

Case report

A 48-year-old woman presented with numerous small flesh coloured papules on her face and neck, see figure 6, below. Previously, these had been treated without a histological diagnosis, by curettage and laser therapy, with only modest improvement.

A small skin biopsy of a lesion on her neck was taken and histological examination showed it to be a fibrofolliculoma. As multiple fibrofolliculomas are a feature of Birt-Hogg-Dubé syndrome, an autosomal dominant cancer syndrome, her family history was explored. This revealed a number of her relatives to have had similar facial lesions, some to have had spontaneous pneumothorax and one to have had a renal tumour, thus confirming the diagnosis of Birt-Hogg-Dubé syndrome.

Further investigation of this patient excluded pulmonary cysts but confirmed that she had a renal carcinoma. Progressive deterioration of her renal function proved fatal.



FIGURE 6 Facial fibrofolliculomas in a woman subsequently diagnosed with Birt-Hogg-Dubé syndrome