STURGE- WEBER SYNDROME

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Sturge-Weber syndrome is an uncommon congenital disorder characterized by venous angiomas of leptomeninges over cerebral cortex, ipsilateral portwine nevus in trigeminal nerve distribution and frequent angiomatous involvement of eyeball, mouth and nasal mucosa. It is caused by faulty development of certain mesodermal and ectodermal elements closley approximated in the brain and meninges during early foetal stages (Peterman et al 1958).

In 1860 Schirmer described a patient with a portwine facial nevus associated with a homolateral glaucoma. Sturge (1879) described a case in a young girl who had extensive portwine nevus over the right side of the head and enlargement of right eye with vascular changes in choroid. He speculated that the convulsions were due to a simiar vascular lesion on the surface of the brain Kalischer (1997) in an autopsy of a similar case described a meningeal angioma on same side of the brain as facial nevus. Weber (1922) gave the characteristic roentgenographic finding of opaque double contoured sinusoidal lines following the convolutions of brain in the occipital and parietal region. Since then this disrder has come to be known as Sturge-Weber syndrome. The occular manifestations were first described by Kirby (1951) in the form of glaucoma and angioma of choroid and he pointed out that this disorder as well as other neurocutaneous syndromes shoud be classified as phakomatoses (mother sports) as suggested by Von-der-Hoeve Only 266 cases of this syndrome including two from India by Dutta Roy and Dubey (1963) and Chakraborty (1964) have been published (Chakraborty, 1964).

CASE REPORT

A. H. 9 years old male patient was born full term with a portwine nevus on right side of face. The mile stones were normal during first year of life. After one year he started with convulsions which were generalized and bilateral. The convulsions were typical of grandmal fits. The fits increased in intensity and frequency with age and he sustained frequent but minor injuries and burns during the fits. The fits have been only partially controlled by phenobarbitone and dilantin sodium. Weakness of left side of body was noticed when he started walking. Patient had shown behavioural problems and became violant on minor excuses. He had a subnormal intelligence though his memory was comparatively good. He talked some times irrelevant. During third year he developed a divergent squint of right eye and eye looked bigger in size. Patient has three sisters. All are well Parents are first cousins.

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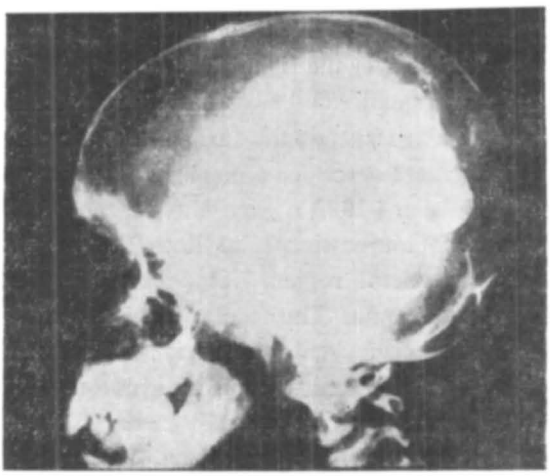
Examination revealed. 1. a portwine nevus on right side of facc in ophthalmic and maxillary distribution. 2. Cavernous angioma of lower lip and adjacent part of mucosa of right cheek, an angioma behind 2nd lower molar tooth on right side. 3. hemiparesis on left side of body of upper motor neurone type. 4. occular manifestations in the form of buphthalmos, circumcorneal injection of vessels, cyclitic membrane behind the pupil, divergent squint in right eye with loss of vision except for slight light perception.

Investigations. 1. Blood, urine and stool examination, all normal. 2. X-ray Skull: Calcification is seen on right side of the parietal region extending towards the temporal region (Fig. 1). Appearance is very typical of Sturge–Weber syndrome.



DISCUSSION

It has been laid down that any two of the following findings usually suffice for the diagnosis (Beeson and Mc Dermott, 1964). 1. Facial portwine nevus. 2. Contralateral convulsions. 3. Contralateral Hemiparesis or hemiplegia. 4. Mental defectiveness. 5. Characteristic roentgen changes. The present case fulfilling all the criteria is therefore a classical patient of Sturge–Weber syndrome.

The vascular lesion of portwine type does not change its relative size with advancing age and roughly corresponds to the trigeminal distribution. However it maycross the midline and may extend to the other regions of the body (Beeson and Mc Dermott, 1964). Unilateral facial lesion usually occurs on the same side as leptomeningeal angioma but has also been reported on the opposite side and rarely when facial lesion is bilateral, intracranial lesion either is on the side of greatest involvement or is bilateral (Beeson and Mc Dermott, 1964).

Evidence of cerebral involvement usually appears in infancy but may be delayed to adult life. It usually results in convulsions (50 – 90 %), mental defectiveness (50%), hemiparesis or hemiplegia (33%) and not infrequently

behavioural problems, homonymous hemianopia or cortical blindness and all these signs are bilateral in about 10% cases (Beeson and Mc Dermott, 1964).

Most important eye changes are buphthalmos and glaucoma which may progress to optic atrophy and blindness. Cataract occurs occasionally and glioma of retina has been reported (Beeson and Mc Dermott, 1964).

Krabbe (1934) demonstrated that roentgenographic shadows represented calcium deposits in the outer layers of the cerebral cortex rather than in the walls of blood vessels as had been thought by previous workers.

SUMMARY

A case of Sturge-Weber syndrome showing portwine nevus, cavernous angioma, convulsions, hemiparesis, mental and behavioural changes, occular manifestatons and characteristic roentgenographic findings is reported.

REFERENCES

1. Beeson, P. W. and Mc Dermott, W., Cecil-Loeb-text book of medicine, 11th. Ed. W.B. Saunders Co Philadelphia and London 1964, pp. 515.
2. Chakraborty, G., Ind Pediet., 1 :279, 1964.
3. Dutta Roy, S. K. and Dubey, M. P., Ind. J. Pediat., 30:204, 1963.
4. Kalischer, S., Berl. Klin. Wchnschr., 34 : 1059, 1897 (Quoted by 7).
5. Kirby, T. J , Am. J. Med. Sc., 222., 227, 1951 (Quoted by 7).
6. Krabbe, H. K., Arch. Neurol and Psychopath, 32 : 737, 1934 (Quoted by 7).
7. Peterman, A F. ; Hayles, A. B. ; Dockerty, M. D. and Love, J. G.; JAMA 167 : 2169, 1958.
8. Schirmer, R., Arch. Ophth , 7 : 119, 1860.
9. Sturge, W. A., Tr. clin. soc London, 12 ; 162, 1879 (Quoted by 7).
10. Weber, F. P., J. Neurol and Psychopath., 3 : 134, 1922 (Quoted by 7).