Case Report

**Tuberous sclerosis with multiple skin lesions**


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**Abstract**

Tuberous sclerosis (TS) is an autosomal dominant neurocutaneous disorder characterized by a triad of epilepsy, mental retardation and adenoma sebaceum with multisystem involvement. We report a case of 22-year-old male who presented with the complaints of uncontrolled seizures since childhood associated with mental retardation and multiple skin lesions. He also had associated ventriculomegaly that was managed conservatively.

**Key words**

Tuberous sclerosis, neurofibromas, angiofibromas, seizures, hydrocephalus, Bourneville-Pringle disease

**Introduction**

Tuberous sclerosis (TS) is an autosomal dominant neurocutaneous disorder with 95% penetrance, and a birth incidence of 1 in 11,000 \(^1,2\) and characterized by a triad of epilepsy, mental retardation and adenoma sebaceum with multisystem involvement secondary to defects in cellular migration, proliferation, and differentiation (Table 1).\(^3,4,5,6,7\) In this article we demonstrate the multiple skin manifestations associated with TS.

**Case report**

A 22-year-old male presented with the complaints of uncontrolled seizures since childhood associated with mental retardation and multiple skin lesions. The patient had multiple cutaneous manifestations of the disease (Figures 1-5).

On examination there were multiple centrofacial, red, exophytic papules and plaques all over the face. There were skin-colored, firm, papillated plaques over forehead, nape of the neck and axilla. Numerous, confetti, hypopigmented macules were present on the lower extremities. He also had subependymal calcified nodules with aqueductal stenosis and obstructive hydrocephalus on a computed-tomography scan of his brain (Figure 6 and 7). There were no other systemic manifestations. There was no family history of similar signs...
Figure 1 Dense lesions of adenoma sebaceum on face

Figure 2 Adenoma sebaceum and shagreen patch over lower back and buttocks

Figure 3 Skin tags, acanthuses, hyper pigmented striate and acneiform eruptions in the maxillary region.

Figure 4 Multiple lentigenes in antecubital fossa

Figure 5 Multiple neurofibromas, Café-au-lait spots, lentigenes and pityriasis versicolor over back

Figure 6 CT scan showing aqueductal stenosis with obstructive hydrocephalus.

Figure 7 CT scan showing sub-ependymal calcification,
Table 1 Various manifestations of tuberous sclerosis

<table>
<thead>
<tr>
<th>System</th>
<th>Manifestations</th>
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<tr>
<td>Brain</td>
<td>Early childhood seizures, calcified subependymal nodules, cortical tubers, giant cell astrocytomas, cerebral white matter radial migration lines</td>
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<tr>
<td>Skin</td>
<td>Facial angiofibromas (adenoma sebaceum), Pringle's birthmarks, fibrous plaques, collagenomas (shagreen patches), periungual fibromas (Koenen tumors), gingival fibromas, dental enamel pits, hypopigmented macules (ash-leaf and confetti), cafe-au-lait macules</td>
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<tr>
<td>Heart</td>
<td>Cardiac rhabdomyomas, arrhythmias (Wolf-Parkinson-White syndrome)</td>
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<tr>
<td>Kidney and liver</td>
<td>Angiomyolipomas, renal cyst, rarely renal cell carcinomas</td>
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<tr>
<td>Lung</td>
<td>Pulmonary, lymphangioleiomyomatosis</td>
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or symptoms. Seizures were controlled with anti-epileptics and fungal infection was treated with anti-fungal agents.

Discussion

TS results from mutations in one of two genes, TSC1 on chromosome 9q34 and TSC2 on chromosome 16p13, that encode distinct proteins, hamartin and tuberin respectively.8,9 The diagnosis of TS is mainly clinical, with neuroradiographic imaging based on the diagnostic criteria of major and/or minor features. Definite tuberous sclerosis complex means either one major feature or two or more minor features.4,10 The most commonly identified brain lesions in TSC are cortical tubers, subependymal nodules, SGCA, and white matter abnormalities.11 Hydrocephalus in TS is usually caused by mass lesions.7,12,13

In our case the hydrocephalus due to aqueductal stenosis was managed conservatively as the patient was not symptomatic for hydrocephalus. In our case patient also had fungal infection that was due to the inability of the patient to take care of him and it was managed with antifungal agents. Patients often seek medical attention for dermal lesions or frequent seizures.10 Investigations and management of these patients should be coordinated between multiple specialties with periodic imaging assessments and symptomatic treatment.4,14

Assessments include brain computed tomography or magnetic resonance imaging, electroencephalogram, retinal evaluation, skin examination, renal ultrasound, electrocardiogram, echocardiogram, and neurodevelopmental assessments. Cutaneous manifestations are often of cosmetic concern and typically can be managed by excision, dermabrasion, and lasers.4,14 Patients with sub-ependymal nodules would require monitoring for early detection of obstructive hydrocephalus.15,16 Sudden death may be due to cardiac arrhythmia, epilepsy, and intratumoral hemorrhage with additional complications including cardiac outflow obstruction, obstructive hydrocephalus, aneurysm rupture, and spontaneous pneumothorax.3,17,18 An awareness of these highly variable tissue manifestations of tuberous sclerosis and the mechanisms are
necessary to establish the diagnosis and for optimal management.

References