Case Report

A Case of Sturge Weber Disease with Portal Hypertension

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Sturge Weber Syndrome is a rare congenital disorder which is characterized by presence of angiomas that involves the face most commonly over the distribution of ophthalmic and maxillary division of the trigeminal nerve and the leptomeninges leading to central nervous system malformation. The disease is characterized by port wine nevus over the face, focal seizures and glaucoma. The disease results from the malformation of the cerebral vasculature which leads to hypoxia in the cortex causing neurological damage.

Portal venous malformations and portal cavernoma are quite rare features of Sturge Weber Syndrome which can lead development of portal hypertension and its complications like gastrointestinal bleeding and hypersplenism.

Here, we shall discuss about a case of a female young mentally retarded patient who presented to us with seizure and anemia

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Sturge Weber Syndrome belongs to congenital neurocutaneous syndromes characterized by abnormal growth of ectodermal tissue, producing distinct skin lesions and malformations and tumours of the nervous system[1]. The hallmark of the disease is a characteristic port wine nevus over the face supplied by the ophthalmic and maxillary divisions of the trigeminal nerve. The facial nevus is usually unilateral[2], rarely bilateral[3]. Associated neurological findings include seizures, intellectual sub normality, visual loss due to glaucoma, focal signs like hemiparesis and sometimes soft tissue hypertrophy[4]. Association with intestinal hemorrhage is also possible[5].

Portal hypertension is an extremely rare condition associated with the syndrome. A Spanish report presented a case of recurrent hematemesis in a patient of Sturge Weber Syndrome from fundal varices as a result of portal hypertension[6].

Case Report

A 16-year-old non diabetic non hypertensive female presented to us with complaints of recurrent episodes of jerky movements suggestive of seizure since 2 years of age, poor scholastic performance less social interactions, failure to attain menarche and generalized weakness for 6 months. The episodes of seizures began when she was 2 years old. Initially one episode occurred every 2-3 months. The abnormal movements used to start in the left side of the body but later used to involve the whole body with loss of consciousness. The seizures became more frequent subsequently and she started having seizures even on medications. There was also complaint of less social interactions. Her scholastic performance was poor from early childhood and stopped going to school after a couple of years. She could not read and write. There were also complaints of reduced feeding for past 5 to 6 months with feeling of generalized weakness and two episodes of passing black coloured stool. No history of vomiting, pain abdomen, jaundice. She did not start menstruating. History of a reddish patch over the right forehead and upper eyelid was there. Apart from the seizure and features suggestive of mental retardation, no significant past history was present. She has 3 other sisters in her family. They are all healthy without any mental retardations or seizures.

Editor's Comment :

- As portal hypertension is a rare manifestation of a rarer disease it may be overlooked.
- High index of suspicion is needed to intervene early.

On examination, the patient was alert, conscious and did not interact much. She could talk a few sentences and could recognize her sister. She was poorly nourished. On facial examination apart from temporal hollowness and malar prominence a distinct maroon colored patch was seen over her right forehead, eyebrows upper and lower eyelids extending up to the nasal side of right face. Significant pallor was found but her vitals were normal. Apart from the higher mental function derangements as described above no other neurological abnormalities were found. On abdominal examination, spleen was palpable, non-tender about 5 cm below the left costal margin. It was firm in consistency, had a smooth surface with well-defined margin. Liver was non palpable and no other abnormalities were found.

After history and clinical examination, we kept a working diagnosis of Sturge Webber Syndrome. But we were also dealing with the issues of anemia,

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splenomegaly and primary amenorrhea.

On complete blood count we found significant decline in all the cell lines with hypochromia and microcytosis. The reticulocyte count was normal. We found hypoalbuminemia in biochemical investigation. Abdominal ultrasonography suggested an enlarged spleen (17cm).

These findings were suggestive of pancytopenia possibly due to hypersplenism and portal hypertension was kept as a possibility of hypersplenism.

An iron profile showed low serum iron and transferr in saturation. An HPLC ruled out Thalassemia and vitamin B12 and folate levels were within normal limit. Further we did a bone marrow aspiration cytology which suggested reactive marrow with erythroid hyperplasia. MP/MPDA and an rK39 ruled out any remote possibilities of Kala Azar and Malaria. Meanwhile, on NCCT brain dense gyral pattern of calcification was seen in the right occipito-temporal cortex with dilatation of sulci – a clearly suggesting Sturge – Weber Syndrome. As per advice of endocrinologist serum FSH/LH were done and the results were normal. At this point, we assumed that the primary amenorrhea was probably a consequence of pancytopenia. Keeping portal hypertension in mind we did Prothrombin time which came to be slightly deranged, ie, 4 secs over the control with an INR of 1.38. An MRI brain was done for the primary disease and apart from atrophy in the right occipeto-temporal region a curious finding of signal change (hyperintensity inT1) was noticed in Basal Ganglia region bilaterally which further suggested liver involvement. Due to all these suggestions, we promptly did an upper G.I. endoscopy where Grade II esophageal varices were found. We also did a CECT abdomen which showed only splenomegaly but did not comment on portal vein. CECT whole abdomen showed a normal uterus and ovaries, thus ruled out any structural cause of amenorrhea. For proper visualization of the portal veins we finally went for an MR portovenography and expert opinion of a radiologist was sought. The imaging was suggestive of portal vein dilation with possibility of portal cavernoma. According to the radiologist, the dilation had probably extended deeper inside the hepatic parenchyma. The patient was treated conservatively, antiepileptics were given to control her seizures and was discharged in a hemodynamically stable state.She was advised to attend gastro medicine and gastro surgery for opinion regarding management of portal cavernoma and was asked to follow up in our department (Figs 1-4).

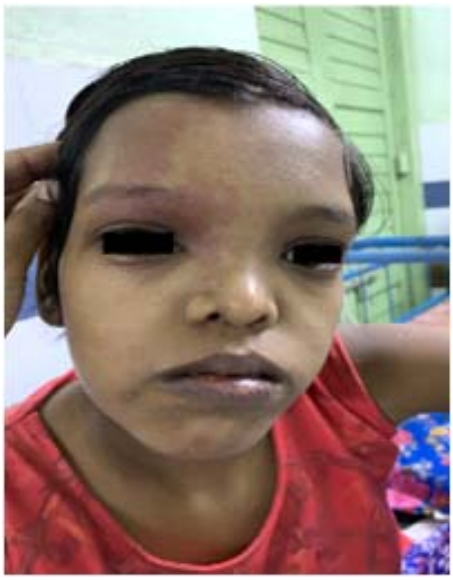

Fig 1 — Patient

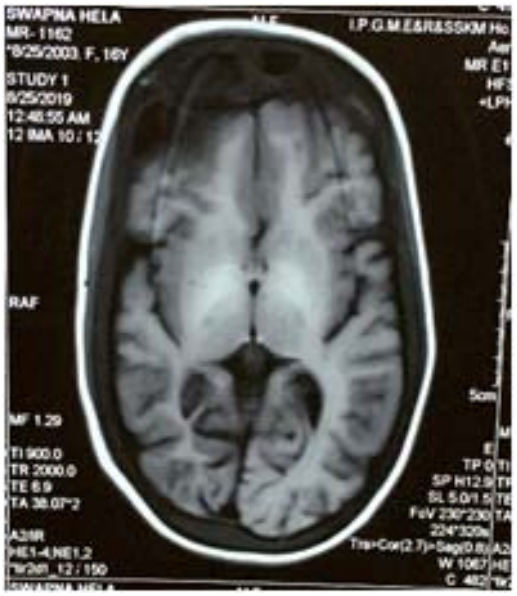

Fig 2 — MRI showing Atrophy and Basal Ganglia Hyperintensity

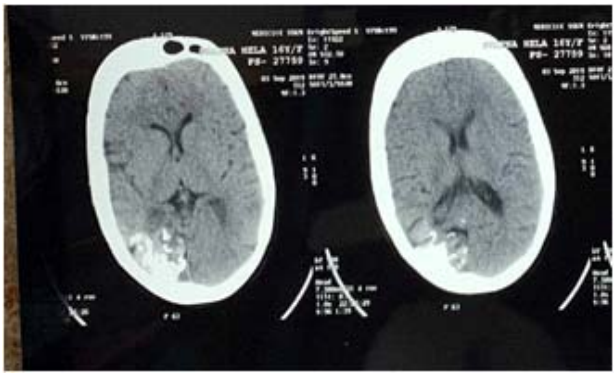

Fig 3 — GyriformCalcification in Right Occipeto-temporal Region

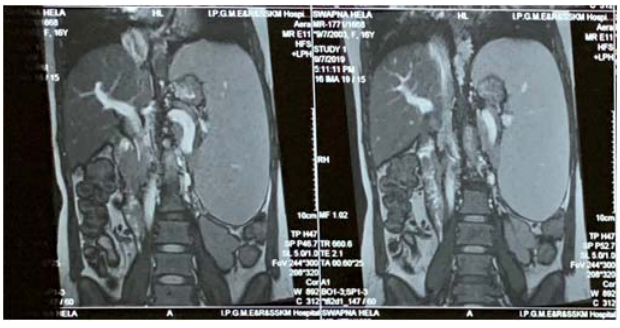

Fig 4 — MR Portovenography Showing Dilated Portal Vein

Investigations

Complete Blood Counts :

	5/9/19	2/9/19	28/8/19	24/8/19	26/8/19	23/8/19
Hb%	4.5	4.4	4.9	4.9	4.9	5.1
PCV	18.1	17.6	19.9	20.6	19.9	20.5
MCV	67.3	64.7	66	68.9	67.5	
MCH	16.7	16.2	16.4	16.4	16.6	
MCHC	24.9	25.0	24.6	23.8	24.6	
RDW		19.4	19.7		19.8	
RBC		2.7	2.9		2.95	
WBC	2000	1000	1700	1400	1300	1800
Neutrophil	55	48	50	39	49	42
Lymphocytes	40	42	43	53	47	46
Eosinophils	03	08	04	03	03	08
Monocytes	02	02	03	05	01	04
Basophils	00	00	00	00	00	00
Platelets	80000	150000	64000	90000	55000	76000

Biochemical Parameters :

	4/9/19	2/9/19	28/8/19	24/8/19
T BIL	0.6	1.05	0.8	1.6
D BIL		0.6		0.6
T Protein	6.1	6.24	4.6	6.5
ALB/GLOB	3.2/2.9	3.43/2.81	2.5/2.1	3.0/3.5
AlkPO4	76	85	82	86
ALT/AST	24/29	12.5/33.7	18/28	11/28
UR/CR	27/0.7	12.6/0.71	13/0.4	25/0.7
Na/K	137/3.2	139/3.5	139/3.4	145/3.7
Ca				7.2
iPO4				1.8
RBS				78

Iron Profile: (2/9/19)
Serum Fe : 28 Percentage Saturation: 7.7
TIBC : 364 Prothrombin Time : (28/8/19)
PT : 16 sec INR: 1.38 Control : 12.2sec

Discussion

Sturge Weber Syndrome (SWS) also known as encephalotrigeminal Angiomatosis is a rare congenital disorder that occurs due to malformations of the cerebral blood vessels located in the pia mater, most commonly over the occipital region. The disease is commonly unilateral and rarely bilateral. It affects males and females equally. The incidence is 1 in 50000 live births[5].

SWS is caused by the persistence of embryonic blood vessels within the developing cephalic part of neuroectoderm beyond 9 weeks of gestation. These persisting immature vessels cause hypoxia, ischemia, venous occlusion, infarction and calcification which result in neurological dysfunctions like seizures, hemiplegia and mental retardation. The skin is affected because of the persistence of the vessels which may be associated lesions in the choroidal vessels of the eye.

Portal hypertension is a rare feature of SWS which is associated with portal venous malformation and hence the diagnosis is often missed.

Therefore, early clinical suspicion with proper imaging is necessary for the diagnosis of this rare condition in patients with Sturge Weber Syndrome.

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