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INTERESTING MEDICAL IMAGE

The Clinical and Paraclinical Manifestations of Tuberous Sclerosis in an Omani Female Patient

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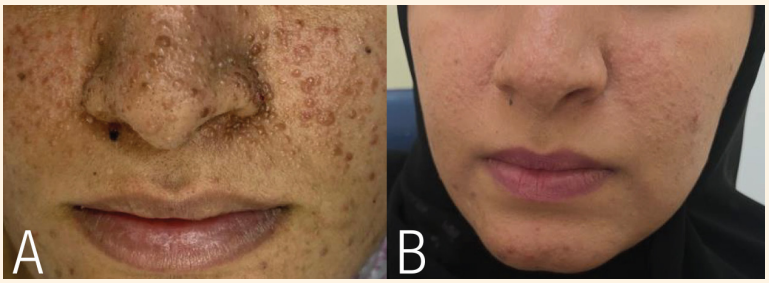

Figure 1: Photographs of the face of a 24-year-old female patient showing facial angiofibroma **(A)** before treatment and **(B)** post-radiofrequency ablation and CO2 laser.

A 24-year-old Omani female patient presented to a dermatology department in Rustaq, Oman, in 2020 with skin lesions over her face and trunk since childhood. Her medical history was unremarkable. There was no family history of similar lesions. Physical examination showed multiple angiofibroma (adenoma sebaceum) over her face, multiple collagenoma over her trunk and a shagreen patch on her lower back [Figures 1 and 2]. Based on the suspicion of tuberous sclerosis (TS), the patient underwent a renal ultrasound, brain magnetic resonance imaging (MRI), echocardiogram, ophthalmic examination and genetics testing. A computed tomography scan of her kidneys, ureter and bladder showed bilateral renal angiomyolipoma [Figure 3]. The brain MRI revealed cortical and subcortical tubers with enhancing subependymal nodules [Figure 4]. Her echocardiogram and ophthalmic examinations were unremarkable. The patient was treated with 1 session of radiofrequency ablation with CO2 laser for the facial angiofibroma with good clinical response. The patient became pregnant due to which other sessions were postponed.

Patient consent was obtained for the publication of these images.

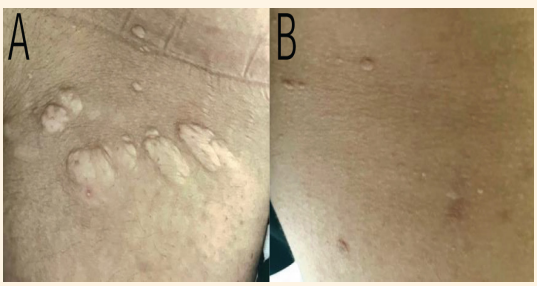

Figure 2: Photographs of a 24-year-old female showing **(A)** Shagreen patch on the lower back and **(B)** multiple collagenoma over the trunk.

Comment

TS complex (TSC) is an autosomal dominant neurocutaneous syndrome that affects multiple organ systems.[1] One third of cases of TS are familial and caused by mutations in 2 tumour suppressor genes, TSC1 and TSC2. The other two thirds of cases are sporadic and due to spontaneous mutations as was seen in the current patient.[2] Both adults and children

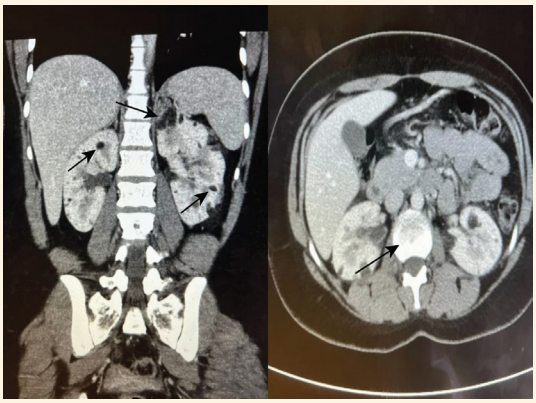

Figure 3: Computed tomography scan of the patient's kidneys, ureter and bladder showing multiple renal angiomyolipomas (arrows).

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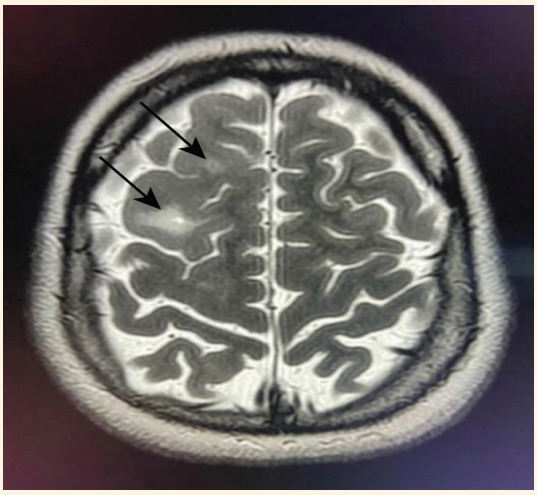

Figure 4: Brain magnetic resonance imaging of the patient showing cortical and subcortical tubers (arrows).

can be affected. Many people have signs of the TS as early as the first year of life, while others may take years to develop signs and symptoms.[3]

Diagnosis is based on clinical and paraclinical criteria; the major criteria includes facial angiofibromas or forehead plaques, non-traumatic ungula or periungual fibroma, more than 3 hypopigmented macules, Shagreen patch, cortical tubers, subepandymal nodules, subepandymal giant cell astrocytoma, multiple retinal nodular hamartomas, cardiac rhabdomyoma, lymphangiomyomatosis and renal angiomyolipoma. Minor criteria consist of more than 14 dental pits, hamartomatous rectal polyps, bone cysts, cerebral white matter radial migration lines, non-renal hamartomas, retinal achromatic patch, confetti skin lesions and multiple renal cysts. Either 2 major criteria or 1 major and 2 minor criteria must be established to diagnose a patient with TSC.[2]

The most common symptoms in TS are dermatological and neurological findings.[2] The skin manifestations of TS are the most common findings;[2] approximately 90% of these patients have hypomelanotic macules, also known as ash leaf spots. These lesions occur in an early onset of life. Furthermore, 75% of patients with TS have facial angiofibromas, formerly known as adenoma sebaceum, which appear as erythematous papules and are typically seen in malar distribution.[4] Approximately 50% of patients with TS develop Shagreen patch which presents in the lumbosacral region as thick leathery skin with a pebbly texture.[5] These dermatological findings of TS are the only manifestations that can be observed in physical examination and considered as major criteria in the diagnosis of TS.[2]

Epilepsy is the most common presenting symptom of TS. Seizure is discovered in 98% of patients with TS and 75% have a seizure in the first year of life.[6] Other neurological findings include cortical tubers, subepandymal nodules and subepandymal giant cell astrocytoma that can lead to seizure attacks.[4] Renal involvement is seen as angiomyolipomas or cysts in TSCs.[2] Ocular manifestations are seen in less than 50% of patients with TS as retinal hamartomas and are bilateral in one third of the cases.[2] Cardiac involvement manifests as cardiac rhabdomyoma and sometimes is the earliest diagnostic finding in TSC. These hamartomas of the heart remain asymptomatic and regress in size and number until 6 years of age in 58–74% of patients.[2]

In the current case, the patient remained asymptomatic, had no history of seizure and she required sessions of radiofrequency ablation with CO2 laser for facial adenoma sebaceum with excellent response. She has 1 healthy child who does not show any clinical features of TS.

AUTHORS' CONTRIBUTION

SaK was involved in the patient evaluation, diagnosis management and review the case. ShK was involved in collecting the data, writing the case with literature review and did the follow-up with the patient. All authors approved the final version of the manuscript.

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