Shapiro's Syndrome

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Case Report

ABSTRACT

Shapiro's syndrome(SS) is an extremely rare disease consisting of paroxysmal hypothermia, hyperhidrosis and corpus callosum agenesis[1] with onset typically on adulthood. We reported a case of 15 year old male presented with classical triad of sudden onset of headache associated with episodes of profuse sweating, low body temperature and excessive shaking/shivering. On MR imaging complete agenesis of corpus callosum with colpocephaly, dilated and highly placed 3rd ventricle. Nasal encephalocele and anterior linear falcine lipoma is seen in this case which had not been recorded in previously reported case.

KEY WORDS: colpocephaly, encephalocele, falcine lipoma, shapiro's syndrome

INTRODUCTION:

Shapiro's syndrome is an extremely rare disorder consisting of paroxysmal hypothermia, hyperhidrosis and agenesis of corpus callosum with onset typically on adulthood.[1] It is described by Shapiro and Plum in 1967.[2] There is variation in the frequency and duration of episodes from person to person. Less than 60 cases has been reported till date. The pathophysiology as well as the prognosis of SS are still debatable. Carbamazepine, Clonidine, Valproate Sodium Valproate, propanolol, pizotifen has been used but there is no treatment consensus.

CASE REPORTS:

A 15-year-old male patient presented with complaint of sudden onset of headache associated with episodes of profuse sweating, low body temperature and excessive shaking/shivering lasting for about 15-30 minutes. He also complains of nausea, weakness, vertigo, polydipsia and polyuria. As narrated by patient's mother he developed similar symptoms at the age of 7 years and had similar complaints every 6-8 months for which he was admitted and given symptomatic treatment. The episodes were self resolving and did not have any aggravating factor. There was no history of seizures, confusion and palpitations during the episode. There was no known past history of head or spinal injuries. Family history was not significant.

The patient was admitted in our hospital with episodes of hyperhidrosis, and subsequent hypothermia. On general physical examination a swelling was present over nose and multiple hyperpigmented macules are present over abdomen and trunk (Figure 1). His neurological examination is insignificant. Routine hematological and biochemical investigations were normal. Thyroid hormonal profile, electrocardiogram, X-ray chest and USG abdomen did not reveal any significant abnormality.

MRI brain shows complete agenesis of corpus callosum, Ventricles- run parallel rather than normal bow type configuration giving "racing car" appreance on axial imaging [Figure-2(a)], characteristic "Moose Head or Viking Helmet" appearance on coronal imaging [Figure-2(b)]due to colpocephaly (dilated and widely seperated posterior horn of lateral ventricle)[Figure-2(c)] and dilated and highly placed 3rd ventricle [Figure-2(d)]. Associated with nasal encephalocele[Figure -3(a)] and linear falcine lipoma anteriorly [Figure -3(b)].

DISCUSSION:

The corpus callosum is the largest interhemispheric connective fibre bundle in brain and the largest fibre tract in the cental nervous system[3].The triad of hyperhidrosis, corpus callosum

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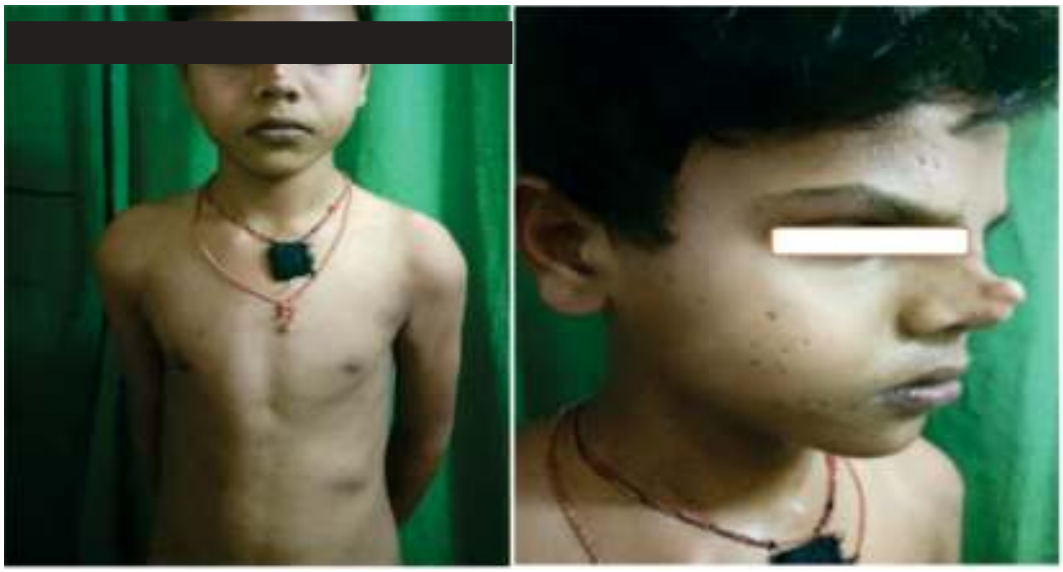

Figure 1: Hyperhidrosis with nasal encephalocele and multiple hyperpigmented macules over trunk.

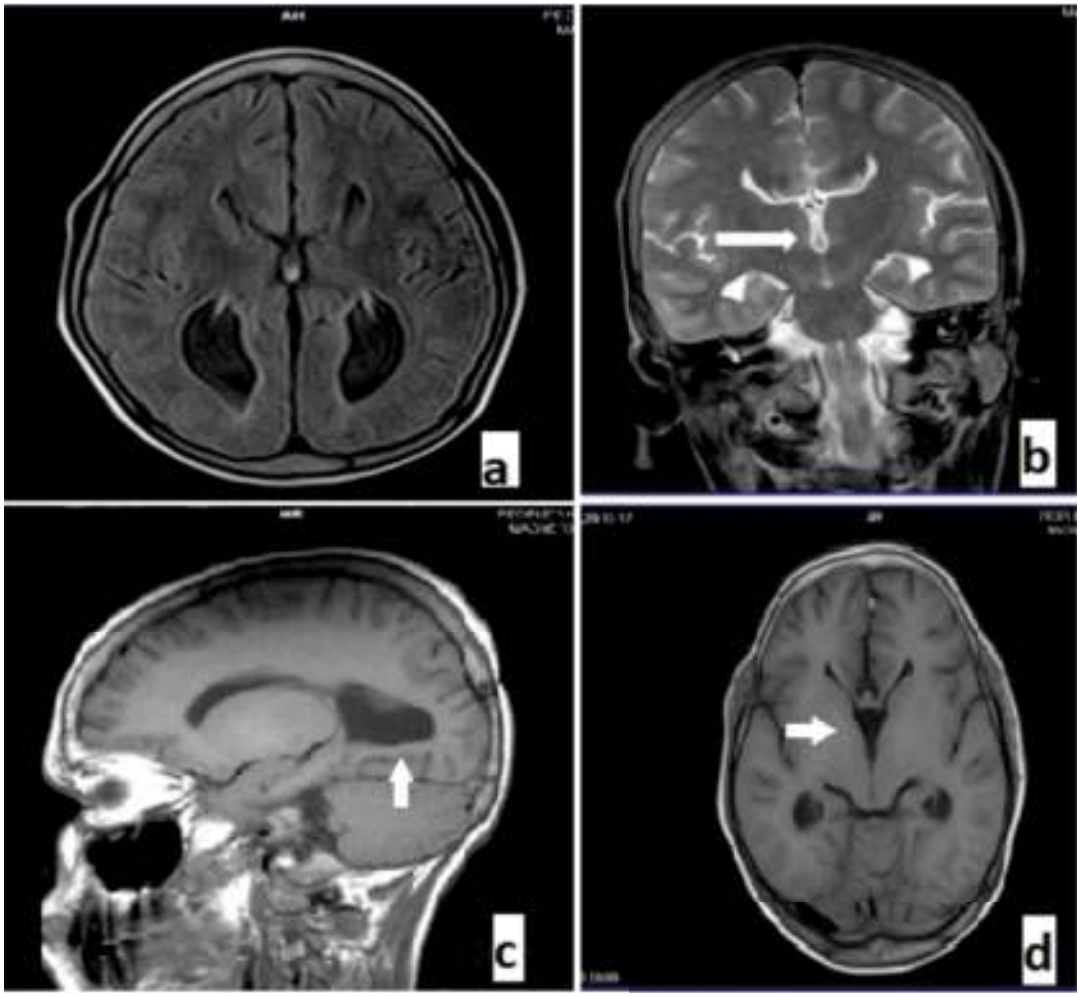

Figure 2: (a) Racing car sign. (b)Moose head or Viking helmet sign. (c) Colpocephaly. (d) highly placed 3rd ventricle.

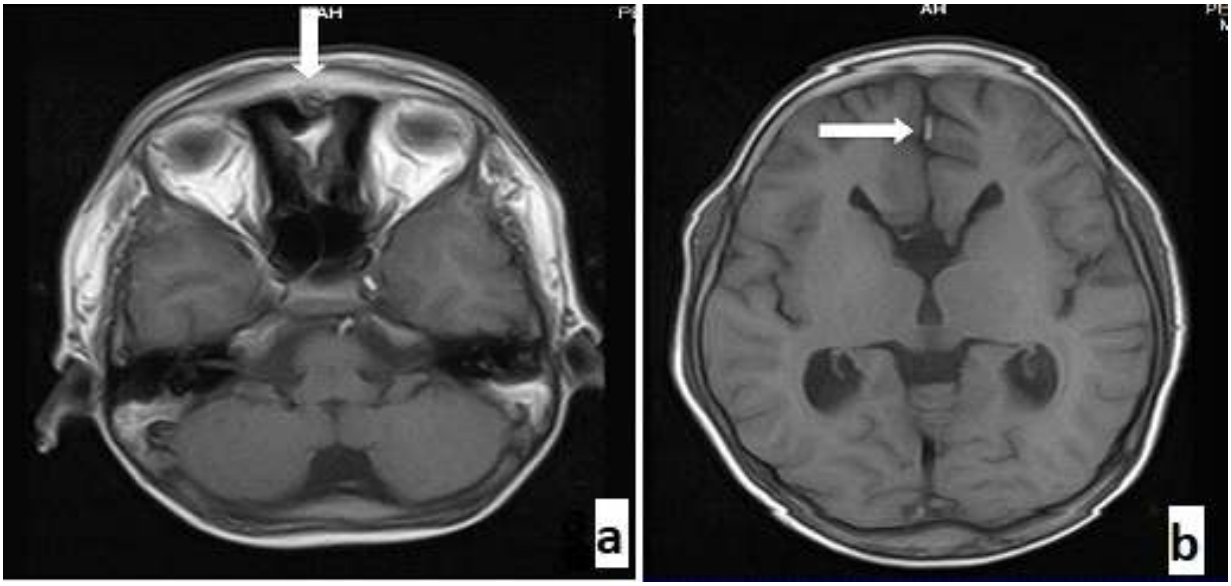

Figure 3: (a)Axial T1WI shows Nasal encephalocele, (b) Falcine lipoma anteriorly on T1WI.

agenesis and spontaneous periodic hypothermia without any hypothalamic lesion pointing towards unusual neurological disorder of SS. It had typical onset in adulthood[3,4]. The duration and frequency of the episodes vary from person to person, with some episodes lasting hours to weeks and occurring from hours to years.[5]

To date, several hypothesis have been suggested regarding the pathophysiologic mechanisms underlying this syndrome. Clearly, the agenesis of the corpus callosum by itself does not cause thermal dysregulation, as callosotomy did not lead to defective thermoregulation and hypothermia .

It is thought that the imbalance between anterior hypothalamic heat-dissipating centre and posterior hypothalamic heat-conserving center causing the fluctuating body temperature in Shapiro syndrome. Suggested mechanisms include central nervous system structural abnormalities, degenerative processes, neurochemical dysfunction, inflammatory processes, and seizure activity.

The differential of the disease are severe hypothyroidism, use of antipsychotic drugs with a strong 5-HT2 antagonistic component and hypoglycemia or attacks of diabetic ketoacidosis [6].

CONCLUSION:

Magnetic Resonance Imaging is the classical imaging tool for the diagnosis of Shapiro syndrome and its variants. There is no cure for Shapiro syndrome. Management is mainly supportive and includes rewarming with warm blanket. We reported this case as its association with nasal encephalocele and anterior linear falcine lipoma which had not been reported till date.

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