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CASE REPORT

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Familial congenital glaucoma and epilepsy: a case series.

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Abstract We present two siblings from consanguineous marriage, both with congenital glaucoma and seizure disorders with progressive visual impairment and blindness. The pedigree showed that five (one male and four females) of the eleven siblings have varied degrees of visual impairment to blindness with seizure disorders.

To the best of our knowledge, familial congenital glaucoma with epilepsy has not been reported, hence the communication to highlight this unusual condition which could be an association or syndromic.

Key words: Consanguinity, Familial, Congenital glaucoma, Epilepsy.

Introduction

Causes of blindness vary between developing and developed countries. In Africa, corneal scarring due to vitamin A deficiency, measles, and ophthalmia neonatorum are the common causes, whereas, hereditary causes such as inherited retinal dystrophies, glaucoma, cataract and other causes like cerebral hypoxia and retinopathy of prematurity predominate in the European countries.[1]

Nearly 1.5 million children worldwide are blind because of simple vitamin A and micronutrient deficiencies.[1,2] Most congenital abnormalities of the eyes are caused by genetic factors or by intrauterine infections and about 10 percent of congenital glaucoma tend to have congenital rubella syndrome.[3]

Stoll and colleagues[4] have established a significant association between eye malformation and consanguinity with risk of more than three times compared to the controls. However, Akinsola and Ajaiyeoba[5] have found no association between congenital glaucoma and consanguinity.

The priority in the management of children with congenital glaucoma include, clinical control of the progression of the disease, correction of refractive errors, regular follow-up and subsequent rehabilitation of the visually impaired handicap as well as genetic counseling.

Case reports

Case 1

BK was an eleven year old girl and the ninth child of consanguineous parents. She was first seen at the age of 7 months with excessive tearing, photophobia, jerky movement of both eyes and generalized, tonic, clonic, seizures. Examination under anaesthesia (EUA) revealed features of congenital glaucoma (bilateral buphthalmos) with intraocular pressure (IOP) indentation of 25mmHg in both eyes. Bilateral trabeculotomy was performed and she did well post-operatively.

A repeat EUA one year later revealed remarkable reduction of the IOP indentation to 7.5mmHg and 8.5mmHg in the right and left eyes respectively. However, she progressively lost her sight in both eyes, but had good seizure control with carbamazepine.

Physical examination after the last follow-up visit revealed no stigmata of neurocutaneous syndrome, her anthropometry including the head circumference were normal and ophthalmic examination revealed no light perception (NLP) in both eyes. There were no clinical features suggestive of homocystinuria such as skeletal abnormalities, blue eyes or mental retardation.
Skull radiography revealed normal findings, electroencephalography and brain neuro-imaging were not done due to lack of facilities. Urinalysis was normal and serology for rubella antibodies was negative.

Case 2

MK was the 18 year old sister of the BK, and the sixth child of the family. She was followed up from the age of three months when she had bilateral trabeculectomy for congenital glaucoma; by 12 years of age, was blind in both eyes.

There was no antecedent history of head injury. She also developed generalized, tonic, clonic, seizures in early infancy and was controlled with carbamazepine.

Physical examination during the last follow-up visit was unremarkable except for 4 café au lait spots measuring about 3mm in diameter on the left axillae. As was the case with her younger sister, urinalysis and skull radiograph were normal but neuro-imaging of the brain was not done. The parents were counseled.

Discussion

Congenital glaucoma is a rare cause of progressive visual impairment with subsequent blindness in childhood and constituted 4.5 percent of Paediatrics Ophthalmic diagnoses in eye clinics of the Ogun State University Teaching Hospital Nigeria.[6]

The pathogenesis of primary congenital glaucoma has been attributed to impaired aqueous outflow as a result of trabeculodysgenesis.

The association between eye malformations especially, congenital glaucoma and consanguinity of parents have been established,[4] as seen in these cases. Majority of cases occur sporadically and about 10 percent of the cases are inherited as autosomal recessive with incomplete penetrance.[7] However, Akinsola and Ajaiyeoba[5] reported five children with glaucoma without known history of consanguinity.

Seizures are common in paediatrics age group and occur in about 10 percent of children.[8] Although, most seizures in children are provoked by somatic disorders of extra cranial origin, such as high fever, infections, hypoxia, hypoglycemia, less than one third are caused by epilepsy; recurrent seizures triggered within the brain due to central nervous system malformations, tumours or idiopathic epilepsy. The prognosis of epilepsy in childhood is generally good, but 10-20 percent have persistent seizures refractory to drugs and thus poses a diagnostic and management challenge.[8] Seizure control in both patients herein reported was good.

There is an association between neurocutaneous syndromes such as Neurofibromatosis type 1, tuberous sclerosis and Sturge-Weber disease with seizure disorders. However, the café-au-lait spots found on MK was unlikely to be significant as there were no other stigmata of neurocutaneous syndromes in either sibling. The cause of afebrile seizures in the patients therefore remains unexplained.

One diagnostic possibility is that of homocystinuria which may present with both glaucoma and seizure disorder. However, there were no other clinical features of this condition such as skeletal abnormalities, blue eyes or mental retardation in either patient. The urinalyses of their freshly voided urine were negative for homocysteine, but their hepatic enzymes were not assayed.

Although, neuro-imaging was not done because of lack of requisite facilities, the absence of clinical features suggestive of congenital rubella syndrome such as microophthalmia, cataract and deafness coupled with negative maternal antibody for rubella infection makes congenital rubella infection unlikely. Lack of clinical features of homocystinuria with normal urinalyses for reducing substances also makes the diagnosis of this condition remote. From the family pedigree and the consanguinity parents, it is tempting to speculate the possibilities of autosomal recessive disorder/familial congenital glaucoma and epilepsy in the cases presented as an association or syndromic, which needs further elucidation.

Recommendation

Despite the fact that both siblings were identified at infancy and regularly followed-up, the outcome was blindness. Therefore, genetic counseling of prospective consanguineous couples about possible manifestation of recessive disorder as exemplified in this report is advocated.

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References

1. Foster A, Gilbert CE. Epidemiology of childhood blindness. Eye 1999; 2(6): 173-6.
2. Community Eye Health. Prevention of childhood blindness. International Centre for Eye Health Institute of Ophthalmology. Slides/text teaching series no. 4, 1998.
3. Jack J Kanski. Primary congenital glaucoma. In: Kanski JJ (editor). Clinical Ophthalmology: A systemic approach, 6th edition. Edinburgh: Butterworth Heinemann Elsevier, 2007: 417-420.
4. Stoll C, Alembik Y, Dott B, Roth MP. Epidemiology of congenital eye malformation in 131,760 births. Ophthalmic Genetics 1992; 13(3): 179-186.
5. Akinsola FB, Ajaiyeoba AI. Causes of low vision and blindness in children in a blind school in Lagos, Nigeria. West Afr Med J 2002; 21(1): 63-5.
6. Haddad MAO, Sampaio MW, Oltrogee EW, Kara-Jose N, Betinjane AJ. Visual impairment secondary to congenital glaucoma in children: visual responses, optical correction and use of low vision Aids. Clinical Science. Clinics, Sao Paulo 2009; 64(8):725-30.
7. Onabolu OO. Pattern of Paediatric Ophthalmic Admissions into the Ogun State University Teaching Hospital, Sagamu. Nig J Paediatr 2002; 29(2): 47-52.
8. Johnston MV. Seizures in Childhood. In: Behrman RE, Kliegman RM, Jenson HB (editors). Nelson Textbook of Pediatrics, 17th edition. Philadelphia: Saunders, 2004:1993-2009.