INTRODUCTION

Sturge-Weber syndrome (SWS) belongs to a group of disorders collectively known as the phakomatoses. It occurs in 1:50,000 to 1:60,000 births. SWS consist of congenital hamartomatous malformations that may affect the eye, skin, and central nervous system (CNS). SWS is referred to as ‘complete’ when both CNS and facial angiomata are present, and ‘incomplete’ when only the face or CNS is affected.

Clinical features in children include focal or generalized seizures, variable degree of learning disability and neurologic deficits such as hemiplegia and homonymous hemianopia. Seizures usually begin in infancy with profound seizure activity with resultant further neurologic and developmental deterioration seen in few cases. Early diagnosis is likely to be associated with a better outcome.

This case describes hemiplegia after a fall in a child with SWS.

CASE REPORT

A 3-year boy was brought to the emergency department in a large teaching hospital in north-eastern India from a remote area with acute-onset headache, vomiting and drowsiness. The symptoms started after an alleged fall from bed during a suspected seizure earlier in the day. He had previously been diagnosed with a seizure disorder and was treated with sodium valproate by a local physician over the preceding 2 years. There was also a history of associated developmental delay. No previous neuroimaging or electroencephalography (EEG) record was available.

Initial observations revealed heart rate of 108/minute, temperature of 36.9°C, respiratory rate of 28/minute and a central capillary refill time of < 2 seconds. He was confused and finding it difficult to verbalise and interact appropriately. The paediatric Glasgow Coma Scale score was 14/15. Neurological examination revealed a conscious child with right sided weakness of both limbs (power 4/5 on MRC scale). An erythematous patch (port-wine stain) measuring 4 x 5 cm was noted over the left forehead extending down to left upper eyelid which parents reported as being present from birth; bilateral bulging eyes were also noted (Figure 1). No obvious bony or soft tissue injury was noted in face or head. Examination of other systems was recorded as normal.

He was admitted to the paediatric ward for neurological observations following the head injury.

In view of the history, a non-contrast computed tomography (NCCT) scan of brain was done which showed a contusion with calcified lesion in the left parieto-occipital region. Basal cisterns were well seen and there was no mid-line shift. Neurosurgical opinion was sought; however, following a review no active intervention was considered necessary. The boy was conservatively managed with intravenous fluid for rehydration, anti-epileptic medication (continued on sodium valproate) and a dose of mannitol upon suspicion of raised intracranial pressure. Continuous monitoring of vital parameters and recording of the GCS...
were done. A repeat NCCT scan of the brain, 8 hours later, showed no change in the size of the cerebral contusion.

In the absence of any previous neuroimaging for comparison and non-progressive nature of the cerebral contusion, uncertainty remained as to whether the contusion was actually caused by the alleged fall or the lesion was present before the head injury occurred. The family was explained regarding this. A magnetic resonance imaging (MRI) scan of the brain done 5 days later which showed venous malformation at choroid plexus on the left side (Figure 2). The child was diagnosed with complete SWS. Conservative treatment continued and the patient gradually improved with an otherwise uneventful hospital stay. He was discharged home 7 days later with plan to continue sodium valproate and follow-up plan to review progress, seizure activity and developmental delay with paediatrician neurologist and ophthalmologist.

**DISCUSSION**

In this case, the episode of head trauma appeared to be co- incidental and did not cause the brain lesion as detected on NCCT scan. The case study also reflects the unexpected diagnostic challenges that may be faced by clinicians in resource-limited settings. Progressive characteristic calcifications in the external layers of the cerebral cortex underlying the angiomatosis associated with ipsilateral cortical atrophy frequently develop and progress with age, occasionally extending anteriorly to the frontal and temporal lobes.

Lack of development of superficial cortical veins, with persistent high flow to the deep venous system, is thought to lead toward engorgement and overloading of the deep cortical veins and venous hypertension. The leptomeningeal angioma and associated venous abnormality is considered to be the basis of cerebral changes seen in SWS. The less effective venous return system results in vascular stasis with chronic hypoxia, resulting in underlying cerebral atrophy, gliosis, and gyral calcification which produce the characteristic radiological features of SWS. Over time, thrombotic venous occlusions occur and aspirin therapy as an antiplatelet agent has been suggested. Spontaneous intracerebral hemorrhage in children with SWS leading to hemiplegia has been described in the literature, the suggested mechanism being sudden congestion leading to obstruction of the internal cerebral vein.

A case series from the UK with 5 children with SWS (4/5 children were already on aspirin) reported transient onset/worsening of hemiplegia following minor head injuries between ages of 10 months and 12 years; it resolved after a variable period of time. The authors also conducted a pilot questionnaire study sent to 135 families of children with SWS identified through the UK Sturge-Weber Foundation register, to identify the incidence of hemiplegia after head injury. Thirty-four returns (25%) were received where 21% (n=7) reported hemiplegia triggered by minor head injury, and one (2.9%) had generalized floppiness triggered by minor head injury. Permanent hemiplegia in SWS is often triggered by the first seizure and tends to worsen with recurring seizures, and transient neurological deficits have been documented in relation to recurrent episodes of hemiconvulsions and migrainous headaches. An improvement in seizure control could be followed by improvement in focal deficit, or at least prevent further events, and thus proper control of seizures in SWS is suggested.

Studies have also described transient hemiplegia and ischaemic stroke following minor head trauma in otherwise healthy children. The acute effects can be severe with dense hemiparesis, reduced level of consciousness, and evidence of unilateral cerebral oedema; but with full recovery being usual. However, permanent deficits with striato-capsular infarction can also occur following mild head trauma in previously healthy children.

Children with undiagnosed cerebral lesions in SWS can present as a diagnostic dilemma as highlighted in this case and more often when there are no previous
neuroimaging available to compare pre-existing intracranial lesions. It is important that neurosurgeons should remain aware of these rare syndromes as a wait-and-watch approach in an otherwise stable child may be the appropriate management strategy. MRI of brain is suggested before any neurosurgical intervention is carried out. Strict seizure control in children with SWS is likely to lead to a better neurological outcome.

REFERENCES


