THREE SIBLINGS WITH PETERS' ANOMALY: Problems of Management in Nigeria

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SUMMARY
The cases of three children with Peters' anomaly anomaly are presented. The children are products of a consanguineous marriage. The clinical features and peculiar problems of managing these patients in a developing country are discussed. With the advent of keratoplasty, some cases, if operated early achieve good vision though the problem of concomitant glaucoma remains a drawback. In Nigeria, and indeed in most developing countries of sub-Saharan Africa, corneal transplantation is still in the embryonic stage.

Key words: Peters' anomaly, Nigeria

INTRODUCTION

Peters' anomaly is one of a group of anterior chamber cleavage syndromes, which exhibit varying degrees of abnormality. This group of iridocorneal dysgeneses includes posterior embryotoxon, Axenfeld's anomaly, Reigers anomaly and aniridia. Axenfeld's anomaly presents with strands of peripheral iris attached to a posterior embryotoxon; which consists of prominent Shwalbe's line displaced anteriorly. Glaucoma develops in these patients with Axenfeld's anomaly and can be present at birth.

Reiger's anomaly is dominant and usually bilateral with asymmetrical involvement of the eyes. It presents with both posterior embryotoxon and iris anomalies, which could be ectropion uveae, pseudopolycoria and corecttopia. Glaucoma presents in about 50% of affected children. Reiger's syndrome consists of Reiger's anomaly with dental and facial malformations.

Peters' anomaly is inherited as an autosomal dominant trait and is usually bilateral in 80% of patients. The bilateral type (as in these patients) is usually described as Peters' anomaly type II. Peters' anomaly is characterized by a central leucoma iris strand that crosses the anterior chamber from the iris. There is an iris collarette with a clear well positioned lens (type 1) or abnormally positioned lens with or without cataract (type II). The posterior stroma, Descemet's membrane and epithelium are also defective. Either type could have other associated ocular and/or systemic anomalies, however, these are more common with type II. It is thought to result from abnormal migration of neural crest cells, which accounts for the posterior endothelial and stromal defects seen and is corroborated by the observation of abnormally large stromal collagen fibrils of up to 360 - 600 Armstrong units in some patients. Another explanation for the posterior leucoma is an anterior subluxation of the lens, either prior to or after its full development, in either case interrupting the normal migration function of the endothelium. There is concomitant glaucoma in over 50% of patients. Other features of Peter's anomaly include anterior polar cataract, corecttopia, iris hypoplasia, microphthalmia, cornea plana and sclerocornea.

One of the earliest references to this disease was by Peters' A in 1906 and since then, various writers have described this group of diseases. We are not aware of any case reports within Africa as at the time of this write up.

CASE REPORT

In April 1993, a 30 year old Hausa woman presented in our clinic with three of her five children. The following history and clinical findings were obtained on all the three cases. For the purpose of this paper, we will call the three patients – Patients A, B and C. The mother gave the history in all the cases.

Patient A: Patient A is an 8 year-old girl born with an unusually white appearance of both eyes. The eyes were also large, and during the first 3 months of life the patient exhibited photophobia and showed excessive lacrimation. The parents also noticed that the child could not see, but medical help was not sought. The girl had no other systemic complaint and had been otherwise healthy from birth.

Patients A, B, and C with mother

She was the product of a full- term gestation and the mother had had a spontaneous vaginal delivery. The mother was healthy throughout the pregnancy, though she did not attend formal antenatal clinic. The child is the second in a family of five children; the last two also have similar eye problems. The parents who are from the same village are first cousins, though no other member of the family (distant or close) has had a similar eye disease (fig. 1).

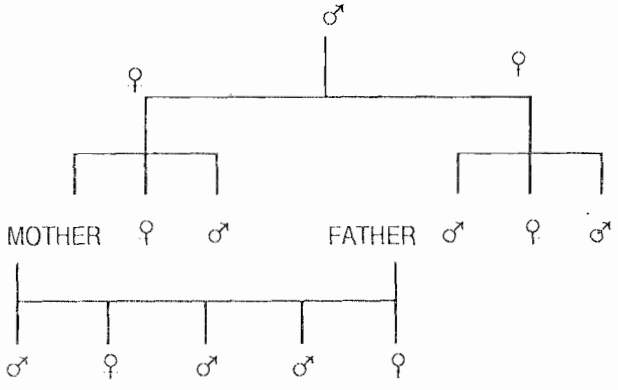
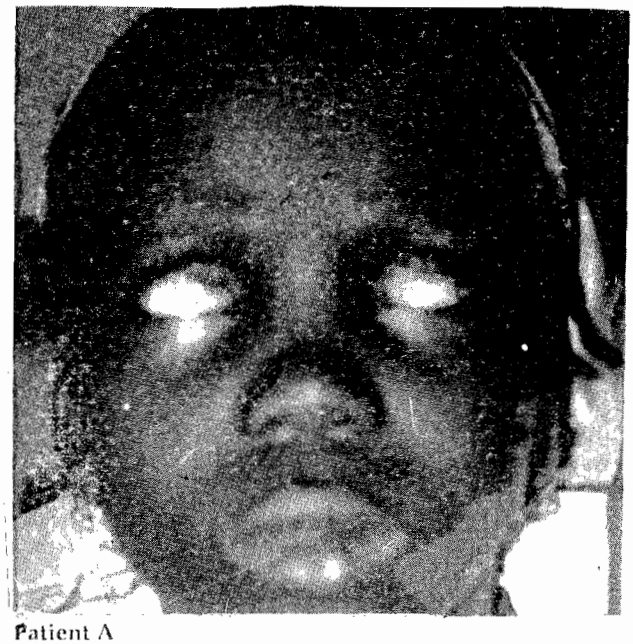


Figure 1. Family Pedigree Chart

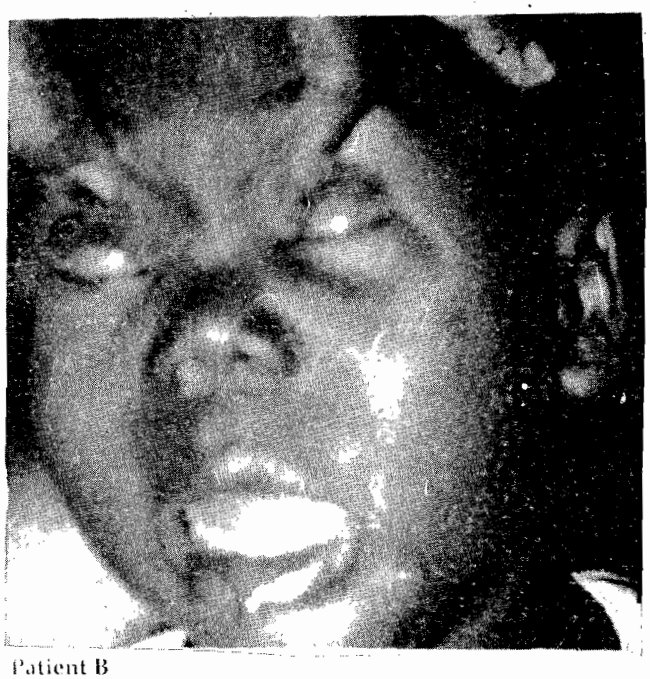
Examination revealed a healthy looking girl with stable vital signs. She had a visual acuity of light perception in both eyes with good light projection in all four quadrants of each eye. The corneal stroma in both eyes were certainly hazy. There was also disorganized epithelium as seen with a slit lamp at the 12 o'clock position. There was iris atrophy between the 12 o'clock and 2 o'clock positions in the right eye, and the pupil was irregular and reacted sluggishly to light. It also dilated poorly to mydriatics. The left pupil was dilated and reactive. Both eyes had dense cataracts. Intraocular pressure (measured with the Goldman applanation tonometer) was 2mmHg in the right eye and 4mmHg in the right and left eye respectively, while the horizontal corneal diameter was 13.5mm in both eyes. Systemic examination was non-contributory while basic laboratory tests showed an AA genotype and haemoglobin of 11.8g/dl. Urea and electrolytes as well as white blood cell (WBC) count were also normal.



Patient A

Patient B: Patient B is a 3 ½ year old boy who was also born with white large eyes. The parents noticed that the eyes were getting bigger with associated photophobia, lacrimation and occasional redness. Later, they noticed that the child could not see, but they still did not seek medical assistance. This child also had no other systemic

illness since birth. He is the fourth child of the family, was delivered at home vaginally after 9 months of gestation and there was no antenatal attendance though the mother claims to have been well all through the pregnancy.

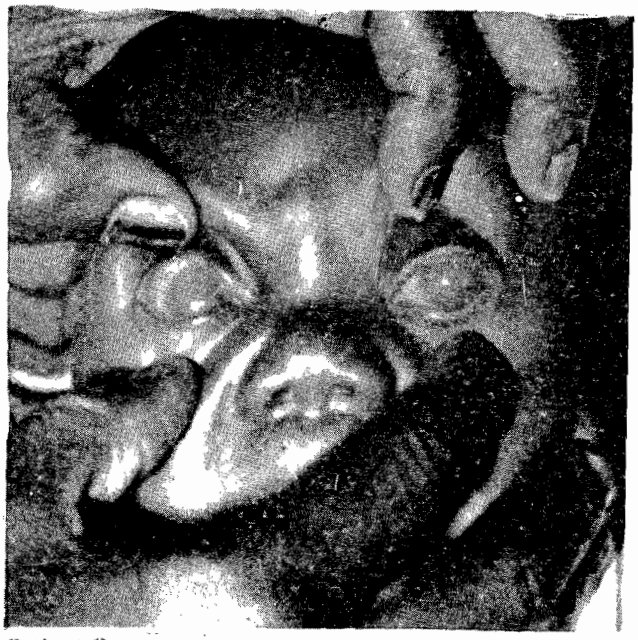


Patient B

Family history is as recorded for patient A. On examination, the child appeared to be healthy with normal temperature and pulse, and with no pallor. Ocular examination showed a visual acuity of light perception with good projection in all quadrants of both eyes. In both eyes, the cornea was completely opaque and bulging, with horizontal diameters of 15mm in the right eye and 15.5mm in the left eye. The opaque corneas made examination of the other eye structures impossible. The intraocular pressure, measured with the applanation tonometer, was 2mmHg for the right eye, while for the left, it was 0mmHg. Nothing significant was found on systemic examination while basic laboratory tests showed a haemoglobin of 10.8g/dl, genotype AA, normal urea and electrolytes as well as normal white blood cell count.

Patient C: Patient C is a girl aged 8 months. She is the 5th child of the family. She was also noticed to have a whitish appearance in both eyes after birth. There was no associated photophobia or lacrimation, but the parents noticed that this child too could not see. This prompted the mother to take all the three children to a general hospital close to their village, from where they were referred to our clinic.

Family history is as recorded above. Examination revealed an otherwise healthy child with stable vital signs. On ocular examination, it was found that the child was able to follow light projected in the different fields of gaze in both eyes. Examination under anaesthesia was done, which revealed bilateral completely opaque corneas with horizontal corneal diameters of 12.5mm. The intraocular pressure for the right eye was 12mmHg and for the left eye was 14mmHg. Corneal opacity made examination of the other eye structures impossible.



Patient C

The mother was also examined. She had clear corneas and a visual acuity (unaided) of 6/6. All other ocular structures were also normal in both eyes. Gonioscopy revealed a grade III open angle (Shaefer), and tension was 14mmHg in both eyes.

The father refused to present for similar examination. In a rural African setting, it is not unusual for the father to leave the responsibility of caring for the children to the mother; especially when she is viewed as being solely responsible for producing abnormal children, in this case blind children.

Based on the history and clinical examination of all the children (normal and affected) and their mother, a diagnosis of Peters' anomaly was made. No treatment could be offered however, as we do not have facilities for keratoplasty in Nigeria. Also the poor economic status of the family ruled out referral abroad.

DISCUSSION

Patients with autosomal recessive or dominant Peters' anomaly often have other associated ocular and/or systemic anomalies.[1, 2, 3, 4, 5] De Almeida et al.[6] in 1991 described two siblings, products of a consanguineous marriage who had Peters' anomaly plus syndrome. The

features which these patients had were short stature, brachydactyly and Peters' anomaly, and they agreed that their inheritance was autosomal recessive. In 1986, Green et al.[7] described a family with congenital cataract with microcornea and Peters' anomaly, it appeared to be variable expressions of a generalized anterior segment disorder inherited as an autosomal dominant condition. Also Salmon et al.[8] described autosomal dominant microcornea with cataract in a seventh generation family. Eight family members had microcornea and cataract and six had Peters' anomaly or sclerocornea. Ciba et al.[9] in 1985 reported a case of Peters' anomaly which was associated with ring 21 chromosomal abnormality. Central leucoma was the ocular finding in this case as the iris, lens, intraocular pressure and the posterior pole in the affected eyes were normal.

The three patients we have described are siblings, products of a consanguineous marriage (fig.1). None of the affected children had any systemic congenital malformation. Cataract, an often associated ocular anomaly,[10, 11, 12] was found in patient B, structures behind the cornea in patients A and C could not be seen however, due to the degree of corneal haziness. The inheritance pattern suggests autosomal recessive inheritance.

Our first patient had bilateral central corneal leucoma, abnormal irides and cataract. The other two had corneal leucoma with peripheral haziness. This prevented examination behind the cornea. They all had light perception vision, megalocornea and normal or abnormally low intraocular pressure.

Some patients with Peters' anomaly attain relatively good vision and clear cornea after keratoplasty, especially if done a few months after birth.[13, 14, 15] Erlick et al.[16] did a retrospective study of corneal transplantation in infants, children and young adults in the Toronto Hospital for Children between 1979 and 1998. In their series, which included at least three months of follow up, 7 of the 16 eyes with confirmed Peters' anomaly had clear corneas, as well as good vision. Eggink et al.[17] reported the case of a patient with bilateral Peters' anomaly whose corneas have remained clear one year after penetrating keratoplasty. This patient, however, had to have a second surgery for glaucoma. In the Parmley[15] study, with a mean follow-up period of 30 months, 26 grafts were done on 16 eyes with Peters' anomaly in 10 patients. Of these, 5 eyes had preoperative glaucoma which persisted postoperatively, 10 eyes developed glaucoma postoperatively, and the remaining one eye did not develop glaucoma at all and vision remained good. Some of these eyes have had graft rejection with subsequent regraft and glaucoma control, surgically or medically. Those that could not be controlled underwent cyclodestructive procedures. With these added procedures, 4 cases have maintained clear corneas and ambulatory vision.

CONCLUSION

The procedures mentioned above are ideal for our three patients who are all members of the same family, with the attendant economic and social burden to themselves and the other family members. The main challenge in Nigeria is the absence of facilities for corneal transplantation. The biggest obstacle to transplantation, however, is that of getting donors.

Although in Nigeria there is already an enabling legislation in place, archaic beliefs, religious taboos, and poor understanding continue to militate against cornea donation.[18] In the northern parts of the country, it is a great honour to bury the dead as soon as possible, which leaves no room to request for donor material. In the southern parts there are so many ceremonial rites that are observed during burial that people do not take kindly to any form of perceived mutilation of corpses. Some people believe that it is against their religious beliefs to donate any part of their body while others believe that the dead should be left alone.

Other inaccessible sources are in cases of trauma resulting in blind eyes with clear useful corneas. The victims are not able to comprehend how their own eyes could be useless to them and yet be useful in anyway to someone else. Cases of retinoblastoma that report early often have clear corneas. Although the eyes get enucleated, it is not advisable to use them as donor material.

For now, our only hope may lie in importing corneas from other countries, after all legislative and administrative difficulties have been sorted out. The few cases of cornea grafts that exist in the country are cases of rich patients who were able to go abroad for such a procedure or cases where sponsorship was provided by non-government organizations such as the Rotary International and Project Orbis.

Thus, in Nigeria and indeed in many developing countries, patients with corneal blindness (which is common) still have a long way to go before they can begin to nurse hopes of having their sight restored through modern technology.

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