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CASE REPORT

STROKE IN SICKLE BETA THALASSEMIA - A CASE REPORT HIGHLIGHTING PITFALLS IN MANAGEMENT IN A LOW PREVALENCE COUNTRY

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Abstract

Stroke in Sickle cell disease is a devastating complication. As Sickle cell disease is uncommon in Sri Lanka many clinicians may be unfamiliar with management of the disease and its complications. A 10-year-old boy presented with a transient ischaemic attack. He has had a silent large parietal infarct previously. He had been managed with transfusions and had undergone a splenectomy. However, he had not received hydroxyurea or undergone trans-cranial Doppler assessment.

Keywords – *Stroke, Sicket beta thalassemia*

Case Report

A 10-year-old boy from Monaragala, diagnosed at the age of 11 months with Sickle beta thalassaemia, presented to the Monaragala Base Hospital with vomiting and sudden onset weakness of his right upper limb, which lasted for 10-12 hours. The patient underwent a CT scan of the brain on admission and an MRI scan of the brain 6 days later. Neuroimaging revealed the presence of an old parietal infarction only (Figures 1 and 2). The patient did not have any significant neurological features referable to this radiological lesion, specifically there was no previous hemiplegia. A diagnosis of transient ischemic attack (TIA) was made and aspirin and dipyridamole were prescribed after the initial assessment.

The diagnosis of sickle beta thalassaemia was made at 11 months of age, when the patient presented with a febrile episode with severe anaemia. Regular monthly transfusions were commenced and mean pre-transfusion haemoglobin of around 8g/dL was maintained. Splenectomy had been done at the age of 4 years due to concerns about increasing transfusion requirement, when the spleen size was 4cm below the costal margin on palpation. The patient had never undergone a trans-cranial Doppler assessment and Hydroxyurea had not been used at any time.

In 2015, the authors stopped transfusions after re-assessment of his disease as it was felt that he did not need regular transfusions. The TIA occured 10 months after stopping transfusions.

His gene analysis was IVS1-5 (G-C)/HbS; αα/αα, Xmn +/- and HbS was located in the Arab Indian haplotype framework.

Discussion

HbS was first identified in Sri Lanka in 1962[1]. Most of the cases reported since are of patients with sickle beta thalassaemia.

The first case of homozygous HbSS was reported in 1991[2]. Due to the low prevalence of the HbS gene homozygous disease is rare in Sri Lanka.

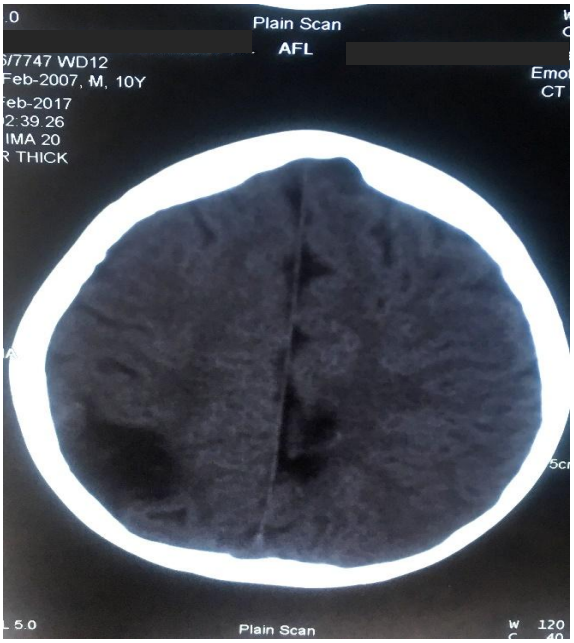

Figure 1
Non-contrast CT scan of the brain showing a right parietal infarction

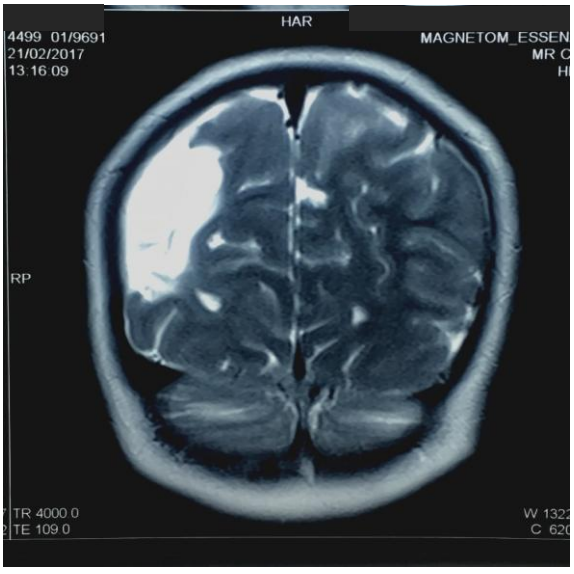

Figure 2
MRI of the brain showing a right parietal infarction

The well-known complication of stroke in sickle cell disease (SCD) has not been reported from Sri Lanka so far.

Approximately 11% of SCD patients have clinically apparent strokes before the age of 20 years[3]. The risk increases to 24% by the age of 45 years. The risk of stroke is highest during the first decade and it is most significant between ages 2 and 5 when it reaches 1% per year. A child with SCD has a stroke risk that is 333 times greater than that of a healthy child. Among those with SCD the risk of stroke is thought to be highest in those with HbSS and lowest in those with HbS β thalassaemia[3]. This however is not uniformly true. In a series from India of 110 adult patients with SCD (80 HbSS and 30 HbS β thalassaemia), all of who had the Arab Indian haplotype, 10 patients had developed strokes and 4 of them were those with HbS β thalassaemia[4].

Prevention and treatment of stokes in SCD is a challenge. Screening children with trans-cranial Doppler to identify abnormal flow over >200m/s and putting them on regular transfusion is the most successful method of prevention. This is however at the cost of iron overload and allo-immunisation. Hydroxyurea, which is remarkably effective in preventing joint crises and acute chest syndrome, may not be that effective in reducing strokes[3]. Once a stroke has occurred the patient is best managed with regular blood transfusions. Sudden cessation of transfusions has been shown to increase the risk of a recurrence of stroke[3].

Our patient's management highlights several important issues pointing to lack of exposure of most Sri Lankan clinicians to strokes in patients with SCD. The reason for commencing regular blood transfusions early in this patient is unclear. Regular transfusions can be justified if considered after risk assessment for stroke, which clearly was not the case here. The splenectomy that the child underwent again

suggests that he had been managed as a patient with thalassaemia rather than SCD. Massive spleens requiring splecentomy are encountered in the Indo Arabic haplotype SCD patients very occasionally, but this is unlikely to be the reason in this patient.

The attempt to stop transfusion was perhaps justifiable given the circumstances of management until then, but the vascular event may have been precipitated by this decision. Starting hydroxyurea prior to stopping transfusions may have prevented the TIA. Aspirin and dipyridamole used in ischaemic strokes in the general population have no defined place in SCD[3].

A recent island wide survey identified that there are over 50 patients with sickle cell syndromes in this country, the majority with HbS β thalassaemia (5). Acute care clinicians in Sri Lanka should be more familiar with management of sickle cell emergencies to prevent mismanagement, since there are likely to be more undiagnosed patients with Sickle beta thalassaemia in the community.

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