Birt-Hogg-Dubé syndrome: identifying patients at risk of renal cell carcinoma, pulmonary cysts and pneumothoraces

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DESCRIPTION

A 42-year-old woman attended the dermatology outpatient department (OPD), with a 2–3 year history of multiple firm flesh-coloured papules localised to her cheeks, periauricular and auricular area (figures 1 and 2). These lesions developed when the patient was in her late 30s. They were asymptomatic. She also had multiple fibroepithelial polyps (skin tags). She had a family history of similar skin lesions (2 sisters and her father). Her medical history was significant for mild hidradenitis suppurativa.

A punch biopsy of lesional skin was performed—histology showed focal increase in perivascular dermal connective tissue, in keeping with an angiofibroma (figures 3 and 4). Genetic testing confirmed a heterozygous variant in the folliculin (FLCN) gene, consistent with a diagnosis of Birt-Hogg-Dubé syndrome.

Birt-Hogg-Dubé syndrome is an autosomal-dominant condition. It has characteristic cutaneous findings, including fibrofolliculomas, angiofibromas, fibroepithelial polyps and trichodiscomas.

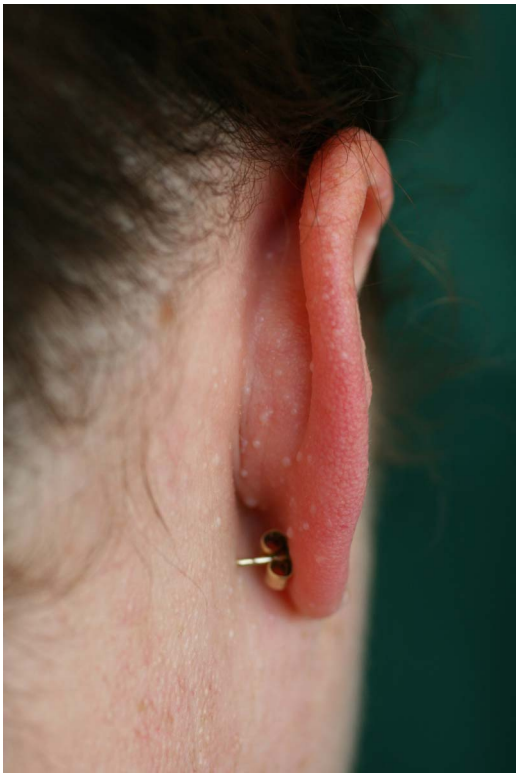

Figure 1 Multiple firm flesh-coloured papules—right post-auricular area.

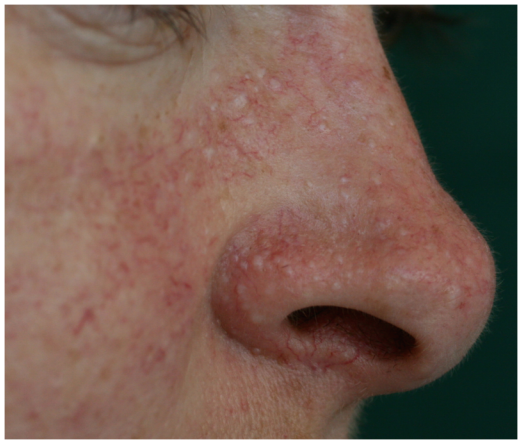

Figure 2 Multiple similar lesions affecting nasal bridge and ala.

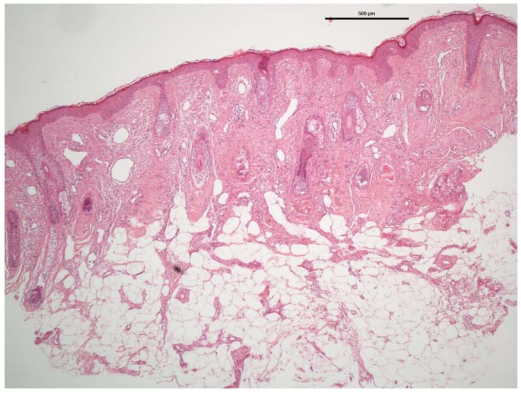

Figure 3 H&E stain—angiofibroma, left earlobe.

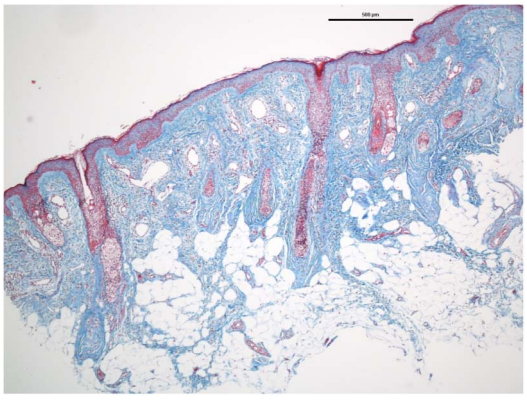

Figure 4 Masson's trichrome—highlighting fibrosis in an angiofibroma, left earlobe.

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Extra cutaneous findings include pulmonary cysts, spontaneous pneumothorax and renal cancer.[1] Management is focused on early diagnosis and treatment of renal cancer.[1] The FLCN gene, located on the short arm of chromosome 17 (17p11.2), is responsible for this syndrome.[2] It codes for the protein folliculin, thought to be an oncogene suppressor protein. It has also been linked to the mammalian target of rapamycin signalling pathway, a serine/threonine protein kinase that regulates cell growth, proliferation and survival.[3]

Following diagnosis of Birt-Hogg-Dubé syndrome, our patient had a renal ultrasound and thorax/abdomen CT to assess for pulmonary or renal disease. These revealed a benign renal cyst with no evidence of pulmonary cysts. She will require long-term follow-up and screening for renal carcinoma.

Learning points

- Multiple angiofibromata are associated with several genetic conditions including tuberous sclerosis, multiple endocrine neoplasia type 1 (MEN1) and Birt-Hogg-Dubé syndrome.
- Skin examination can help detect patients who may be at risk of certain malignancies and who may therefore benefit from regular cancer screening.
- Diagnosing a genetic condition has implications for both the patient and all family members.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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