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CHARGE SYNDROME - AMBLYOPIA AND COLOBOMA: A CASE REPORT

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Purpose: CHARGE syndrome is caused by a genetic disorder, with different associated anomalies is a recognizable pattern of congenital malformations. Coloboma, one of the developmental anomalies, is characterized by a poor closure of the embryonary fissure, which can affect the eye different structures. The association with systemic pathology makes the ophthalmologic diagnosis essential for patients multidisciplinary orientation. From an ophthalmological point of view, patients may present with coloboma associated with other changes, namely strabismus and nystagmus, and with amblyopia and variable visual acuity.

Methods: Case report on a 9-month-old boy with diagnosed CHARGE syndrome with bilateral optic nerve coloboma, amblyopia and other anomalies.

Results: Magnetic Resonance and Ocular Ultrasound showed the existence of optic nerves coloboma. In the level of electrophysiology there is a diminished overall retinal response. In the orthoptics’ evaluation, it was verified that the visual acuity 0.50ccpm (Teller Acuity Cards), also presenting nystagmus with slow beats and fast phase to the right, ocular motility with limitation of abduction and elevation, bilaterally.

Conclusion: This case requires a multidisciplinary approach, with adaptive recovery follow-up, particularly regarding 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 that functional vision is a process of maturation and learning, visual acuity should be stimulated to promote a good functional residue. Orthoptists play a fundamental role in this process since they intervene both at the level of diagnosis and follow-up and throughout the functional vision rehabilitation process.

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TREATMENT OUTCOMES OF OCCLUSION THERAPY IN CHILDREN WITH BILATERAL ISOMETRIC AMBLYOPIA

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Purpose: To investigate the treatment outcomes with spectacle correction and/or occlusion treatment (OT) in children with bilateral isometric amblyopia.

Methods: Forty-one previously untreated bilateral isometric amblyopia were enrolled. Inclusion criteria: Visual acuity (VA) being worse than 0.25 logMAR in both eyes with spherical equivalent (SE) difference less than 1.0 Diopters (D) and cylindrical difference less than 1.50. Full myopic and astigmatic errors were corrected, and hyperopia were corrected full or under-corrected within 1.5D. Baseline VA, cycloplegic refraction, stereopsis, VEP were assessed. VA assessed at every 4 weeks until achieving 0 logMAR VA for both eye.

Results: Mean age was 5.12±1.91 years. OT was applied to twenty-two children. Mean baseline VA were 0.41±0.19 logMAR for better eye, 0.56±0.25 logMAR for fellow eye. When better eye achieved 0 logMAR VA, OT was significantly helpful in whom showed VA of fellow eye over 0.23 logMAR (p=0.012). There were no significant correspondence with baseline age and the time achieving 0 logMAR VA in both eye. Baseline SE difference or VEP results didn’t show correspondence with OT needs or full VA improvement period. Near stereoacuity was significantly improved from 283.13 to 43.44 arcsec, but had no correlation with OT needs. Baseline lower VA showed positive correlation with full VA improvement period (R=0.266, p=0.016).

Conclusion: Baseline lower VA was important in predicting resolution time of amblyopia and occlusion was significantly helpful in isometric amblyopia with different VA.