CASE REPORT

Giant pilomatricoma in a patient with tuberous sclerosis, both diagnosed in the adult life

Cristina Isabel Pinho Resende, Joana Gomes, Maria da Luz Duarte, Celeste Brito

Department of Dermatology and Venereology, Hospital de Braga, Braga, Portugal

Correspondence to
Dr Cristina Isabel Pinho Resende,
cristinapresende@gmail.com

SUMMARY

Pilomatricoma is a relatively common benign skin neoplasm originating from the hair follicle matrix cells. β-Catenin is a subunit of the cadherin protein complex. It acts as an intracellular signal transducer that influences cell differentiation and proliferation. This protein was recently involved in the formation of hair follicle-related tumours, including pilomatricomas. Tuberous sclerosis (TS) is an inherited neurocutaneous disease, which is characterised by pleomorphic features involving many organs, hamartomas in multiple organ systems and by the fact that it is usually diagnosed early in life. We reported a case of a Caucasian patient with TS and a giant pilomatricoma, both diagnosed in the adult life.

BACKGROUND

Pilomatricoma is a benign neoplasm of follicular structure.[1 2] It commonly affects children and adolescents, but it may develop at any age.[2]

The size of the tumour rarely exceeds 3 cm, although very large tumours were reported.[2] Most of these tumours occur in the head and the neck region, followed by upper extremities.[2] The lower extremities are rarely affected.[1]

Tuberous sclerosis (TS) is a multisystem neurocutaneous disease, which is characterised by the formation of benign tumours or hamartomas in multiple organ systems, including the brain, eyes, heart, skin, lung, liver, kidney and it is usually diagnosed early in life.[3 4]

We reported a case of a Caucasian patient with TS and a giant pilomatricoma, both diagnosed in the adult life.

CASE PRESENTATION

A previously healthy 34-year-old caucasian man presented an asymptomatic slowly growing nodule on his right thigh. The nodule was associated with yellowish coloured papules and plaques on his left lumbar area and on occiput. These lesions appeared progressively over 9 years and there was no history of local trauma.

A clinical examination revealed a solitary, firm, non-tender nodule on the lateral surface of his right thigh, measuring about 6.5×2.8 cm (figure 1). The nodule was mobile and it was not fixed to the deep dermal layers. The examination also revealed multiple yellowish-coloured, firm, non-tender papules and plaques, with diametres from 5 mm to 3 cm, on his left lumbar area and on occiput (figures 2 and 3). These lesions were isolated and confluent and they had an uneven surface, with a rough texture, resembling an orange peel. The remainder of the examination revealed the presence of eight hypopigmented, well-demarcated, round and polygonal macules, with diametres from 0.5 to 2.0 cm. They were located on the abdomen and on the lower limbs. We also observed pink papules, with a smooth surface. They were symmetrically distributed over centrofacial areas, suggesting facial angiofibromas. He also presented with numerous soft, pedunculated, skin coloured papules, located on neck, axillae and groin, suggestive of molluscum pendulum.

There are no nail, hair, teeth or mucosal changes to report.

In his personal history, it is important to report infantile spasms, developmental delay and mental retardation. This patient is not taking any chronic medication.

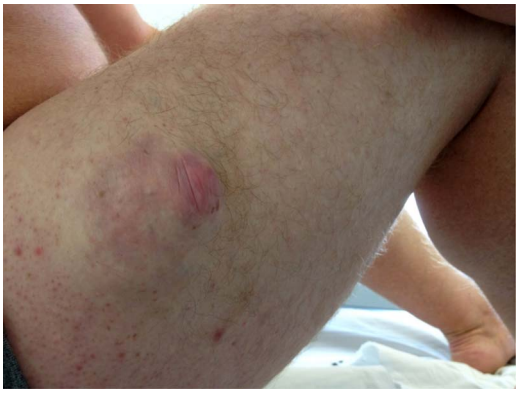

Figure 1 Nodule on the lateral surface of his right thigh.

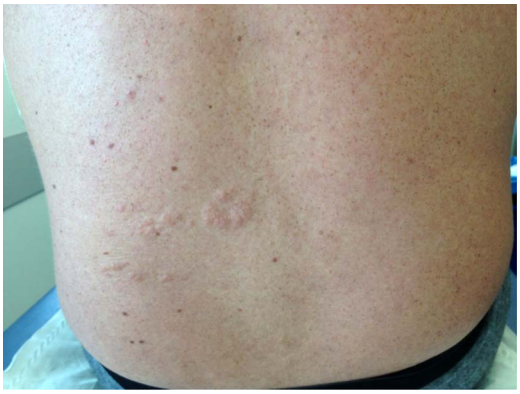

Figure 2 Papules and plaques on his left lumbar area.

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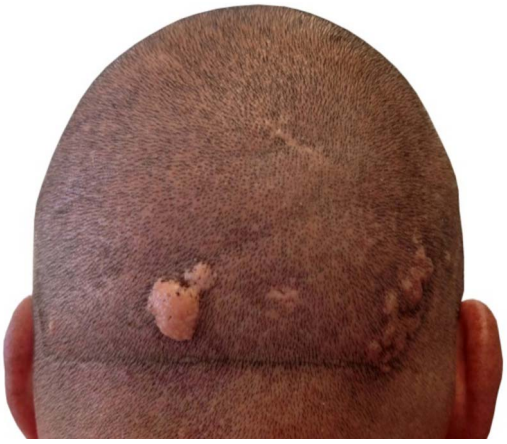

Figure 3 Papules and plaques on occiput.

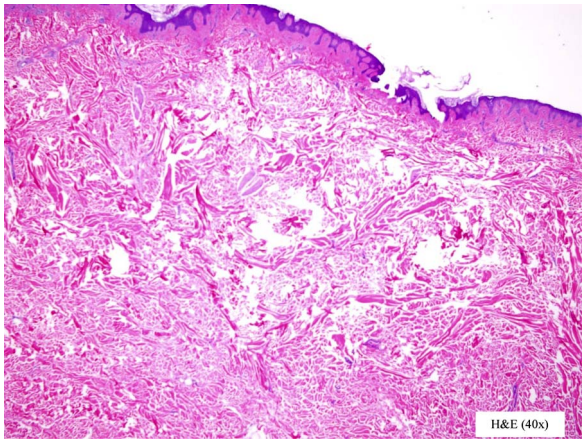

Figure 5 Within the reticular dermis there were areas with randomly arranged dense, coarse collagen fibres of various sizes.

His mother and his brother have a history of similar lesions, namely, numerous hypopigmented macules, facial angiofibromas and numerous molluscum pendulum. Moreover, they have periungueal fibromas on the toenails.

Clinical features suggested the diagnosis of TS. In light of this diagnosis, we continued our study.

Histopathological examination of excisional biopsy of a lesion taken from the right thigh displayed the typical features of a pilomatricoma (figure 4). In particular, it displayed small round basophilic cells adjacent to pale, eosinophilic, enucleated shadow cells. Focal areas of calcification were scattered throughout the tumour. A histological section of a lesion taken from the left lumbar area demonstrated that within the reticular dermis there were areas with randomly arranged dense, coarse collagen fibres of various sizes. Based on these findings, the diagnosis of collagenoma was made (figure 5).

Routine laboratory blood tests demonstrated no abnormalities. The echocardiogram was also within normal limits. The ultrasonographic examination revealed two small echogenic cysts in the left renal cortex.

Altogether, the clinical and pathological data suggested the diagnosis of a giant pilomatricoma in a patient with TS.

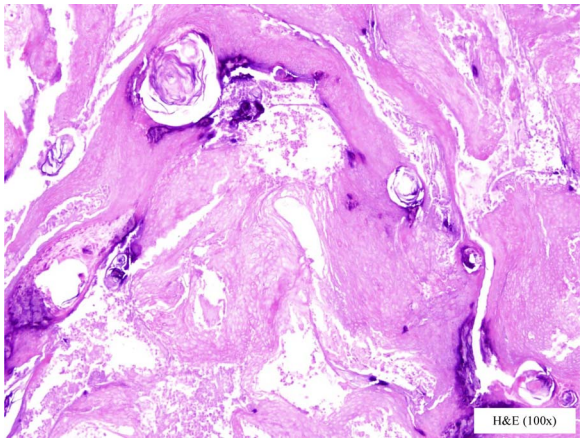

Figure 4 Small round basophilic cells adjacent to pale, eosinophilic, enucleated shadow cells.

OUTCOME AND FOLLOW-UP

Our patient was referred to genetic counselling and he is also under vigilance in the dermatology, neurology, cardiology and oftalmology consultations.

DISCUSSION

TS is an autosomal dominant disorder with an incidence of 1–10 000 births.[4] The disease is caused by the mutation of one of two tumour-suppressor genes: TSC1 and TSC2, which encode hamartin and tuberin, respectively.[3 5] Two-thirds of all the cases are caused by sporadic mutations.[2]

The diagnosis of TS is based on clinical criteria.[4] Genetic testing is not required to make a diagnosis in patients who fulfil the criteria for definite TS, but it is helpful for family studies, defining reproductive risks for relatives.[4] Our patient was diagnosed with TS because he had three major features for the clinical diagnosis of TS, namely facial angiofibromas, hypomelanotic macules and Shagreen patch. Histopathological examination of the lesion taken from the left lumbar area demonstrated the typical features of a collagenoma. The latter was compatible with a Shagreen patch, which is a connective tissue nevous composed of various amounts of vascular structures, adipose tissue, collagen, elastic fibre, smooth muscles and cutaneous appendages without increased vascularity.[4] The Shagreen patch mostly occurs in the lumbosacral area. In addition, it typically begins to develop at the age of 2 years (approximately), affecting nearly half of the patients with TS. It is also worth noting that our patient also has a history of infantile spasms, which can occur in approximately 70% of infants with TS.[4]

Nearly every organ can be affected by TS and a multidisciplinary approach is essential for an early, accurate diagnosis and proper management of the affected patients.[4 5] It is also important to provide genetic counselling to the affected individuals and families.[4]

We would also like to emphasise that our patient had a giant pilomatricoma in the right thigh. This is interesting because of the large size and the rare location of the pilomatricoma.

Pilomatricoma is a relatively common benign skin appendage neoplasm originating from the hair follicle matrix cells.[1 2 6] It has been observed in association with various disorders. However, to the best of our knowledge, in the literature, there is only one case, suggesting the association of pilomatricoma and TS.[1] According to the reported case, mutations of TS1 or

TS2 could contribute to β-catenin overactivity, resulting in the formation of a pilomatricoma.[1]

β-Catenin is a subunit of the cadherin protein complex that acts as an intracellular signal transducer. It is involved in signalling pathways that influence cell differentiation and proliferation. It has recently been shown that β-catenin plays an important role in the formation of hair follicle-related tumours, including pilomatricomas.[1 7–9] However, this is probably not the only explanation for the concomitance of TS and pilomatricomas. If this was the only mechanism involved in this association, there would be much more cases described in the literature.

The treatment choice in cases of pilomatricoma is surgical excision.[2 6] Recurrences are rare after complete ressection.[4 5]

We presented this case because not only the diagnosis of TS and pilomatricoma in adult life is a rare phenomenon, but also because this case emphasises the rare concomitance of the two disease conditions, which could theoretically, be linked to each other.

Learning points

- TS is a multisystem neurocutaneous disease, which is characterised by the formation of benign tumours or hamartomas in multiple organ systems. It is usually diagnosed early in life, although it can also be diagnosed in the adult life.
- Pilomatricoma is a relatively common benign skin appendage neoplasm. It rarely exceeds 3 cm, although some very large tumours have been reported in the literature.
- Pilomatricoma and TS can occur simultaneously and there could be a theoretical link between the two.

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Competing interests None.

Patient consent Obtained.

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