CASE REPORT

A Rare Occurrence of Shagreen Patch on the Face of a Pediatric Patient with Tuberous Sclerosis

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Abstract

Tuberous sclerosis is a neurocutaneous syndrome characterized by a clinical spectrum varying from severe mental retardation and incapacitating seizures to normal intelligence and lack of seizures, often within the same family(1). The younger the patients present with symptoms and signs of TSC (tuberous sclerosis complex), the greater are the likelihood of mental retardation (2). Skin features are very characteristic of the disease and help in the early diagnosis of the disease. Shagreen patch is one of the major diagnostic features of the disease. It is a less common skin lesion consisting of an excess amount of fibrous tissue similar to that found in scar tissues (3). It is a section of thickened, elevated pebbly skin like an orange peel. The name is derived from the French phrase "peau chagrinee". It is usually found on the lower back, buttock, thigh and the nape of the neck commonly. In this case report we report a characteristic skin lesion, the shagreen patch in a six-year-old child with TSC at an unusual site (the cheek) never reported before with the best of our knowledge.

Keywords: Tuberous sclerosis; Shagreen patch; Child; Cheek

Introduction

Tuberous sclerosis is a rare genetic disorder inherited as an autosomal dominant trait with variable penetrance and a prevalence of 1/6000-9000 of the population. Spontaneous genetic mutations occur in up to 75% of the cases. Molecular genetic studies have identified two foci for the TS complex. The TSC1 gene is located on chromosome 9q34 and the TSC2 gene is placed on chromosome 16p13. The 8.6-kb TSC1 transcript encodes a protein of 130 kd called hamartin. The TSC2 gene encodes the protein tuberin. Hamartin and tuberin act together as a single molecular complex at the Golgi apparatus. TS is an extremely heterogeneous disease with a wide clinical spectrum varying from severe mental retardation and incapacitating seizures to normal intelligence and lack of seizures, often within the same family. As a rule, the younger the patient presents with symptoms and signs of TS, the greater is the likelihood of mental retardation. The disease affects many organ systems, other than the skin and brain including the heart, kidney, eyes, lungs and the bone. Definite tuberous sclerosis complex is diagnosed by the presence of two major features or one major feature plus two minor features (2).

Case report

A 6-year-old Indian boy presented in Kamlaraja Hospital, Gwalior complaining of abnormal body movement with altered tone and consciousness. When asked for history, the parents revealed that the boy is the product of a non consanguineous

marriage without any significant family and sibling history. The birth history was uneventful, but as the child reached the first year of life, the parents noticed a lag in all domains of development compared to siblings and peers. But because of the poor socioeconomic background and illiteracy, they did not seek medical help. The boy was apparently normal till the age of 6 years, then started developing abnormal body movements which seemed to be focal tonic-clonic seizures, gradually increasing from two-three to eight-ten episodes per day from two months before, confirmed after witnessing an episode after admission and relieved by intravenous valproate.

By history and examination, the boy had global developmental delay with a moderate IQ and normal anthropometry for age. Head to toe examination revealed multiple adenoma sebaceum (Fig. 1), ash leaf spots (Fig. 2) and pilonidul sinus (Fig. 3). The unique feature of this case is the presence of a large shagreen patch (Fig. 4) on the left cheek and another on the tragus above the first one. This as the prominent feature in this case caught our attention because of its unusual site and size, as in the literature no past reports of such a presentation were found and our case appears to be the first of its type. Neurological examination revealed differential hypotonia with unilateral plantar extensor. MRI brain demonstrated subependymal nodules (Fig. 5, 6). The rest of the system and all other investigations such as blood, CSF, echocardiography, renal ultrasonography, optholmological and skeletal work up was normal.

Discussion

The criteria for diagnosing tuberous sclerosis complex have been revised (4-6).The features have been divided into major features and minor features. Major features of tuberous sclerosis are namely, facial angiofibromas or forehead plaques (7), nontraumatic ungual or periungual fibromas, hypomelanotic macules (≥3), shagreen patch (connective tissue nevus), multiple retinal nodular hamartomas, cortical tuber, subependymal nodule, subependymal giant cell astrocytoma, cardiac rhabdomyoma, single or multiple lymphangiomyomatosis and finally, renal angiomyolipoma.

Minor features of tuberous sclerosis are multiple randomly distributed pits in the dental enamel, hamartomatous rectal polyps, bone cysts, cerebral white matter radial migration lines, gingival fibromas, nonrenal hamartoma, retinal achromic patch, confetti skin lesions and multiple renal cysts.

Diagnosis of tuberous sclerosis

Definite tuberous sclerosis complex is diagnosed by the presence of two major features or one major feature plus two minor features. Probable tuberous sclerosis complex is indicated by one major feature plus one minor feature. Possible tuberous sclerosis complex is indicated by either one major feature or two or more minor features (4-6).

The most common skin lesions are hypomelanotic macules (ash-leaf spots) that occur in more than 90% of patients with TSC. Other cutaneous clinical features include a forehead plaque, shagreen patch, facial angiofibromas and periungual fibromas (8). Interestingly, all the skin lesions mentioned above come in major features. To the best of our knowledge, shagreen patch has never been reported on the face so far. Shagreen patch poses cosmetic problems especially for teen girls. It may be easily removed by surgery but there is a risk of scar which should be discussed with a dermatologist and plastic surgeon regarding the risks and benefits. Currently, there is no non scarring method for removal of these lesions, but carbon dioxide laser might be of some help in this regard (3).

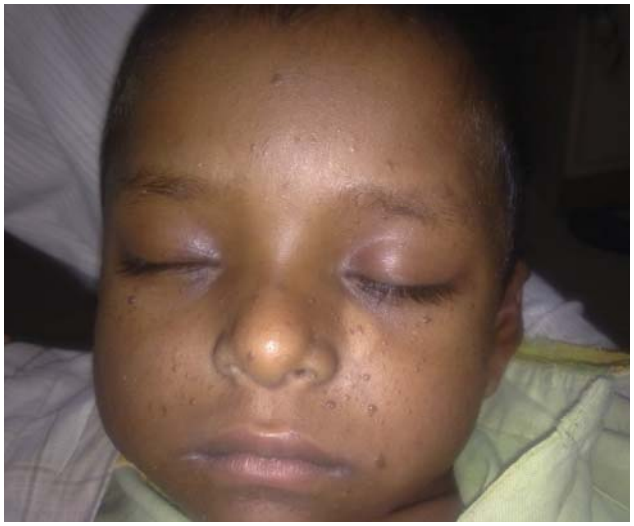


Fig 1. Numerous adenoma sebaceum on the patient's face

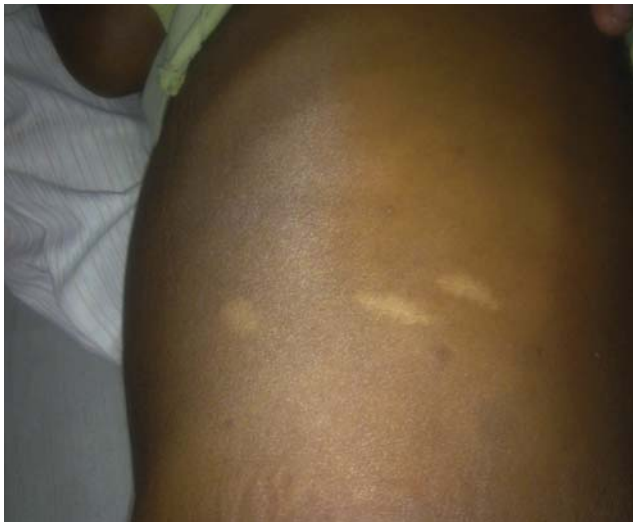


Fig 2. Ash leaf spots on the patient's trunk

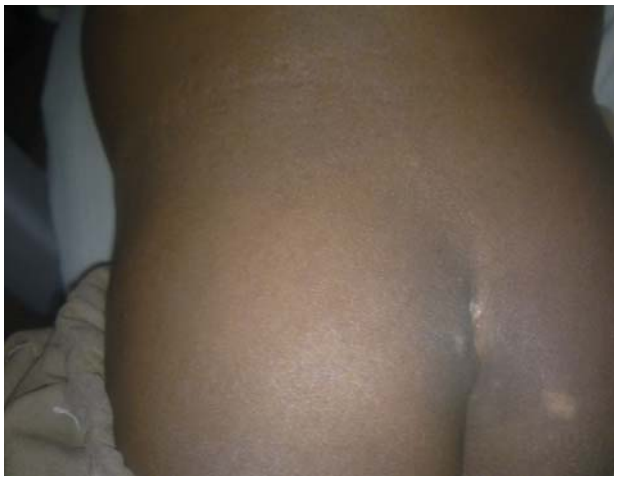


Fig 3. Pilonidal sinus

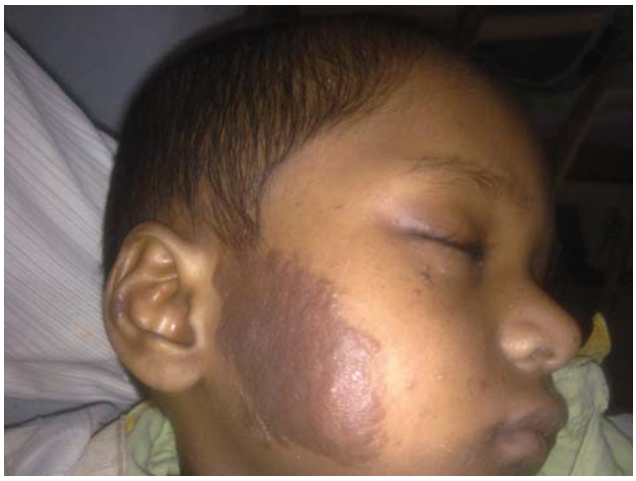
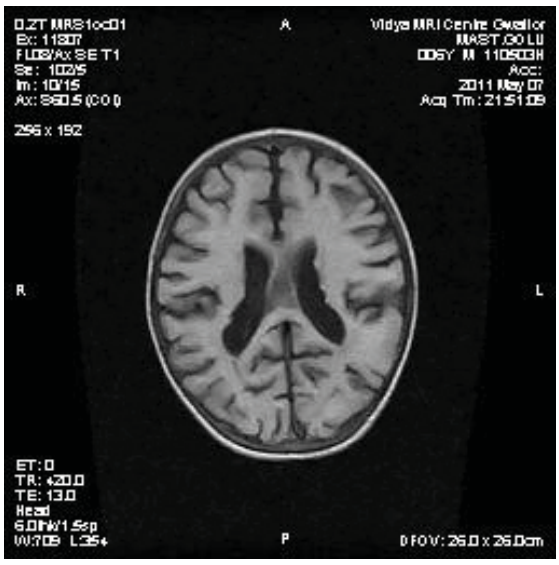
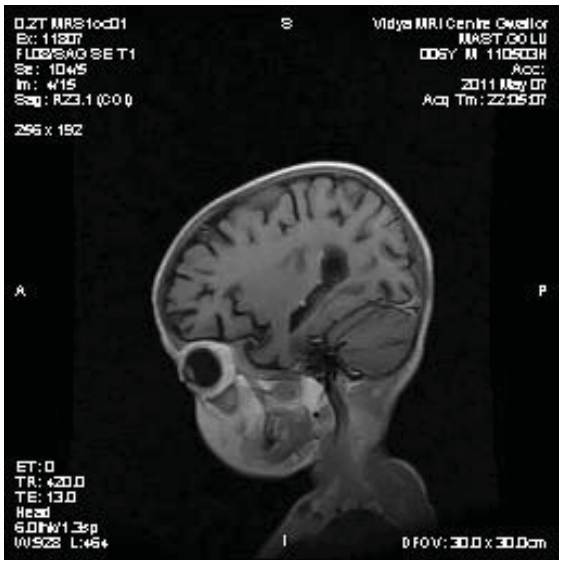


Fig 4. Shagreen patch on the patient's cheek and tragus





Figs 5, 6. Brain MRI showing subependymal nodules

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