Sturge Weber Syndrome Overlapping with Klippel Trenaunay Syndrome

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Abstract

Sturge Weber syndrome (SWS) is a mesodermal phakomatosis characterized by meningo-facial angiomas with cerebral calcification. Klippel Trenaunay syndrome (KTS) is another very rare type of phakomatosis with cutaneous angiomas, varicose veins and enlargement of soft tissue or bones. Overlap between SWS & KTS is very rarely encountered. We report a three and half year old girl with overlapping features of both SWS and KTS.

Key words: Sturge Weber syndrome, Port wine stain, Hemihypertrophy, Klippel Trenaunay syndrome.

Introduction

The phakomatoses are disorders characterized by hamartomas of the skin, eye, CNS and viscera[1]. Sturge Weber syndrome (SWS) is a mesodermal phakomatosis characterized by a port-wine haemangioma on the upper part of the face over the distribution of trigeminal nerve, choroidal vascular lesions associated with glaucoma, early onset seizure, meningeal calcification and neurologic deterioration[2]. Klippel Trenaunay syndrome (KTS) is characterized by a triad of port-wine stain, varicose veins, and bony and soft tissue hypertrophy involving an extremity[3]. Both the syndromes occur sporadically, but very rarely patients with overlapping features of SWS and KTS have been reported[4-6]. Here we describe a case with features suggestive of overlap between them with some other rare associations.

The Case

A three and half year old female child, born out of non-consanguineous marriage, presented with first episode of left sided focal convulsion with secondary generalization. Perinatal and developmental history was normal. Convulsion was controlled with multiple anticonvulsants and other supportive measures.

Physical examination revealed extensive port-wine staining over right side of her face, with extension to opposite side at the level of chin (Figure 1A). The port wine stain also descended on to the neck, chest, part of the trunk on both sides and the right leg (Figure 1B). Further, the right half of the hard and soft palate was found to be reddish in colour due to the presence of port wine stain. There was obvious discrepancy (>5%) in length and circumference between the two sides of both the upper & lower limbs (Figure 1C). No varicosities were noted. On ophthalmological examination, there was right-sided megalocornea and features suggestive of glaucoma on the same side.

The routine blood parameters were normal. The computerized tomography (CT) scan of brain (Figure 2A) showed serpentine calcifications in the right cerebral hemisphere with atrophy of the brain parenchyma. Electroencephalography (EEG) was suggestive of cerebral dysrhythmia. Ultrasound of abdomen (Figure 2B) was done, which pointed out increased volume of the right kidney (48.63cc) compared to the left one (37.52cc); no other abnormality was detected.

According to the results of physical examinations and imaging study, the patient was diagnosed to have SWS overlapping with KTS. She was discharged home on oral carbamazepine and is being followed up on an outpatient basis.

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How to cite this article ?
Sarkar S, Patra C, Dey C, Dasgupta MK, Kundu TK. Sturge Weber Syndrome Overlapping with Klippel Trenaunay Syndrome. J Nepal Paediatr Soc 2013;33(2):147-149.

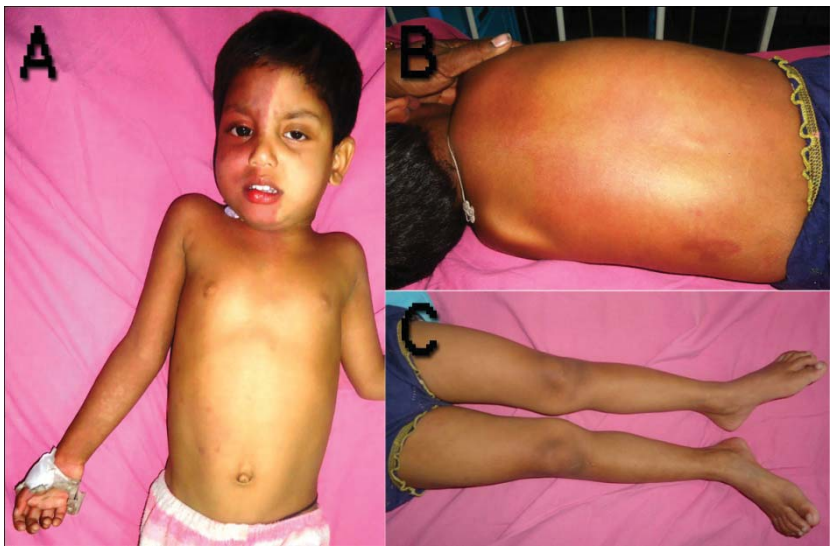

Fig 1: **(A)** Extensive port-wine staining over right side of her face, with extension to opposite side at the level of chin
(B) Port wine stain on the left part of trunk
(C) Hemihypertrophy of right lower limb

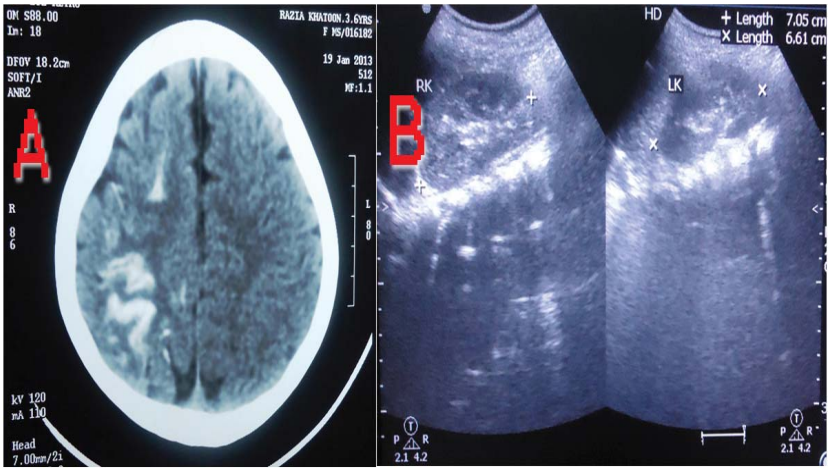

Fig 2: **(A)** Serpentine calcifications in the right cerebral hemisphere with atrophy of the brain parenchyma.
(B) Increased volume of the right kidney compared to the left one.

Discussion

Sturge Weber Syndrome occurs sporadically with a frequency of approximately 1 in 50,000[7]. Port-wine stains typically in the ophthalmic and maxillary divisions of the trigeminal nerve, ocular involvement and cortical vein dysplasia with consequent alterations in the brain are considered the three major signs of Sturge-Weber syndrome[2]. Port wine stain generally does not cross the midline, but in our case it had.

Klippel Trenaunay syndrome is a triad of cutaneous angiomatosis, varicose veins and enlargement of soft tissue as described first by Klippel & Trenaunay in 1900[4]. Associated A-V malformation causing high output heart failure was first described by Parker & Weber, when present is known as Klippel Trenaunay Weber syndrome. Not all the three abnormalities need to be present for the diagnosis of the syndrome. The lower limb is involved in about 95% of patients while upper limb involvement is seen in only 5%. Venous varicosities develop in about 80% of the patients[2] by 12 years of age[8]. In our case, both upper & lower limbs were affected and varicosities were yet to develop. Different viscera like bladder or intestine may be involved by hemagioma leading to hematuria or hematochezia respectively. But, associated hemihypertrophy of kidney without any renal haemangioma like in our patient has not been reported in literature till date.

In the present case, port wine stain over face and other parts of the body, calcification in the brain, seizures and right sided hemihypertrophy indicates presence of SWS co- existing with KTS. Cases with features of this overlap are seen rarely[5,6,9]. The exact pathogeneses of these syndromes are still not clear [9]. But these different syndromes and their overlap can be linked by postulating an insult to neural crest occurring at the time, when these structures are in close proximity during development[10].

It was Schnyder et al, who first bridged the gaps between these three syndromes, namely Klippel Trenaunay, Parkes Weber and Sturge Weber[11]. Baskerville et al[12] pointed out that the persistence of the embryonic vascular network which usually regresses in the developing limb bud, can increase skin blood flow and temperature leading to increase in the size and number of veins. This may result in increased bone growth and ectasia of superficial veins. Moreover, Furukawa et al and Gottron considered the two disorders as differing only in the location of involvement[13,14]. There is no specific curative treatment for this combined disorder. The management is directed towards the underlying local and systemic anomalies.

Conclusion

Our patient has features suggestive of both SWS and KTS. Though rare, a few cases have been reported with overlapping features of these two syndromes. But uniqueness of our case is that, though right side is affected in both SWS & KTS, port wine stain has also been extended to left side in face and trunk. There was also unilateral hypertrophic kidney associated with the overlap syndrome which to the best of our knowledge has not been reported till date.

References

1. Smirniotopoulos JG, Murphy FM. The phakomatoses. *AJNR* 1992;13:725-46.
2. Sturge WA. A case of partial epilepsy apparently due to a lesion of one of the vasomotor centers of the brain. *Trans Clin Soc Lond* 1879;12:162–67.
3. Janniger CK, Ortonne JP, Wells MJ, Libow LF, Gelfand JM, Elston DM. Klippel-Trenaunay-Weber Syndrome [online]. 2012. [cited 2012 Sep 17]. Available from: http://reference.medscape.com/article/1084257-overview.
4. Kiley MA, Oxbury JM, Coley SC. Intracranial hypertension in Sturge- Weber/Klippel-Trenaunay-Weber overlap syndrome due to impairment of cerebral venous outflow. *J Clin Neurosci* 2002;9:330-33.
5. Deutsch J, Weissenbacher G, Widhalm K, Wolf G, Barsegar B. Combination of the syndrome of the Sturge–Weber and the syndrome of Klippel-Trenaunay. *Klin Pediatr* 1976;188:464–71.
6. Lee CW, Choi DY, Oh YG, Yoon HS, Kim JD. An Infantile Case of Sturge-Weber Syndrome in Association with Klippel-Trenaunay-Weber Syndrome and Phakomatosis Pigmentovascularis. *J Korean Med Sci* 2005;20:1082–4.
7. Haslam Robert HA. Sturge-Weber disease. In: Behrman RE, Kliegman RM, Jenson HB, Stanton BF, editors. *Nelson's Textbook of Pediatrics*. 18th ed. Philadelphia: Saunders; 2008. p. 2487.
8. Verhelst H, Van Coster R. Neuroradiologic findings in a young patient characteristics of Sturge-Weber syndrome and Klippel-Trenaunay syndrome. *J Child Neurol* 2005;20:911-13.
9. Bonse G. Rontgenbetunde ber emer phakomatose (Sturge Weeber Kombinert nut Klippel-Trenaunay). *Fortscher Roentgenstr* 1951;74:727-29.
10. Cushing H, Bailey P. Tumors arising from the blood vessels of the brain: angiomatous malformations and hemangioblastomas. Springfield, Ill.:C Thomas; 1928.
11. Schnyder UW, Landolt E, Martz G. Syndrome de Klippel–Trenaunay avec colombe irien atypique. *J Genet Hum* 1956;5:1-8.
12. Baskerville P. A. et al. The etilogy of the Klippel Trenaunay syndrome. *Ann Surg* 1985;202:624-27.
13. Furukawa T. et al. Sturge Weber and Klippel Trenaunay syndrome with nevus of Ota&Ito. *Arch Dermatolr* 1970;102:640-45.
14. Gottron HA., Schnyder UW. Verenbung von Hautkrankheiten. In: Handbuch der hautund Geschlechtskrankheiten. volumee 7. Berlin: Springer Verlag; 1966.