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CHARGE Syndrome – Amblyopia and Coloboma: a case report

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Introduction:

Hall, in 1979, after observing the combination of choanal atresia with other congenital malformations in patients with normal karyotype described the Charge Association. Pagon (1981), proposed the acronym CHARGE to describe this set of findings: C - Coloboma. H - Congenital heart disease, A - Choanal atresia, R - Growth retardation, G – Genital anomalies, E - Ear abnormalities[1,2]. Although the cause is not known, it seems to result from specific anomalies of brain differentiation. The criteria for clinical diagnosis of CHARGE syndrome were reformulated by Verloes in 2005, and the existence of ocular coloboma is a major signal for the diagnosis of the disease[3,4]. With a prevalence of 1:10 000, it is an autosomal dominant disease. The CHD7 gene, located at the 8q12 locus, is the only gene known to be associated with CHARGE syndrome and is mutated in 95% of patients who meet the clinical criteria of the typical Verloes form and 60-70% of suspected cases[5-7] .

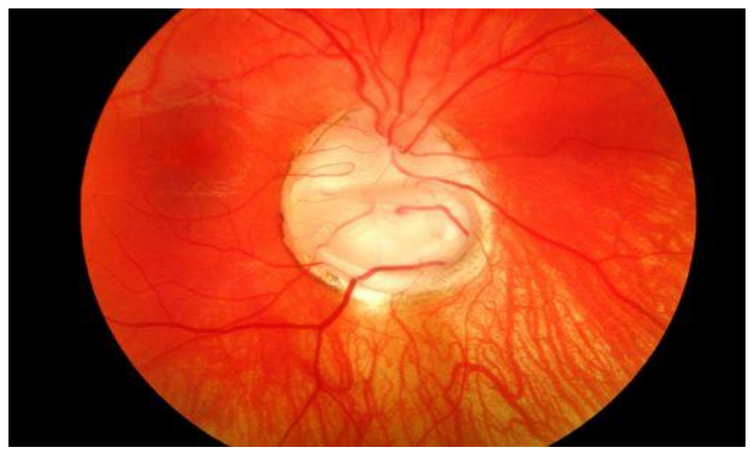

Figure 1-Retinography in a case of coloboma of the optic nerve.
Source:https://www.imagenesmy.com/imagenes/optic-nerve-coloboma-32.html (illustrative image)

Coloboma (Figure 1), is characterized by a poor closure of the embryonic fissure, which can affect various structures of the eyeball (iris, retina or optic nerve)[7]. The association with genetic pathology makes the ophthalmologic diagnosis fundamental for the multidisciplinary orientation of the patients. From the ophthalmologic point of view, patients may present with coloboma alone or in association with other changes: strabismus, nystagmus, and variable visual acuity (VA)[4,8]. The treatment of motor and sensory alterations secondary to coloboma follows the classical indications of functional strabismus and amblyopia. At structural level it is essential to monitor the evolution of the coloboma with imaging techniques because there is a risk of secondary retinal detachment. The techniques should be chosen according to the age and collaboration of the patient[9]. In children of reduced age it is usually opted for observation of the ocular fundus with indirect ophthalmoscopy and ocular echography. With the advancement of the age and with the increase of the capacity of collaboration of the patients can be introduced new techniques of image (retinography and OCT).

Case-report: A 9-month-old male child with a diagnosis of Charge Syndrome. Cesarean delivery at 37 weeks, with a history of growth below the normative for the age, being Cryptoquia presence. No family history with clinical relevance

Ophtalmic and Orthoptics Examination:

- VA= ODE 0.50 cpcm (Lea Greattings a 27.5 cm). Reference values 1-8 cpcm, below the normal value for age.
- Fundoscopy (Indirect ophthalmoscopy): Retina applied in four quadrants .
- Appearance and coloring normal and vasculars architecture in relief changes.
- Coloboma of NO in ODE - reduction of the neuro-retinal ring in ODE / increase of the Cup/Disc Ratio.
- Functional Vision: Follow the light on all positions of the look. Slow-beat nystagmus, with fast phase on the right. Limitation of ODE Abduction and of the elevation. The movements of infraversion are maintained. Grab objects at 18 cm.

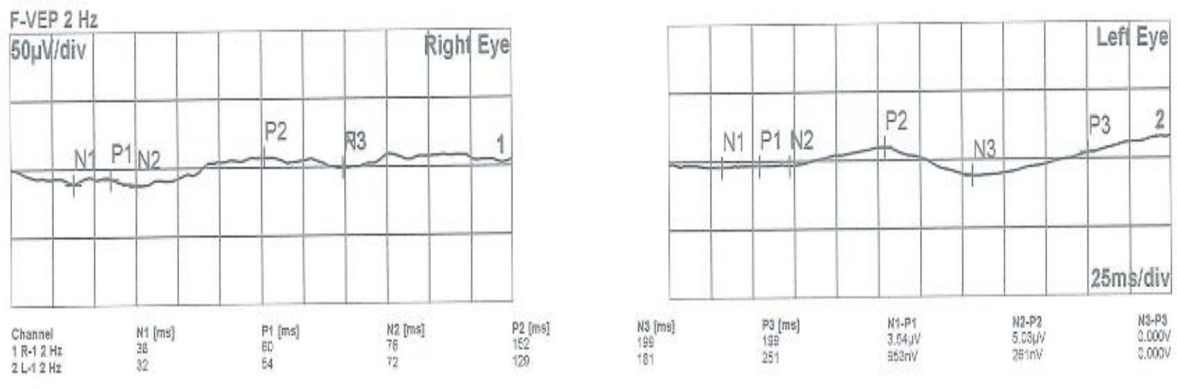

Figure 2- Electrophysiology - The fPEV

Complementary diagnostic tests:

- MRI - focal bulging of the right eye wall at the level of the optic nerve papilla and small segment below, there being on the left a similar but smaller aspect and circumscribed to the papilla region. Remaining extension of ocular globes with normal morphology, density and position. Discrete asymmetry of the optic nerves, presenting the orbital segment of the right nerve less thickness .
- Electrophysiology (ISCEV protocol) - pPEV - not identified unequivocally cortical response of both the eyes for the spatial frequencies of 60 ". The fPEV- (figure 2) - evidence response in the 20th pole centers of the retinal ODE, present and symmetric, establishing the presence of the retinal function /pre-quiasmical pathway.
- Ocular ultrasound (mode B) - Retina applied in the four quadrants in ODE Vitreous silence in ODE. Irregularity in the contour of the nasalposterior wall and lower in ODE and with a "pit" of about 3 mm peri-disc in OD. Minimal enlargement of the left disc diameter (figure 3).

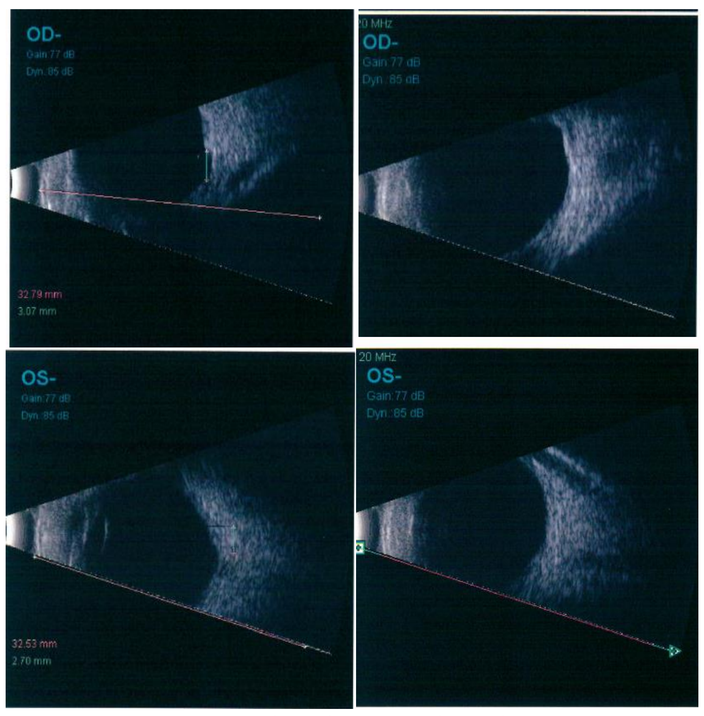

Figura 3 – Ocular ultrasound mode B ODE

Conclusion: This case requires a multidisciplinary approach, with adaptive recovery follow-up, particularly with regard to functional vision. The ocular manifestations arising from the coloboma should be treated as functional abnormalities. Given that there is a decrease in visual functions and functional vision is a process of maturation and learning[8], the VA should be stimulated to promote a good functional residue. At this stage, aiming at visual stimulation is intended to motivate the child to explore the surrounding environment and the objects that surround it, through a visual rehabilitation plan adapted to the age and characteristics of the case. The orthoptist, integrated in a multidisciplinary team and in complementarity with the ophthalmologist, plays a fundamental role in these types of cases, both at the level of diagnosis, follow-up and rehabilitation program through the accomplishment of the various complementary diagnostic exams, design and implementation of the strategies of necessary rehabilitation.

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The authors declare no conflict of interest