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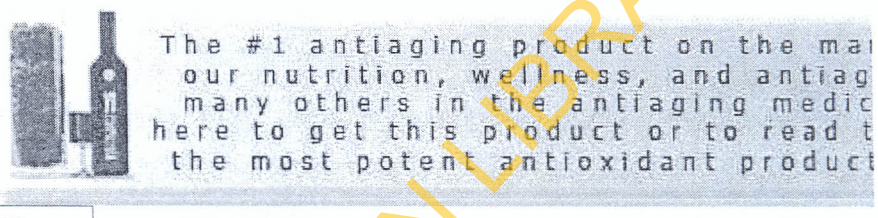

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A Report Of A =ase Of Bilateral Iris Coloboma In A 9 Year Old Boy

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Abstract

Introduction

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

Subsequently a series of events including multiple inductive =nd morphogenic events, proliferation and differentiation of cells =nto mature tissue, and establishment of neural networks connecting the retina =o the higher neural centres (superior colliculus, lateral geniculate =odies, occipital lobes, etc) occur.[2]

Stages of development include formation of the optic vescicles, =ptic cup, choroidal fissure, hyaloid artery, and the primitive =etina.

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

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

This cleft is most commonly located in the iris only, and is =nown as a coloboma iridis. However it may extend into the ciliary body, the =etina, the choroidal and the optic nerve. This malformation is frequently =een in combination with other eye abnormalities.[3]

A coloboma is a localized absence or defect of the uveal tract. =hey are caused by the incomplete closure of the choroidal fissure =roducing incomplete development of mesodermal and neurectodermal component =n the area of the cleft.

They typically occur inferonasally, and can be complete or =ncomplete. Atypical coloboma can occur in any other area.

In this article we present iris coloboma which has an incidence =f 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 =nd normal general features presented at the eye clinic of the =niversity College Hospital Ibadan on account of photophobia and difficulty =n seeing in bright light.

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

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

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

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

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

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

The mother gave a history of uneventful pregnancy, birth and =eonatal history. She had routine antenatal care in a private hospital, and =as given 'routine' drugs, namely fesolate tablets, =olic 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 =efractive 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 =he peripheral iris defect, leaving the central corneal region as the =nly point of entry of light into the eyes. This will relieve =hotophobia and possibly improve his distant and near vision.

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

Discussion

A large proportion of sporadic, unilateral or bilateral =olobomas are likely due to non genetic factors. Many non-Mendelian, =ultisystemic malformation syndromes are associated with colobomata. Examples =nclude the CHARGE syndrome where approximately 86% of patients have uveal =r iris colobomata [4], and naevus sebaceous =f Jodassohn where some patients have iris and choroidal colobomata.[5] The

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underlying =echanisms are not known for such syndromes, which constitute a significant =roportion of coloboma cases.

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

In phenotypes where there is no mapping information, 13 show =utosomal dorminant inheritance, 14 are autosomal recessive, 3 are thought =o 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 =rugs during pregnancy may be associated with ocular coloboma. These =nclude thalidomide and alcohol abuse.[6,7] *Other reported causes =f ocular coloboma are maternal infections caused by cytomegalovirus, =oxoplasmosis, vitamin E deficiency, ionizing radiation and =yperthermia.*[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 =bnormalities and growth retardation. One should also rule out dental anomaly =nd mental retardation.

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

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

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

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

Summary

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

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

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

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