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Case Report

Cerebral sinovenous thrombosis in a child with homocystinuria

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Introduction

Cerebral sinovenous thrombosis (CSVT) is a potentially fatal condition which can have multiple etiologies and varied presentation in children. Management of CSVT is incomplete without a search for possible etiology. Sébire et al (2005) found that the incidence of pediatric CVST is at least 0.67 per 100,000 children per year.[1] We present an interesting case of a six-year old-boy with extensive CSVT who was diagnosed to be suffering from homocystinuria.

Case report

A six year old patient was brought to hospital with complaints of multiple episodes of vomiting for two days. He had no diarrhea, fever, oliguria or high-colored urine. The child was a second product of non-consanguineous marriage. He had moderate mental subnormality (IQ: 50). A child psychologist had worked him up at the age of five years when his thyroid status, karyotype, blood lead levels, parental questionnaire for autistic spectrum disorder were detected to be normal. Perinatal, family, socioeconomic, immunization and treatment history was unremarkable.

On admission, he was irritable, pale and had tachycardia (120/min). Temperature, respiration, blood pressure, capillary filling time, oxygen saturation (SaO_2) and skin turgor were normal. He did not have icterus, cyanosis, lymphadenopathy, rash and neck rigidity. He had hyper-pigmentation over dorsa of fingers, axilla and groin as shown in [Fig. 1]. His parents and sibling did not have similar hyper-pigmentation. His weight and height at admission were 16.8 kg (81.95% of 50th centile of WHO reference i.e. 20.5 kg) and 115 cm (99.13% of 50th centile of WHO reference i.e. 116 cm). His arm-span was 114.5 cm and ratio of upper body segment to lower body segment was 1.1:1 (both normal for age). His systemic examination was normal. The boy was managed as a case of acute gastritis with mild dehydration with Oral Rehydration Solution and anti-emetics. He became asymptomatic within 12 h of admission. After about 30 h of admission; he became irritable and febrile. He had hair-pulling and head-banging which were suggestive of headache. After about 2 h of the first spike of fever, he had an episode of vomiting. Fever, intermittent in type, lasted for about 12 h (maximum temperature: 101 °F). He had no meningeal signs. Fundoscopy was normal. Investigations

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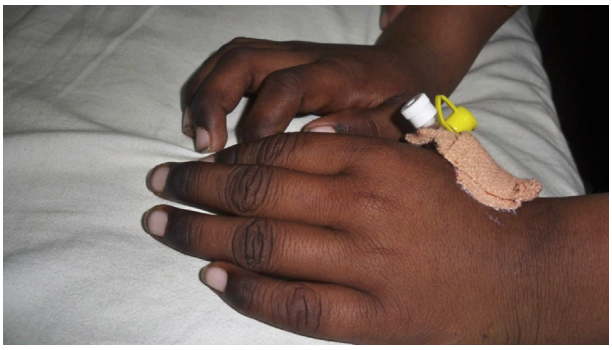

Fig. 1 – Photograph showing hyper-pigmentation over the dorsa of fingers.

revealed Hb: 9.8 g/dl Total Leukocyte Count: 14,200/mm^3 Differential Leukocyte Count: $P_{80}L_{14}M_{02}E_{04}$ Platelets: 1,90,000/mm^3. Peripheral blood smear showed microcytic hypochromic anemia with anisocytosis, poikilocytosis and leukocytosis with leftward shift. No hemoparasites were seen. Liver Function Test, Blood urea, Serum creatinine, Blood sugar, Serum electrolytes, urinalysis and Blood culture were within normal limits. Lumbar puncture released CSF with high pressure (not quantified). Cerebro-spinal fluid studies were normal. On day 3 of admission, the patient had generalized tonic clonic seizures lasting 3 min followed by drowsiness, bradycardia and projectile vomiting. He was managed with intravenous phenytoin sodium, mannitol, broad spectrum antibiotics and intravenous fluids. Neuroimaging was locally not available. Due to rapid clinical deterioration with signs of raised ICT, he was electively ventilated for 72 h. After extubation, the boy was asymptomatic with no signs of neurological impairment. CT scan brain showed thrombosis of superior sagittal and right transverse sinuses [Figs. 2 and 3]. His PT-INR was 0.8 and d-dimer levels were 8.8 μg FEU/ml (>0.5 μg FEU/ml). Hb electrophoresis, echocardiography, urine for homocystinuria and reducing substances, Mantoux test, sickle screen, HIV (I & II) screen, serum cortisol, aldosterone, T_3/T_4/TSH, ANA profile, ECG, chest radiograph, USG abdomen and Doppler study of renal, abdominal and neck vessels were essentially normal. Low Molecular Weight Heparin (LMWH) (1 mg/kg/dose 12 hrly s) was initiated which was substituted

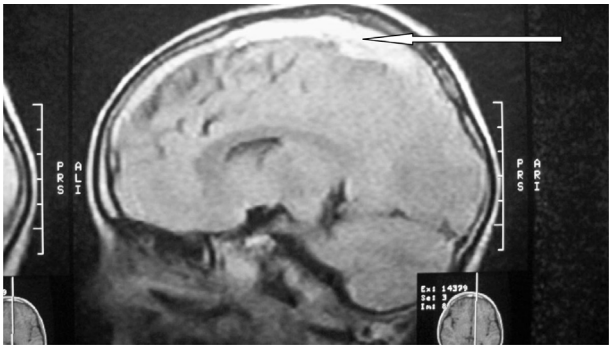

Fig. 2 – CT Scan brain showing thrombosis of the superior sagittal sinus.

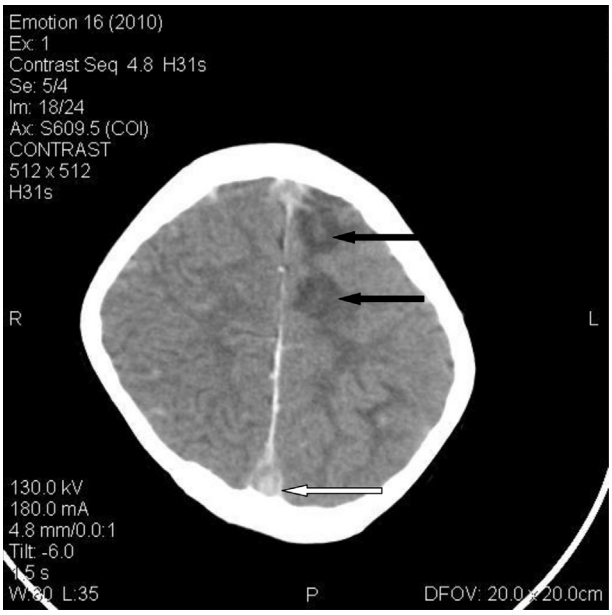

Fig. 3 – CECT brain showing the enhancing dura around the thrombosed superior sagittal sinus, the "empty delta sign"(white arrow). Multiple hypodense (black arrows) are noted involving the centrum semiovale on left side.

one week later with oral coumarol. He was discharged home after two weeks on oral iron, phenytoin sodium, acenocoumarol with PT-INR of 1.5.

The child was readmitted after two weeks with headache, vomiting and irritability followed by generalized status epilepticus, right sixth cranial nerve palsy and decorticate posturing. He was managed with Inj Midazolam, Phenytoin Sodium, Phenobarbitone and Propofol in stepwise fashion. Cerebral decongestive therapy including head elevation,

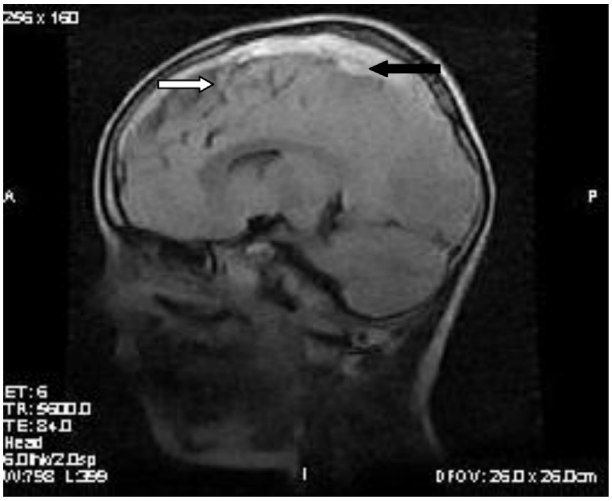

Fig. 4 – MRI sagittal FLAIR image of brain which reveals irregular and expansile hyperintense thrombus (black arrow) in the superior sagittal sinus and an ill defined hypointensity (white arrow) in the superficial part of the frontal lobe, consistent with venous infarct.