Overlap of Klippel Trenaunay and Sturge Weber syndrome

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Abstract

Klippel Trenaunay syndrome and Sturge Weber syndrome are rare disorders of neurological and cutaneous signs of vascular origine. Klippel Trenaunay syndrome is triad of cutaneouse vascular malformation along with bony and soft tissue hypertrophy and venous varicosities. Sturge weber syndrome occurs sporadically with frequency of one in 50,000 characterised by meningofacial angiomatosis with cerebral calcification. We report a case with features suggestive of overlap between them for its rarity.

Introduction:

We describe patient with complex neurocutaneous syndrome of congenital vascular malformation, abnormalities of brain , venous varicosities and soft tissue hypertrophy of lip and limbs of affected left side. According to eponymous classification scheme ,the patient can be assigned to two different clinical entities- Klippel Trenaunay and Sturge Weber syndrome. Association between these two syndromes is very rare. It has been recognized in the past and only 40 cases of such combination have now been published all over world according to our knowledge.

Case report :

28 years old male patient resident of Amravati, Maharashtra, was referred to our department for Neavus Flammens on whole face , left side of chest ,abdomen, back, left upper and lower limb(fig.2).Further we noticed his stepping gate, and obvious increase in axial length and circumference of left upper and lower limb(Fig 1&2).

At birth his family members noticed that he had several areas of redness of skin. No further data available regarding birth history .History of grand mal type of epilepsy was present since he was 2 years old.

Physical examination revealed congestion of eyes and soft tissue hypertrophy of lower lip (fig 4).Left hand was longer than right by 2 cm and had larger circumference. Left leg was larger than right leg by 2.5cm and was larger in circumference (Fig 1 &2). Varicose veins were present on lateral side of left leg (figure 1).Neurological examination revealed no obvious defects.

On further investigations, CT scan of brain revealed serpiginous calcification(Fig.3) noted at left side of occipital lobe, suggestive of Sturge Weber syndrome. CT-angiography revealed no abnormality . Slit lamp examination of eye revealed reddish hue of iris and conjunctiva. Fundus examination and intra ocular pressure was normal. All other routine investigations revealed no significant abnormalities.

Diagnosis of Klippel Trenaunay syndrome and Sturge Weber syndrome was made on basis of physical examination and radiological evaluation.

Discussion:

Sturge Weber syndrome is a mesodermal phacomatosis characterised by port wine stain covering face and cranium supplied by 1st division of trigeminal nerve and calcification of cerebral hemisphere. Other associated features are seizures, glaucoma, neurodevelopmental delay. Neuroradiological examination shows leptomeningeal enhancement along with signs of cortical atrophy and calcification. In our patient CNS abnormality on CTscan along with port wine stain ,seizures pointed towards Sturge Weber syndrome. But lip and left sided limb hypertrophy and varicouse veins could not be explained. Klippel Trenaunay syndrome is rare entity with triad of port wine stain,varicosities and soft tissue hypertrophy which were present in our patient.

Thus the patient we described above has complex congenital syndrome of vascular malformations with

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internal abnormality of brain combined with soft tissue swelling. According to eponymous classification, patient would meet criteria for both the syndromes described above. Hence the use of eponymous classification is of little use for complex abnormalities as described here .Also it is very important to carefully map the symptoms and their extent in patients suffering from vascular malformations. Further this cases illustrates the necessity of carefull follow op of these vascular malformations by a multidisciplinary team in order to prevent complications.

Key Messages:

In a patient of congenital vascular malformation, one should always search for all possible associated clinical abnormalities by keeping in mind overlap phenomenon such as in our case. This will help in making early diagnosis and prompt management of any serious complication.

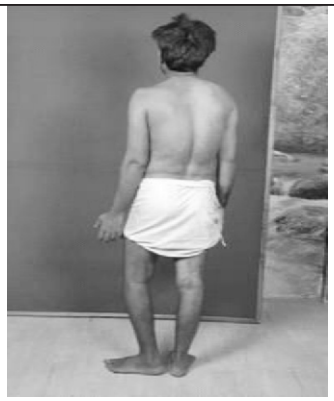

Fig1. posterior view of patient showing portwine stain on one half of body with limb length discrepancy

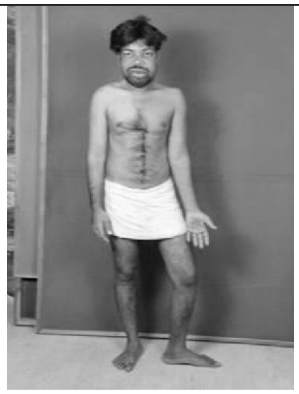

Fig.2 anterior view of patient showing portwine stain on one half of body with limb length discrepancy and also varicosities over lt leg (lateral surface)

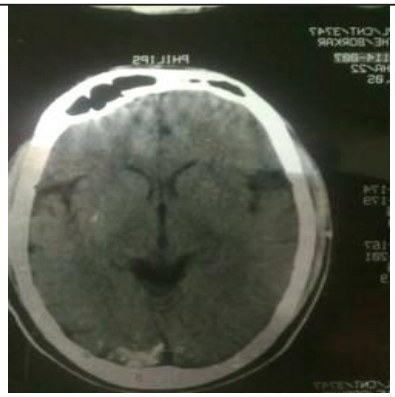

Fig3. CT BRAIN PLAIN of same patient showing serpigenous calcification in left occipital lobe

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