

A Report Of A Case Of Bilateral Iris Coloboma In A 9 Year Old Boy

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Abstract

INTRODUCTION

Development of the eye begins in the 22 day embryo with the appearance of a pair of shallow grooves on each side of the invaginating forebrain.¹

Subsequently a series of events including multiple inductive and morphogenic events, proliferation and differentiation of cells into mature tissue, and establishment of neural networks connecting the retina to the higher neural centres (superior colliculus, lateral geniculate bodies, occipital lobes, etc) occur.²

Stages of development include formation of the optic vesicles, optic cup, choroidal fissure, hyaloid artery, and the primitive retina.

During the 7th week, the lips of the choroidal fissure fuse, and the mouth of the optic cup then becomes a round opening, the future pupil.

Under normal conditions, closure of the choroidal fissure occurs during the 7th week of development, failure of which results in a persistent cleft.

This cleft is most commonly located in the iris only, and is known as a coloboma iridis. However it may extend into the ciliary body, the retina, the choroidal and the optic nerve. This malformation is frequently seen in combination with other eye abnormalities.³

A coloboma is a localized absence or defect of the uveal tract. They are caused by the incomplete closure of the choroidal fissure producing incomplete development of mesodermal and neurectodermal component in the area of the cleft.

They typically occur inferonasally, and can be complete or

incomplete. Atypical coloboma can occur in any other area.

In this article we present iris coloboma which has an incidence of 0.007% in the UK population. No data exists on the incidence in Nigeria.

CASE PRESENTATION

A 9 year old male child in primary 5 with normal intelligence and normal general features presented at the eye clinic of the University College Hospital Ibadan on account of photophobia and difficulty in seeing in bright light.

He was brought in by his mother who claimed he had had the problem since early childhood. There was no family history of a similar occurrence.

On careful and thorough examination, which entailed slit lamp examination, slit lamp biomicroscopy with +78DS Volk's lens and binocular indirect ophthalmoscopy, he was noted to have unaided distant visual acuity of 6/9 in each eye, not improved by pin hole.

He had unaided near visual acuity of N6 in each eye. He had no nystagmus or strabismus.

On slit lamp examination, the eyelids, conjunctiva and cornea of both eyes were normal. The anterior chambers were deep and normal bilaterally. There was an inferior iris defect extending to the ciliary body in both eyes (Figure 1-3).

Figure 1



The anterior chamber angles were normal bilaterally and both lenses were clear. The vitreous was clear and the retina and optic nerve were normal in both eyes.

Systemic examination revealed no abnormalities, and radiological investigations carried out were all within normal limits.

The mother gave a history of uneventful pregnancy, birth and neonatal history. She had routine antenatal care in a private hospital, and was given 'routine' drugs, namely fesoate tablets, folic acid tablets, and vitamin B complex tablets.

The main aim of treatment was to relieve the discomfort from photophobia especially when in bright light, and to correct any refractive error present. Refraction did not however improve his vision significantly.

Cosmetic opaque contact lenses with clear optical centres were prescribed. This cuts out the excess light entering his eyes via the peripheral iris defect, leaving the central corneal region as the only point of entry of light into the eyes. This will relieve photophobia and possibly improve his distant and near vision.

Informed consent: Both oral and written informed consent was given by the boy's mother for his pictures to be taken, and for this article to be written and possibly published.

DISCUSSION

A large proportion of sporadic, unilateral or bilateral colobomas are likely due to non genetic factors. Many non-Mendelian, multisystemic malformation syndromes are associated with colobomata. Examples include the CHARGE syndrome where approximately 86% of patients have uveal or iris colobomata⁴, and naevus sebaceous of Jodassohn where some patients have iris and choroidal colobomata.⁵ The underlying mechanisms are not known for such syndromes, which constitute a significant proportion of coloboma cases.

Majority of inherited coloboma cases are associated with systemic disease. Twenty seven genetic loci have been mapped to specific chromosomal regions, and 21 of the genes have been identified. Eleven chromosomal aberrations have been documented and 3 of these overlap with known coloboma associated genes (SHH, CHX 10, MAF).²

In phenotypes where there is no mapping information, 13 show autosomal dominant inheritance, 14 are autosomal recessive, 3 are thought to be X- linked, and in 7 phenotypes the mode of inheritance is yet to be established.

A number of studies in humans suggest that the use of certain drugs during pregnancy may be associated with ocular coloboma. These include thalidomide and alcohol abuse.^{6,7} Other reported causes of ocular coloboma are maternal infections caused by cytomegalovirus, toxoplasmosis, vitamin E deficiency, ionizing radiation and hyperthermia.^{8,9,10,11,12,13,14,15,16,17}

Management of iris coloboma entails a thorough general examination to rule out any associated abnormalities such as heart abnormalities, chonal atresia, genital abnormalities, ear abnormalities and growth retardation. One should also rule out dental anomaly and mental retardation.

Ocular examination should be done to rule out nystagmus, strabismus, reduced vision from refractive errors or amblyopia. The patient should also be examined for aniridia, microphthalmia, anophthalmia, and also coloboma of other parts of the eye.

Investigations should include Electrocardiography, Echocardiography, Cranial MRI, abdominopelvic ultrasound and chromosomal studies.

Treatment involves refracting the patient and prescribing spectacles if needed, preferably tinted. Tinted iris contact lenses and tinted intraocular lenses are also useful. Amblyopia and strabismus should also be treated.

Surgical care entails surgical iridoplasty to cover the iris defect. Other ocular and systemic anomalies that require correction should also be attended to.

SUMMARY

Iris coloboma is a very rare ocular condition and can be unilateral or bilateral. They are mostly sporadic, but could also be associated with other ocular or systemic malformation syndromes; hence thorough ocular and systemic examination/ investigation is essential.

Photophobia and refractive errors are the commonest presentations. Cosmetic opaque contact lenses with clear optical centres can be prescribed. This cuts out the excess light entering the eyes via the iris defect, leaving the central corneal region as the only point of entry of light into the eyes, relieving photophobia and improving distant and near vision.

Refractive errors should also be corrected. In some cases, where indicated, surgical iridoplasty could be performed.

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