

CASE REPORTS

Blindness from bilateral bullous retinal detachment: tragedy of a Nigerian family

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Abstract

Objective: To present a scourge of blindness possibly due to an inherited condition causing retinal detachment in 3 siblings

Methods: In February 2004, three siblings from a monogamous family from Ipetu-Ijesha in Osun state, Nigerian presented to the author consecutively with history of visual impairment/ blindness. A detailed history and comprehensive ocular examination was conducted on each of them. Information obtained included patients demographic, family history of blindness, eye diseases and other medical conditions

Visual acuity assessment, slit lamp examination, direct and indirect ophthalmoscopy, intra-ocular pressure measurement, visual field test and systemic evaluation were conducted on each of the patients.

Results: Two males and a female patient from the same parents who were systemically healthy are reported. Their ages were 67 years, 58 years and 52 years respectively. Each presented with bilaterally poor vision ranging from 6/36 to no light perception, intra-ocular pressure ranging between 6 and 18 mmHg and bilateral grayish white bullous retinal detachment.

Conclusion: The mode of inheritance in these patients was most probably autosomal dominant or X-linked recessive. Ignorance and poverty causing failure to seek prompt specialized eye care were responsible for blindness in these siblings.

Key Words: Bullous Retinal detachment, Aetiology, Inheritance

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Introduction

Retinal detachment (RD) refers to separation of the inner layers of the retina from the underlying retinal pigment epithelium. Exudative or serous detachments occur when subretinal fluid accumulates and causes detachment without any corresponding break in the retina. Exudation of material into the subretinal space from abnormal retinal vessels such as in hypertension, central retinal venous occlusion, vasculitis, or papilledema may result in bullous retinal detachment. Ocular tumors (e.g. primary malignant melanoma of the choroid, hemangioma of the choroid, retinoblastoma or metastatic carcinoma to the choroid from breast cancer, lung cancer) and inflammations are characterized by a broken blood-retinal barrier leading to RD.

Although RDs usually are sporadic events, certain pedigrees may be prone to detachment. The mode of inheritance in most of the disease associated with bullous exudative RD (BERD) have been widely studied.^{1,2} Vascular causes such as angiomas of the retina, Von Hippel disease, telangiectasia retina, juvenile Coat disease, adult Coat disease, Eales disease and retinal vein occlusion show variable form of inheritance.³⁻⁵ Systemic diseases associated with retinal detachment include diabetes, tumors (eg, breast cancer, melanoma), angiomas of the CNS, sickle cell disease, Leukemia and eclampsia.⁶⁻⁸

Tragically, RDs were uniformly blinding until the 1920s when Jules Gonin, MD, pioneered the first repair of RDs in Lausanne, Switzerland.⁹ Today, with the advent of scleral buckling, intravitreal gas, microscopic, laser, and cryotherapy techniques, rapid emergency department diagnosis and treatment of an RD truly can be a vision-saving opportunity. Ultimate outcome depends upon the time the retina is detached, the underlying mechanism of the RD, and whether the macula is involved. Prognosis is related inversely to the degree of macular involvement and the length of time the retina has been off.

Given the diverse nature of the underlying causes of retinal detachments², we are not aware of any reports on the frequency locally or abroad. Also, reports of simultaneous occurrence of bullous exudative retinal detachment (BERD) in siblings were not found in literature. This report aimed at presenting three siblings of a monogamous African family suffering from possibly preventable severe visual impairment and blindness resulting from bilateral bullous retinopathy with a view of advocating for a more intensive eye health education, community eye screening for eye diseases and provision of appropriate facility for specialized eye care in the developing country. These are necessary in achieving the goal of VISION 2020.

Case reports

Informed consent was obtained from the patient before writing up this report.

Case 1 is a 58 year old Ijesha female farmer from Osun state. She presented to a private eye clinic on the 2nd of February 2006 with a 6 months history of sudden loss of vision in the left eye and rapidly progressive loss of vision in the right eye of 3 months duration. At the initial stage of her eye ailment patient experienced floaters, photopsia and metamorphosia. As the photopsia became progressively more disturbing the patient's vision worsens. She had visited several chemist shops and opticians who prescribed various eye drops at each occasions with no improvements in her symptoms. She also visited traditional healers who prescribed herbal remedies both topically and orally with no positive effects.

She is the 2nd child in a monogamous family with five children. Mother and father had significant visual impairment (could not recognize faces though move round comfortably) but neither of them was blind before they died. She was not aware of any other family history of blindness.

A presentation her visual acuity was count finger in the right eye and no light perception in the left eye, vision did not improve with a pin hole. There was poor pupillary reaction to light in the right eye and apparent pupillary defects in the left eye with grayish fundal reflex. On dilated funduscopy, there was bilateral bullous retinal detachment involving the maculae, this was confirmed by ocular ultrasound. Intra-ocular pressures measured with the Perkins' Applanation tonometer was 6mmHg and 11 mmHg in the left eye and right eye respectively, no peripheral retinal breaks were seen with careful scleral depression

A diagnosis of bilateral bullous total retinal detachment was made and patient was urgently referred to Eye Foundation, Lagos or National eye Centre, Kaduna (both in Nigeria) for fundal photography, fluorescein angiography and further management especially of the right eye. She was placed on gtt. Mydracyl and gtt Betnesol N. She was counsel to seek urgent attention from either of the referral centre. Patient reported back in three weeks to say that she could not afford the treatment in one of the hospital and the machine in the other hospital was faulty and as such no treatment had been offered. Patient however defaulted and could not be followed up further. Lack of facility for imaging is a major limitation to providing appropriate illustration of the patients clinical presentation likewise in the two other affected siblings.

Case 2 is a 67year old farmer in Ipetu-Ijesha, Osun state, Nigeria. He is the elder brother of case 1.

Information about the need to refer his sister for treatment at Lagos prompted him to seek for treatment. He presented with a history of loss of vision in the right eye of 8 months duration and severe visual impairment in the left eye of 2 months duration. There was no history of trauma, previous eye surgery, intra-ocular foreign body, ocular inflammation or use of glasses. The patient was not a known diabetic, hypertensive or sickle cell patient. The patient was aware that his younger sister was having an eye problem but was not sure of the nature or its aetiology.

His visual acuity was hand movement in the right eye and 6/60 in the left, vision did not improve with a pin hole. There was no detectable abnormality on slit-lamp examination. Intra-ocular pressure was 12 mmHg and 15mmHg in the right and left eye respectively. Dilated funduscopy revealed extensive bullous retinal detachment involving the macula in the right eye with macular sparing in the left eye. No retinal holes were detected. The patient could not afford an ocular ultrasound.

A diagnosis of bilateral bullous retinal detachment was made and treatment was instituted as for the first case while patient was counsel to seek for further evaluation and management including fundal photography, fluorescein angiography at either of the two institution mentioned earlier. This prompted him to inform his younger brother who had complained of an eye ailment to him earlier.

Case 3 is a 52 year old trader who resides in Ibadan, Oyo state of Nigeria. He was the youngest of the three. He was informed of the eye problem in his siblings; this prompted him to come for an ophthalmologic assessment. He had already noticed significant reduction in his left eye vision with blurring of vision in the right eye.

There was no history of trauma, previous eye surgery, intra-ocular foreign body, ocular inflammation nor use of glasses. The patient was not a known diabetic, hypertensive or sickle cell patient. His visual acuity was 6/36 in the right eye and Count Fingers (CF) in the left eye.

Slit lamp examination of the anterior segment revealed no abnormality. Intra-ocular pressure was 16 mmHg and 14mmHg in the right and left eye respectively. Dilated funduscopy revealed extensive bullous retinal detachment involving the macula in the left eye with macular sparing in the right. Funduscopy in the right eye also exhibited peripheral avascular, a dragged disk, but no retinal holes were detectable. The patient could not afford an ocular ultrasound.

A provisional diagnosis of bilateral bullous retinal detachment was made and topical treatment was instituted (gtt Mydracyl tds and gtt. Betnesol N qid)The patient was counsel and referred for further evaluation and management including fundal photography and fluoresceine angiography at either of the two institution mentioned earlier.

Unfortunately none of these patients could have detailed evaluation and necessary care at any of the referral centre due to reasons mentioned earlier and so they were permanently blind.

Discussion

Over the past decade, the pace of gene identification of the causes of inherited eye diseases has increased dramatically as the complete human genome information becoming available.¹⁰⁻¹⁴ Molecular genetic analysis establishes reliable clinical diagnostic criteria and improves the accuracy of diagnosis. We encountered a very uncommon presentation of retinal detachment in African patients in Nigeria.

The pathetic thing about the presentation is that the three siblings involved were either almost blind or having severe visual impairment before one of them sought for help, thereby prompting the remaining two to present for treatment of their visual ailments. The three were the remaining surviving children out of 5 children from the same parents. Both parents had some degree of visual impairment before they died but the nature was not known because they did not have any medical care then.

The diagnosis of bullous retinal detachment was made based on the complaint of painless visual loss, poor visual acuity not improved with pin hole, abnormal pupillary response, and findings of a bullous grayish white reflex on direct funduscopy and presence of bullous detachment of the retinal in both eyes on binocular indirect ophthalmoscopy. Lack of retinal breaks on careful indentation of the retinal was also documented. Ocular B-scan ultrasonography was only done in the first patient but not the remaining two due to lack of funds. Further detailed evaluation using fundal photography and Fluoroscein angiography was not done in any of these patients due to lack of appropriate facilities in the managing private hospital nor any other hospital in this region of the country. The only two centers in Nigeria where fundal photography and fundal fluoresceine angiography could be done were so far away and non of these patients could afford the very high cost of the investigations.

Extensive spectrums of conditions that cause bilateral bullous retinal detachments have been

documented.¹¹⁻¹³ They have been classified according to similar pathogenic mechanisms. Some of the causes are: Idiopathic (Coats disease, Central serous chorioretinopathy, Vogt-Koyanagi-Harada syndrome, retinoschisis, syphilis, scleritis, Sympathetic ophthalmia, other vasculitic entities e.g. rheumatoid arthritis, Wegener granulomatosis and other uveitic conditions e.g. toxoplasmosis, cytomegalovirus [CMV] retinitis; Congenital (Nanophthalmos, colobomas of the optic nerve and familial exudative vitreoretinopathy); Neoplastic (Choroidal melanoma, choroidal metastases, choroidal nevus, choroidal hemangioma, retinoblastoma, primary intraocular lymphoma); Iatrogenic (Excessive pan-photocoagulation, and scleral buckling) and Vascular factors (eclampsia, exudative age-related macular degeneration, chronic renal failure and hypertension.⁴⁻¹⁰

There is no doubt that some of these causes have a direct genetic origin. A few of these conditions which have familial tendencies include Vogt-Koyanagi-Harada syndrome, choroidal melanoma and exudative age related macular degeneration.^{1,4-6} Vogt-Koyanagi-Harada syndrome appears to be more common in Asians and Hispanics than in Caucasians. Choroidal melanoma is more common in Caucasians than in other races. Also, exudative age-related macular degeneration is more common in Caucasians than in other races and usually a disease of the elderly. The presentation in 3 siblings from a monogamous family suggested that the most likely mode of inheritance of those affected was autosomal dominant or probably X-linked, though we could not determine whether the condition was expressed in other generations, they denied family history of blindness and ocular surgery in other family members. These patients could have had a rhegmatogenous or exudative retinal detachment arising from an inheritable cause. The report of some form of poor vision among their parents supported the possibility of heredity in their ailments. The parents were likely to have had exudative age related macular degeneration which may be inherited.

Some researchers reported that a novel mutation in the FZD4 gene was identified in Japanese patients with familiar exudative vitreoretinopathy (FEVR).¹² Karyotype studies ruled out involvement of chromosome 11 or others at a gross level, but did not exclude them at the location for some genetic defect related to FEVR because single base changes and small deletions or insertions may be undetectable by the methods utilized.¹¹ FEVR often presents much earlier in early childhood with poor vision but all the three patients reported presented with visual loss in their middle age. Another previous report described the unusual occur-

rence of Coats'-like massive exudative retinal detachments in patients with X-linked retinoschisis.¹²⁻¹³ Total exudative detachment as a first presentation of von Hippel Lindau disease as also being reported.¹⁴ In Japan, a 14-year-old girl with Alport's syndrome who developed bilateral exudative retinal detachment in the macula had been reported.¹⁰

The study reported bilateral asymmetrical nature of visual involvement and differences in the age of the patients at presentation may be explained by the concept of variable penetrance and expression which often characterize autosomal dominant inheritance.

When diagnosed or highly suspected, RD requires an emergency ophthalmologic consultation for confirmation and treatment. This is particularly true for RDs that threaten the fovea or central vision.

Ideally, patients with RD should be referred to a retinal-vitreous specialist as soon as they are suspected. However, immediate retinal-vitreous specialist consultation is not necessary in all cases because many general ophthalmologists are capable of performing indirect ophthalmoscopy and determining the need for further intervention. Frequently, time is critical; however, the time frame is hours and not minutes, and many cases do not require emergency surgery. Inflammatory retinal detachments, for example, usually are treated medically. Acute retinal breaks should be surgically repaired within 24 hours if at all possible.

These patients were ignorant of the significance of early and prompt diagnosis of their visual impairment hence the delay in seeking for specialized and appropriate ophthalmic care. Delayed presentation to eye care specialists has been the pattern in most Nigerian with visual problems and this often contribute to poor visual outcome and blindness.

There are two major specialized regional Vitro-retinal centres in Nigeria at present. These are (1) Eye Foundation, a private hospital located in the heart of Lagos, South Western Nigeria and National Eye centre, a government referral centre located in Kaduna, Northern Nigeria. These two centers sub serve all patients who need specialized surgical intervention for RD throughout the country. Poverty is a major factor in the prevalence of blindness as one can deduce from these cases presents. None of the patients was able to seek for specialized care due to lack of funds.

In conclusion, loss of acuity and blindness is the most common complication of a retinal detachment. Delay in presentation and poverty were identified as major reasons why three siblings went blind in their middle

age. There is a need to subsidize the cost of health services to the common man and develop more specialized and accessible vitro-retinal centres in Nigeria, in order to reduce the scourge of blindness resulting from vitro-retinal disorders especially RD.

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