Optic Disc Coloboma in Two Nigerian Female Siblings

Yewande O. Babalola,[1,2] Olusola O Olawoye, [1,2] Patrick O Idam[2] and Adeyinka Ashaye[12]

[1]Department of Ophthalmology, College of Medicine, University of Ibadan, Nigeria
[2]Deparment of Ophthalmology, University College Hospital, Ibadan, Nigeria
Corresponding author: Patrick Idam. Email: idampat@yahoo.com

Introduction: Optic disc coloboma is characterized by a bowl-shaped excavation of the optic disc and results from incomplete closure of the embryonic fissure.[1] We report a case of optic disc coloboma in 2 Nigerian female siblings.

Methods: *Case report.*

Sibling 1: A 9-year old girl was referred from a catholic hospital on suspicion of having congenital glaucoma. She presented with a 3-year history of poor vision in both eyes with history of learning disability following childhood febrile convulsions. She lacked adequate social skills expected for age but was otherwise healthy. Her visual acuity could not be assessed as she was uncooperative, however fixation in both eyes was central, steady and maintained. Both anterior segments were normal and IOP was 12mmHg bilaterally. Binocular indirect ophthalmoscopy (BIO) of the RE revealed an enlarged optic disc with inferior whitish bowl-shaped excavations, a strip of pink NRR superiorly, normal vessels/macula and flat retina. Her LE revealed an enlarged optic disc with inferior whitish bowl-shaped excavation and superior wedge of pink NRR. A coexisting optic disc pit was also seen at the temporal aspect of the left disc with normal vessels/macula and flat retina. A diagnosis of Bilateral Optic Disc Coloboma and Left Optic Disc Pit was made. She had refraction and spectacles were prescribed. She was referred to a paediatric neurologist who made an assessment of severe intellectual disability and recommended a special tutor.

Sibling 2: Her 8-year-old sister subsequently presented with a 5-month history of poor vision in the LE. VA was 6/6 in the RE and CF in the LE with a 15^0 esotropia. She had normal anterior segment examination and IOP was 14mmHg bilaterally. BIO of the RE revealed a large excavated disc with a strip of pink NRR superiorly, normal vessels/ macula and flat retina. BIO of the LE revealed a small hypoplastic disc with peripapillary atrophy, normal vessels/ macula and flat retina. A diagnosis of a Right Optic Disc Coloboma and Left Optic Disc Hypoplasia was made.

Discussion: Optic disc coloboma is rare and usually sporadic but may be inherited as an AD disorder.[1] The latter may be the case in these sisters. It may occur as an isolated defect or in association with other congenital disc anomalies as seen in our patients, i.e. optic disc pit and hypoplasia. Developmental retardation seen in this case appears to be acquired but could also occur as part of congenital syndromes such as CHARGE syndrome [2]. The patient was thought to have congenital glaucoma by the referring center. Optic disc colobomas may be mistaken for glaucoma[1] and though a rare disease, a high index of suspicion may help identify cases and save both patients and caregivers from lifelong burden of unnecessary glaucoma treatment. Multidisciplinary management is vital as other co-morbidities such as neurodevelopmental deficits, cardiac and renal anomalies may be present.[5]

Conclusion: Optic disc coloboma does occur in our environment and may be misdiagnosed as glaucoma. Routine screening of relatives of patients who present with hereditary ocular disorders should also be encouraged.

References

1. Norisiki A, Yamaguchi Y, Handa H, Tadokoro K, Asaka A, Kawase E et al: Mutations of the PAX6 Gene Detected in Patients with a Variety of Optic Nerve Malformations Am J Hum Genet. 2003; 72: 1565-150
2. Onwochei, B. C., Simon, J. W., Bateman, J. B., Couture, K. C., & Mir, E. Ocular Colobomata. Survey of Ophthalmology, 2000; 45; 175–194.
3. Amador-Patarroyo, M. J., Pérez-Rueda, M. A., & Tellez, C. H. Congenital anomalies of the optic nerve. Saudi Journal of Ophthalmology. 2014; 29: 32–8.