SIGHT THREATENING RETINOPATHY IN AN EIGHT YEAR OLD NIGERIAN MALE WITH SICKLE CELL β° THALASSAEMIA: CASE REPORT

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Abstract
Sight threatening changes in the retina are a well-recognized complication of sickle cell disease (SCD). However they usually occur in older patients with Haemoglobin SC or Sβ+thal patterns. It is rarely found under the age of 20 years in patients who are Hb SS or Sβthal. This is a report of sight threatening retinopathy in an 8-year-old male Nigerian patient with Sβ°thal—one of the youngest reported cases to our knowledge. The patient had been diagnosed at birth and had his first ophthalmic examination done at 6 years of age when he developed an acute cerebral syndrome with transient blindness and hemiplegia. Retinal examination at that time was normal. In the subsequent years, he had several episodes of vaso-occlusive crisis including renal papillary necrosis. Two years later despite minimal visual symptoms, he had developed abnormal conjunctival vessels and bilateral retinopathy. He had high levels of Hb F and irreversibly sickled cells (ISC). Four years after treatment with Argon photocoagulation, he has developed no further neo-vascularization and his visual acuity remains normal with correction. As sight-threatening retinopathy could occur even in children, there is need for early detection and treatment in patients with SCD to prevent progression of lesions. Hence, a yearly examination is recommended for children, irrespective of age or electrophoretic pattern of patient. Another option would be to screen high-risk patients—those with frequent vaso-occlusive crisis, and high ISC counts.

Key words: sickle, cell, retinopathy, beta thalassaemia

Introduction
Sickle Cell Disease (SCD) is a genetic disorder affecting millions of people all over the world. In Nigeria it is the commonest genetic disorder with over two million people estimated to have the disease and a gene frequency of about 25% in adults. Approximately 100,000 babies in Nigeria are born with SCD each year. Sight threatening changes are a well recognized complication of this disorder and may affect any structure in the eye. However the part of the eye most often affected is the retina. It is thought that microvascular occlusion of the vessels of the retina occurs, leading to vasospasm especially in the periphery of the retina where maximal sickling occurs, resulting in retinal ischaemia. This leads to formation of new vessels (proliferative sickle cell retinopathy), which develop, from abnormal arteriolar venous communications, usually at the border of vascular and avascular lesions. The natural history of these abnormal vessels is that in a varying percentage of cases they lead to vitreous haemorrhage, which can result in tractional retinal detachment and visual loss. These abnormal vessels leak intravenous administered fluourescein and can progress rapidly. In some patients these lesions undergo spontaneous autoinfarction.

Patients who mostly develop proliferative sickle cell retinopathy (PSR) are those with the Hb electrophoretic pattern SC or Sβ thal. Those with Hb SS are less often affected, and retinopathy is rarely reported in those with Hb Sβ°thal, a variant that is clinically, haematologically and electrophoretically similar to Hb SS (except for the absence of Hb A). Sickle cell retinopathy usually occurs in patients over 20 years of age and as such many authorities advise that regular examinations be carried out in people with SCD from the age of 20 or 25years. This is a report of sight threatening retinopathy in an eight-year-old child with Hb Sβ°thal, and represents one of the youngest reported cases.

Case report
The patient was first seen when he was 6 years old at the ophthalmic unit of the Guinness Eye Hospital, Ahmadu Bello University Teaching Hospital, Kaduna, Nigeria. He was a known patient with Hb Sβ°thal having been diagnosed in the United Kingdom at birth. A few days prior to being seen at the unit, he
he had suffered an acute cerebral syndrome with complications of hemiplegia and blindness, which lasted for a few hours. Ophthalmic examination done at that time revealed a normal retina. Visual Acuity (V/A) was 6/18. The haematological parameters were haemoglobin A: 0, haemoglobin S: 79.8%, haemoglobin F: 16.1%, haemoglobin A: 4.1 %, haemogram 7.5 gm/dl, reticulocytes 0.1% and irreversibly sickled cells (ISC) 213/100.

In the following 2 years the patient had no visual complaints. However he had several episodes of vaso-occlusive crises of the limbs and once there was haematuria due to renal papillary necrosis.

At the age of 8 years, the ophthalmologists again saw him when he was sent for a routine ophthalmic examination (Table 1). At this time the child’s father was the only one who had noticed that the patient had a little bit of trouble with distance vision. Both direct and indirect ophthalmoscopy was carried out on the patient after papillary dilation with phenyl ephedrine 10% and cyclopentolate 1% topical preparation. The patient also had a slit lamp examination and because there was no facility for fluorescein angiography he was referred to the United Kingdom for this and for laser treatment. However in the United Kingdom, Fluorescein angiography was not carried out for fear of precipitating further vaso-occlusion in the patient. Argon laser application to the areas of chorio-retinal infarcts and abnormal vessels was performed. At the age of 9 years, further argon laser application to the remaining telangiectatic vessels in the periphery of both fundi was carried out. At the age of 13 years, post argon laser treatment examination showed no new vessel formation.

Table 1: Ophthalmic findings

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Finding</th>
</tr>
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<tbody>
<tr>
<td>Unaided visual acuity</td>
<td></td>
</tr>
<tr>
<td>Right eye</td>
<td>6/6</td>
</tr>
<tr>
<td>Left eye</td>
<td>6/24</td>
</tr>
<tr>
<td>Visual acuity with pinhole</td>
<td></td>
</tr>
<tr>
<td>Right eye</td>
<td>6/5</td>
</tr>
<tr>
<td>Left eye</td>
<td>6/9</td>
</tr>
<tr>
<td>Anterior segment (conjunctiva – both eyes)</td>
<td>Multiple short comma shaped capillary segments seemingly isolated from the vascular network. Few corkscrew shaped vessels found more in the lower conjunctiva.</td>
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<tr>
<td>Posterior segment (retina – both eyes)</td>
<td>Dilated and tortuous main vessels with appearance of new abnormal vessels in the retinal periphery, especially in the temporal quadrants. Numerous discrete dark circular chorio-retinal infarcts.</td>
</tr>
</tbody>
</table>

Discussion

This report of sight threatening retinopathy in Hb Sβ°thal has several unusual features. The patient developed chorio-retinal infarcts and new vessel formation over a period of 2 years. These findings are indicative of retinal ischaemia. Other workers have described signs of choroido-retinal atrophy in sickle cell disease (SCD), and the pathogenesis is thought to be due to retinal vessel occlusion. SCD is characterized by microvascular occlusion that occurs all over the body. This patient had previously experienced frequent episodes of vaso-occlusion in various parts of his body including the limbs, kidney and brain. In the eye microvascular occlusion of the retinal vessels resulted in ischaemic changes. The numerous chorio-retinal infarcts found in this patient may have been the result of this or of subsequent autoinfarction. Unfortunately, fear of precipitating further vaso-occlusion in this vulnerable patient prevented fluorescein examination.

This patient represents one of the youngest reported cases to our knowledge of proliferative sickle retinopathy (PSR) especially in a patient with Sβ°thal a condition in which the clinical manifestations are similar to those of Hb SS. PSR is rare in young children, and usually develops between the ages of 20-30 years. In a study of Nigerian children with sickle cell disease (Hb SS), Abiase found mainly conjunctiva signs in them. Only one child out of 92 had signs of neo-vascularization. In the Jamaican Cohort study, arteriovenous anastomoses were evident in only 3% of them, and proliferative retinopathy in none. Sight threatening retinopathy most commonly occurs in patients with the electrophoretic pattern Hb SC where the youngest reported case was 8 years. Hb Sβ°thal clinically resembles Hb SS, but the youngest reported patient with Hb SS was aged 13 years old. Retinopathy is rarely reported in patients with Sβ°thal. It could be patients with that this variant of SCD, which is clinically, haematologically and electrophoretically similar to Hb SS (except for the absence of Hb A), are often mistaken for Hb SS. In many areas, the equipment needed to distinguish the two types of patients is not available.

It is of note that the patient had minimal visual
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symptoms, which were not serious enough to warrant him seeking medical attention. Had a routine ophthalmic examination not been requested, it is possible that the lesions could have progressed to visual loss. Identification of those patients whose PSR likely to proceed to visual loss is of great importance but this has not been proven to be an easy task. Some studies suggest that a low level of Hb F, high level of irreversibly sickled cells (ISC) in patients is associated with PSR. This patient had a high level of ISC but he also had a high level of Hb F. In SCD, patients who have high levels of Hb F usually have milder clinical features. Surprisingly, one earlier study by Talbot et al. suggested that a high level of Hb F was significantly associated with retinal vessel closure in sickle cell retinopathy in Jamaican children. However in a follow up study, the opposite conclusion was reached.

The success of scatter and laser therapy in early small lesions of PSR is enough justification for lesions to be treated soon after their development. This would prevent progression to large lesions that require more complex therapy. In this patient laser therapy appears to have halted the progression of the lesions and the development of new ones. However there is a need to follow up this patient for a much longer period.

It is difficult to draw definite inferences from this case as to patients that are at high risk of developing PSR. A pointer might be the frequent episodes of vaso-occlusion, which indicate a high propensity for microvascular occlusion. There is thus a need for further studies that might be able to identify risk factors especially in children with frequent episodes of vaso-occlusion. This case demonstrates that unsuspected sight-threatening retinopathy can occur even in children with SCD. There is need for early detection and treatment in them to prevent progression of lesions. Hence, it is recommended that a yearly examination be carried out in children, irrespective of age or electrophoretic pattern. Where this is not feasible, another option would be to screen high-risk patients- those with frequent vaso-occlusive crisis, and high ISC counts.

References