloboma) or rhegmatogenous retinal detachment (in case of retinochoroidal coloboma[1]) - such complications should be treated and prevented with laser treatment [2]; and 2. Changes in upper visual field and visual acuity - they depend on the involvement of the papillomacular bundle. [3] Finally, the following criteria should be taken into consideration at the time to perform the differential diagnosis of coloboma: macropapilla (papilla presenting normal features, but bigger size), hypoplastic papilla (malformed and small papilla, which can indicate association with neurological disorders) and tilted disc (tilted, hypoplastic, asymmetrical papilla associated with astigmatism [2]).

The aim of the current report was to present a case of bilateral retinochoroidal coloboma, whose correct ultrasound examination interpretation helped selecting the appropriate therapeutic approach and, consequently, enabled the prompt reestablishment of patient's vision.

Case Report

E.R.S.S., female, 43 years old, married, housewife. She first came to our office on January 31st, 2018 reporting that she always had eye issues: previous history of phacoemulsification with IOL (intraocular lens) implantation in the left eye (LE), as well as other unsuccessful procedure and retinal detachment treatment. The patient reported to have been recently subjected to ultrasound examination on her right eye, which was diagnosed with advanced cataract and retinal detachment - it was the reason why she sought a second opinion.

Upon examination:

O.D.V.: hand movements

O.E.V.: NLP (no light projection)

Biomicroscopy of the right eye: nystagmus, microcornea, inferonasal iris coloboma, 4+/4+ nuclear cataract.

Biomicroscopy of the left eye biomicroscopy: atrophic (phitisic) eyeball.

Tonometry of both eyes (BE): hampered by the presence of nystagmus.

F.O.D.: impossible due to media opacity (cataract);

F.O.E.: impossible due to corneal opacity;

The ultrasound examination of the right eye (RE) was carried out with B-mode ultrasound and a tightly positioned limbar probe at 12, 3, 6 and 9 hours. Results have shown the presence of coloboma originating behind the optic papilla; extended towards the anterior segment of it at inferonasal direction (Figures 1-6). In cases like these, it is essential noticing the presence of spicule-like technical artifacts due to sudden changes in the eye wall. We made the option to perform only phacoemulsification with IOL implantation because we did not find retinal detachment at ultrasound examination and because the patient presented visual acuity at hand movements and good light projection.

The ultrasound examination of the left eye (LE) has shown hypotonic-to-compression eyeball with reduced anteroposterior diameter and extensive choroid calcification areas.

Based on results of clinical and ocular ultrasound examinations, it was possible concluding that the patient did not have retinal detachment; she only had cataract and bilateral retinochoroidal coloboma. Thus, the adopted procedure was based on a less invasive approach, which resulted in visual acuity of 0.05.

Ultrasound examination of RE:

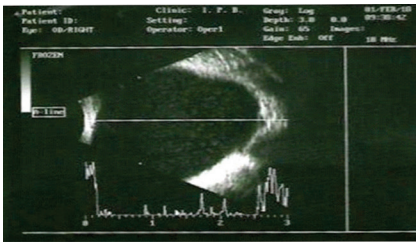

Figure 1: Inferonasal coloboma starting from the papilla.

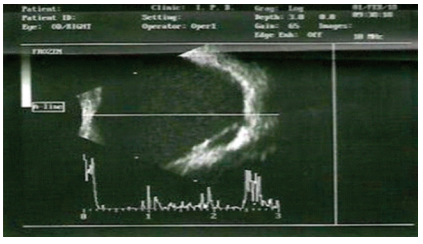

Figure 2: Fixed retina presenting detachment aspect; actually, it was just the vitreous membrane over the coloboma area.

Ultrassonografia de L.E.:

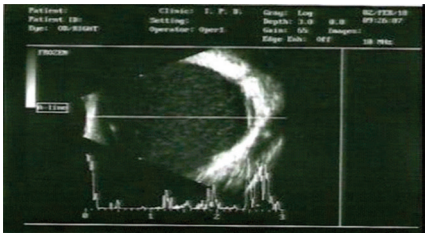

Figure 3: Another image that can be misinterpreted as detachment of a fixed retina.

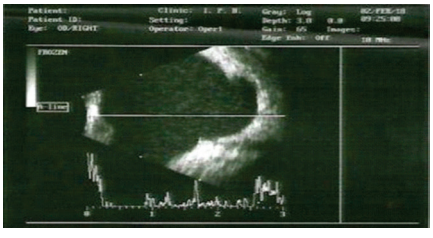

Figure 4: Image showing "spicules" next to the great eye wall "step" (technique artifact), which can be misinterpreted as retinal detachment.

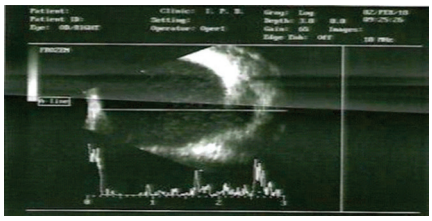

Figura 5: Outro aspecto de "espícula" que pode levar o examinador pouco experiente a idéia de descolamento de retina.

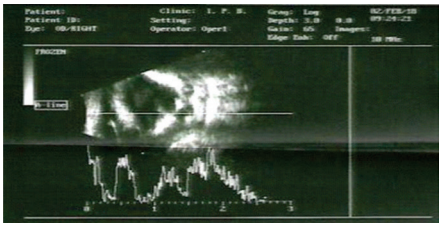

Figura 6: Globo ocular de dimensões reduzidas, hipotônico e com extensas áreas de calcificação de coroide.

Discussion

The case reported in the current study refers to a bilateral retinochoroidal coloboma (also called retinochoroidal coloboma) resulting from incomplete closure of the choroidal fissure [4] during eyeball embryogenesis, which happens between the fourth (period when the fissure appears) and sixth (period when the embryonic fissure should be fully closed) week of intrauterine life. [3] Assumingly, there is the involvement of hereditary factors associated with the lack of fusion in the edges of the inner (formed as an undifferentiated and vascularized membrane) and outer (not formed) retinal layers, pigmented epithelium and, consequently, with no choroid formation,[2] since this structure does not develop from the outer layer, [3] but from a thin interlayer membrane.[2]

The current study addressed retinochoroidal coloboma, which can present different sizes in fundoscopic examinations, mainly in the lower part of the eye, [1] in bright white, yellowish color; crystalline lens or cataract, without zonules; absence of retina and choroid (large as beveled pupil, or small - simulating chorioretinal scars on the inferonasal retina);[2] and inferior and pigmented irregular borders (sites presenting failed closure are filled with abnormal retinal tissue). They extend towards, and involve, the optical disc and reach the iris (Figure 7); or they are isolated and, finally, can be associated with multisystem diseases (e.g., Trisomy 13 and Goldenhar Syndrome), as well as with dominant or recessive autosomal factors; or they can happen as isolated manifestations.

The patient reported history of surgery on the left eye, which was phitisic at the time of our first meeting. She reported to have been subjected to ultrasound examination of the right eye in another ophthalmic service. Based on this exam, besides advanced cataract, she was diagnosed with retinal detachment; thus, surgery was recommended to improve her condition.

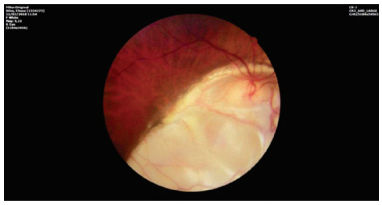

Figure 7: Fundoscopic aspect of coloboma starting from the optic nerve. Notice the absence of the inferior temporal and nasal arches.

As previously mentioned, the patient underwent ultrasound examination in the presence of media opacity.

We believe that the eye ultrasound examination interpretation was used by the other ophthalmic service as the basis to indicate facectomy and retinal detachment treatment, as well as that visual acuity measurements may have been neglected. This combined surgery can be quite complex in case of microcornea and other eye malformations, since not even the gateway of the posterior segment - the ora serrata - is well-located.

However, when there is indeed the need of intervening in the posterior segment, this challenge must be faced and surgery must be performed, mainly in the case of a young patient with a single eye who is mother of two.

The importance of addressing this case report lies on reinforcing the essential role played by ultrasound examination of the eye as a complementary exam to help getting to an accurate diagnosis and defining the best treatments in daily ophthalmologists' practices. Therefore, the correct indication, performance and interpretation of such exam, which depend on examiner's study, dedication and experience, are of paramount importance.

With respect to the herein addressed case, the recommendation of previous ultrasound examination in the other ophthalmic service was correct; however, its interpretation was not. Thus, if the patient had not had the initiative to ask for a second opinion, her case could have had a different outcome.

Thus, we made the option for adopting a more conservative, although not less risky, surgical approach, since cataract was very advanced given the patient's justifiable fear of operating her single eye. In addition, in these cases, the pupil dilates well, but there is also the absence of zonules in the region corresponding to the coloboma; therefore, there is certain instability in the crystalline, which leads to higher risk of complications.

We performed cataract surgery and implanted the intraocular lens - the surgery went well. Postoperative examinations no longer found the media opacity that was previously hindering the visualization of the fundus. It was possible seeing that the retina was, as we suspected, fully attached to the eye (Figure 8). We performed YAG laser iridoplasty at the 30th postoperative day, since patient's visual axis was impaired. Nowadays, patient's right eye has visual acuity of 0.05, which allows her to have some autonomy.

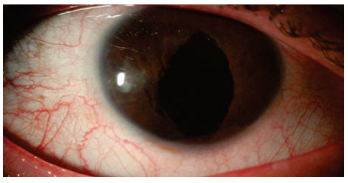

Figura 8: Biomicroscopy after facectomy and pupilloplasty favoring the visual axis.

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