An overview on coloboma: A congenital malformation of the eye.

San Arrie*

Department of Ophthalmology, University of Lisboa, Lisbon, Portugal

Accepted on 10 December, 2021

Description

Coloboma is a Greek term for mutilation. The embryonal fissure fails to close properly, resulting in coloboma of the fundus. The term "Typical coloboma" refers to deformities visible in the inferior/infero-nasal region of the fundus that may be clearly traced to a fault in the closure of the embryonal fissure. Atypical colobomata are malformations that have been encountered in other places. The term "Complete coloboma" refers to abnormalities in the optic disc, choroid/retina, ciliary body, zonules, lens (notching), and iris. Cysts can form from the coloboma's margin and extend into the orbit, turning the eye non-functional, but modest involvement, such as moderate hypoplasia of the iris, a notch in the pupillary border, and so on, have no influence on the eye's function. Fundus coloboma has been linked to varying degrees of microphthalmos. Although sporadic coloboma is prevalent, many systemic abnormalities and syndromes have been identified, some of which are hereditary in nature.

The involvement of the macula and optic disc in the coloboma, as well as the increased chance of Retinal Detachment (RD) over the individual's lifespan, all create a threat to vision. With the introduction of pars plana vitrectomy procedures, the care of coloboma-related RD's has undergone substantial improvements and now gives good consistent results.

Several eye field transcription factors, including PAX6, SIX3, LHX2, and RAX, are required in various combinations at each stage of eye development. Intrinsic transcription factors interact and modify extrinsic signals. The optic vesicle includes retinal stem cells, which can differentiate into neuro-sensory retinal cells, RPE, or optic stalk depending on the appropriate signal combination. The presumed neural retina (distal region of the optic vesicle) expresses VSX2, while the proximal section that develops into RPE expresses MITF. PAX2 is expressed in the potential optic stalk. The mutual antagonism between MITF and VSX2 establishes the barrier between RPE and neural retina, whereas the antagonism between PAX2 and PAX6 provides the boundary between optic stalk and neural retina. Several external factors, including members of the TGF, fibroblast growth factor, Sonic Hedge Hog (SHH), and WNT signaling families, influence the WNT signaling pathway

Ocular coloboma manifests itself in a variety of ways. While hereditary instances and those with chromosomal abnormalities are extensively recognized, spontaneous colobomata are more prevalent, and environmental and maternal factors are more likely to be responsible. Visual acuity may be compromised by the coloboma itself if it affects the disc and fovea, or by complications such as RD, choroidal neovascularisation, and so on. The identification of ICM detachment is critical for understanding the role of coloboma in the pathogenesis of RD. Pars plana vitrectomy with silicone oil tamponade and endolaser along the coloboma margin has a high success rate.

Conclusion

Future research should focus on (a) Improved methods of detecting causes of non-syndromic coloboma; (b) Improved imaging with sweeping source OCT and longer scans that provide a panoramic view of the anatomy; (c) Randomized controlled studies to investigate the function of laser photocoagulation in the prevention of RD; (d) Randomized controlled trials to examine at the role of optimum internal tamponade in the treatment of coloboma-related RD's.

*Correspondence to

Dr. San Arrie

Department of Ophthalmology

University of Lisboa

Lisbon, Portugal

E-mail: sanarrie$@umlub.pt

Citation: *Arrie S. An overview on coloboma: A congenital malformation of the eye. J Clin Ophthalmol. 2021;5(S6):499.*