Picture Story

A case of Klippel-Trenauny-Weber syndrome

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Introduction

Klippel-Trenauny-Weber syndrome (KTS) has a sporadic occurrence with an incidence of 1:100,000 births and comprises a triad of capillary malformations, varicosities and limb hypertrophy[1]. No definite diagnostic criterion or specific test exists for this condition and patients may not exhibit all the classical signs of this condition. This is the first Sri Lankan paediatric case of KTS reported in the literature.

Case report

An 8-year-old boy presented with 3 episodes of afebrile generalized tonic clonic seizures, each lasting 5 to 10 minutes with no drooling, urine/bowel incontinence or post-ictal drowsiness. He had no previous episodes or a family history of seizures. The child had delayed speech; only talking sentences at 7 years of age, and has a low intelligent quotient. On dermatological examination the child was found to have hyper-pigmented reddish irregular macular skin lesions over his neck and abdomen (Figures 1 and 2). The child had facial asymmetry (figure 3) with segmental left foot hypertrophy, disproportionate growth of left foot digits and marked leg length discrepancy (figure 4).

Figure 1: ***Hyper-pigmented reddish macular lesions over left aspect of face and neck***

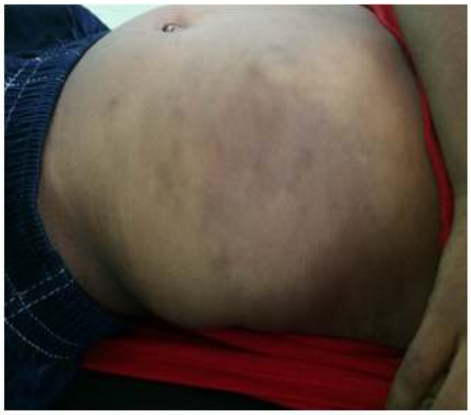

Figure 2: ***Hyper-pigmented reddish macular lesions over abdomen***

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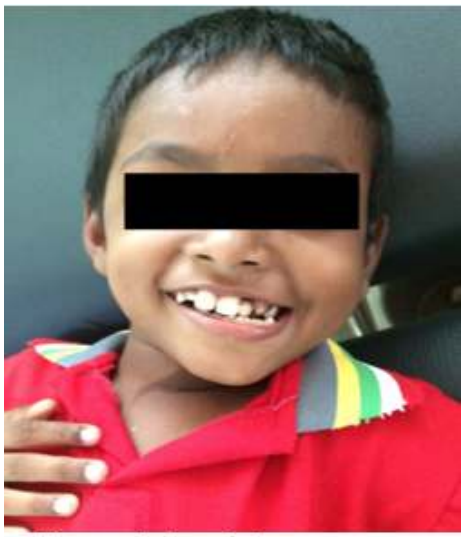

Figure 3: ***Facial asymmetry***

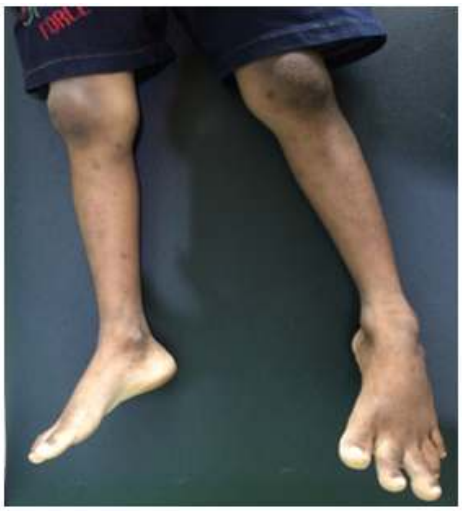

Figure 4: ***Leg length discrepancy with segmental left foot hypertrophy***

The asymmetrical limbs caused the child to have an abnormal gait. The child had no obvious varicosities. Both ear, nose and throat (ENT) and eye examination were normal, but cardiac examination revealed a grade 1 soft systolic murmur. All biochemical investigations were normal and ultrasound scan of abdomen found no obvious abnormalities. Contrast enhanced computed tomography (CECT) of the brain, done to rule out intracranial haemangiomas, which may have resulted in seizure activity, was normal.

Discussion

A study conducted by Jacob AG *et al* in 1998, reviewed 252 patients with KTS and found that 98% of patients exhibited capillary malformations, 72% had varicosities and 67% had limb hypertrophy[2]. Our patient had capillary malformations (port-wine stains) and limb hypertrophy but has not as yet exhibited varicosities/venous malformations. Seventy five percent of cases have hypertrophy of only one leg[3], as in our case. The incidence of seizures amongst those suffering from KTS has not been documented, but mental deficiency with seizures in patients with facial haemangiomas has been noted[3].

KTS is a disease requiring a multi-disciplinary team approach but some patients may not require any treatment at all. Orthopaedic intervention may be sought for extreme limb deformity or leg length discrepancy. Most patients with KTS can lead a normal life, but may suffer from complications such as pulmonary embolisms and bleeding.

Our patient was commenced on sodium valproate to manage the seizures. The child is doing well and has not suffered from any further seizures. He is followed up regularly at the hospital paediatric clinic.

References

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