CASE REPORT

Scalp fibroma: a rare cutaneous manifestation of tuberous sclerosis

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SUMMARY

We report a case of a 23-year-old woman with a history of generalised tonic-clonic seizures, reddish brown maculopapular swelling over the face and an enlarging swelling over the scalp. Physical examinations revealed angiofibroma of the face and other typical cutaneous lesions of tuberous sclerosis, for example, shagreen patch and periungual fibroma. Scalp swelling was labelled as fibroma by dermatologists, which was further supported by the histopathological findings. Fibroma of the face is one of the commonest lesions, however, fibroma of the scalp is a rarely described entity.

BACKGROUND

Tuberous sclerosis, a neurocutaneous syndrome, has varied neurological and cutaneous manifestations. Neurological manifestations like seizures are perhaps the most common reason to seek medical help, whereas, cutaneous manifestations are the ones which help make a spot diagnosis. Facial angiofibromas are the most evident and one of the commonest cutaneous lesions but fibroma of the scalp is a rarely described entity.

CASE PRESENTATION

A 23-year-old right-handed woman, born out of non-consanguineous marriage with normal developmental milestones, without any significant personal or family history, presented with a 18-year history of generalised tonic-clonic seizure (GTCS). Over the next 14 years GTCS increased in frequency up to three to four times a day. At that stage consultation was sought and she was started on antiepileptic drug, carbamazepine (CBZ) 200 mg twice a day. On antiepileptic treatment her seizure frequency decreased and for the past 10 months she is seizure free. She also had symptoms of multiple small reddish brown swellings over the face for the past

18 years, which increased in size and number until 8 years ago, becoming static thereafter; and a swelling over the right scalp increasing in size for the past 5 years. There was no history suggestive of any mental subnormality or behavioural disorders. General examination revealed facial angiofibromas (figure 1), a firm, non-tender swelling, 5×3 cm in size on the scalp over the right parietal region (figure 2), shagreen patch over the lumbosacral region (figure 3) and periungual fibroma of left little toe (figure 4). Neurological examination including fundus was normal. Other systemic examinations were also within normal limits. Dermatologist's opinion was taken, who confirmed

Figure 1 Angiofibromas in typical butterfly distribution, sparing upper lip.

our cutaneous observations and opined regarding scalp swelling as collagenoma or fibroma.

INVESTIGATIONS

Routine investigations were normal except low haemoglobin (8.7 g/dL). X-ray of the chest, renal ultrasound, echocardiography and EEG were normal. CT of the brain showed multiple, bilateral cortical and subcortical calcifications suggestive of tubers (figure 5). Skin punch biopsy from the scalp swelling was suggestive of fibroma.

TREATMENT

The patient was continued on CBZ 200 mg twice a day, as her seizure was well controlled on this

Figure 2 Scalp swelling (fibroma) measuring 5×3 cm.

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Figure 3 Shagreen patch over left lumbar region.

medication. She was planned for laser therapy for facial angiofibroma by dermatologists and surgical excision of the scalp fibroma by plastic surgeons.

OUTCOME AND FOLLOW-UP

The patient is on regular follow-up and her seizures are well controlled on CBZ.

DISCUSSION

Tuberous sclerosis complex (TSC) is a rare autosomal dominant neurocutaneous syndrome caused by the mutations of the genes; TSC-1, coding for hamartin and TSC-2, coding for tuberin at chromosomes 9q34.3 and 16p13.3, respectively.¹ TSC is characterised by the hamartomas of the brain, skin, retina, viscera and various cutaneous manifestations.² Cortical tubers are perhaps the most important hamartomas and are responsible for various neurological manifestations such as seizure, mental retardation and behavioural abnormalities.³ Seizures may be focal or generalised and usually occur in 80–90% of patients.^{3 4} Other intracranial lesions are subependymal nodules (SENs), focal cortical hypoplasia, and heterotopic grey matter. Some of the SENs enlarge and transform into subependymal giant cell astrocytoma (SEGA) resulting into increased intracranial pressure, poor seizure control and appearance of new focal neurological deficits.¹

Cutaneous lesions, seen in more than 90% of patients, help in the diagnosis and include hypomelanotic macules (ash leaf spot), facial angiofibroma (adenoma sebaceum), shagreen patch and non-traumatic periungual or unguinal fibroma.¹ Hypomelanotic macules, present in approximately 90% of patients, are seen at birth much before the appearance of other characteristic skin lesions (Fitzpatrick *et al*).⁵ These are leaf-like, linearly arranged hypopigmentation of a few millimetres to several centimetres over the trunk or extremities. These are common in normal population but the presence of three or

Figure 4 Periungual fibroma of left little toe.

Figure 5 Multiple bilateral cortical and subcortical calcifications suggestive of tubers.

more such lesions constitute one of a major diagnostic feature for TSC.^{1 6} Angiofibroma of the face are pink to red glistening nodules, classically distributed in a butterfly pattern over the cheeks and forehead but sparing the upper lip (figure 1). Although specific for TSC, angiofibroma are present only in 75% of patients and appear late during preschool age. Forehead plaque is a variant of angiofibroma, seen in 20% of the patients.^{7 8} Although fibromas are common on extremities, face or forehead, scalp fibromas (figure 2) in association with TSC are rarely described.^{7–9} They are composed of fibrocollagenous tissues with a few dilated vessels. Other uncommon locations of fibromas are gums, mucosa, lips and base of tongue.^{10 11} Shagreen patch is an irregular, slightly elevated area of skin, 1–10 cm in diameter, with pigskin, orange peel or elephant hide appearance, found most commonly in the lumbosacral region and present only in 20–30% of patients.^{1 6}

Ungual fibromas are nodular or fleshy lesions around and underneath the nails, considered specific for TSC but they are present only in 15–20% of patients that too after puberty.^{1 6}

Diagnosis of TSC is mainly clinical supported by imaging and other ancillary tests.¹ According to TSC consensus revised criteria, TSC can be labelled definite, probable or possible depending on the number of major and minor features present. For definite TSC, either two major or one major plus two minor features, for probable TSC, one major plus one minor features and for possible TSC, either one major or two or more minor features should be present.^{1 4 12} Ours was a case of definite TSC, as four major features were present (figures 1, 3–5).

Although no cure is available for the disease, yet symptoms such as seizure can be well controlled with antiepileptic drugs, steroids (adrenocorticotrophic hormone or prednisolone) and surgeries. Laser and other cosmetic surgeries could be offered for the disfiguring cutaneous lesions.⁷

So to conclude, TSC is a rare neurocutaneous syndrome with seizure as the most common neurological presentation and varied cutaneous lesions serving as the pointer to the diagnosis.

Fibroma of the face and extremities are common but scalp fibroma is very scarcely described in literatures. So proper clinical examination should be carried out in all the patients to pick these lesions in uncommon locations which can aid in the diagnosis of this neurocutaneous syndrome.

Learning points

- ▶ Tuberous sclerosis is a rare neurocutaneous syndrome.
- ▶ Neurological manifestations such as seizure incite patients to seek medical help, whereas, cutaneous lesions are the ones which serve as the pointer to the diagnosis.
- ▶ Among cutaneous lesions, angiofibroma of the face is one of the commonest and perhaps the most evident lesions, but fibroma of the scalp is very rare.
- ▶ Hence one should carry out thorough clinical search to pick these rare lesions in uncommon locations, which could serve as cutaneous markers or a clue to the underlying disease and aid in diagnosis.

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Competing interests None.

Patient consent Obtained.

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