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Nephroquiz
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Particular cutaneous lesions and chronic renal failure

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Keywords: Bourneville tuberous sclerosis; developmental defects; hamartomas; Koenen's tumours; Pringle's adenoma sebaceum

Case

A 40-year-old woman with no particular history was referred to our nephrology unit for long-standing, slowly developing chronic renal insufficiency. The blood pressure was 180/80 mmHg. Serum creatinine was 340 μmol/l and GFR 32 ml/min. Proteinuria was 0.35 G/24 h. The urinary sediment was bland. Ultrasonography disclosed two symmetrical, moderately atrophic kidneys.

The diagnosis was made at fist glance.

What is your answer?

Pringle's adenoma sebaceum and Koenen's tumours led to a diagnosis of Bourneville tuberous sclerosis involving the kidneys.

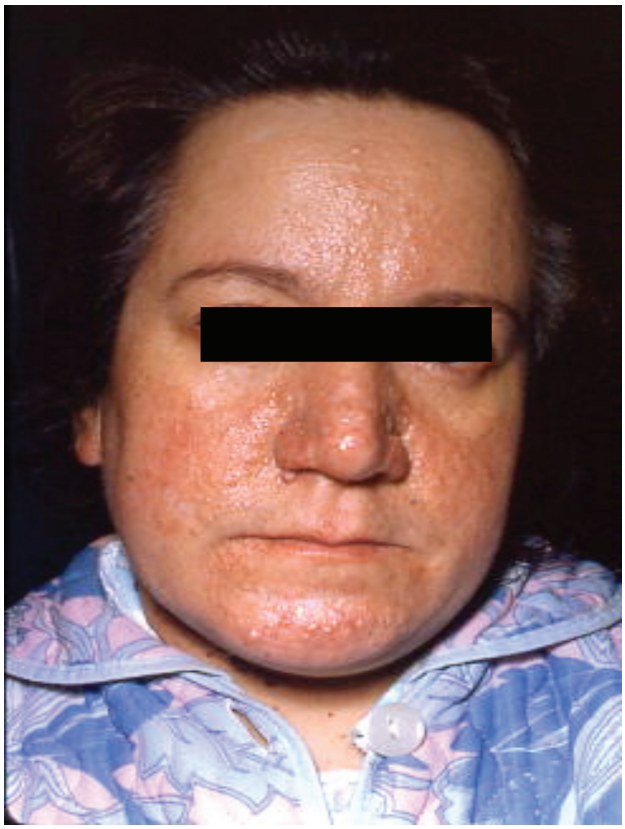

Fig. 1. Pringle's sebaceous adenomas (angiofibromas).

Bourneville tuberous sclerosis

The Bourneville–Pringle complex was partly described at the end of the 19th century [1] and fully identified as a 'phakomatosis' in 1999. It is a hamartomatous disease with localizations in various organs and tissues [2]. Two autosomal dominant responsible genes (hamartin, TSC1; tuberin, TSC2) have been identified. They function as a cytoplasmic protein complex with inhibitory effects on cell growth, proliferation, adhesion, trafficking and migration [3]. The disease, however, can be sporadic. Amongst the pathological localizations of tuberous sclerosis, the kidney may be involved by hamartomas. Histology discloses angiomyolipomas, benign cysts and, much less often, lymphangiomas [4]. The angiomyolipomas are composed of mature adipose tissue and smooth muscle fibres, and are richly vascularized by thick-walled blood vessels. The histopathologic diagnosis should never be made by a kidney biopsy that may be complicated by severe perirenal bleeding. The renal disease may be clinically silent. High renin hypertension is not uncommon. Spontaneous perirenal haematomas may occur and require haemostasis by an interventional radiologist. Bilateral forms with progressive destruction of the renal tissue are rare. Progression to renal cancer (or the co-existence of a malignant tumour) has been described.

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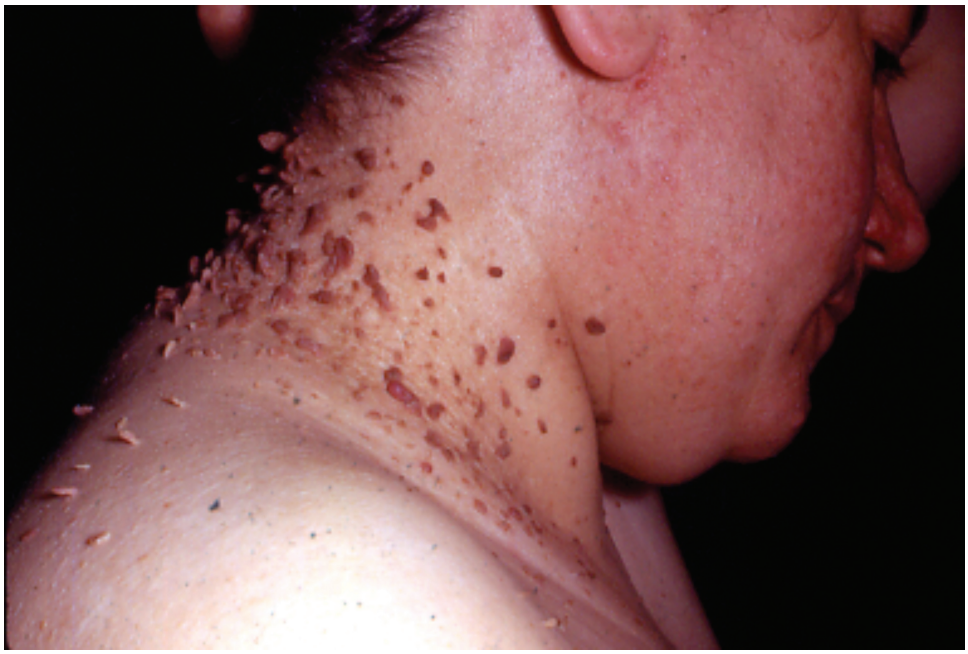

Fig. 2. Widespread cutaneous soft fibromas.

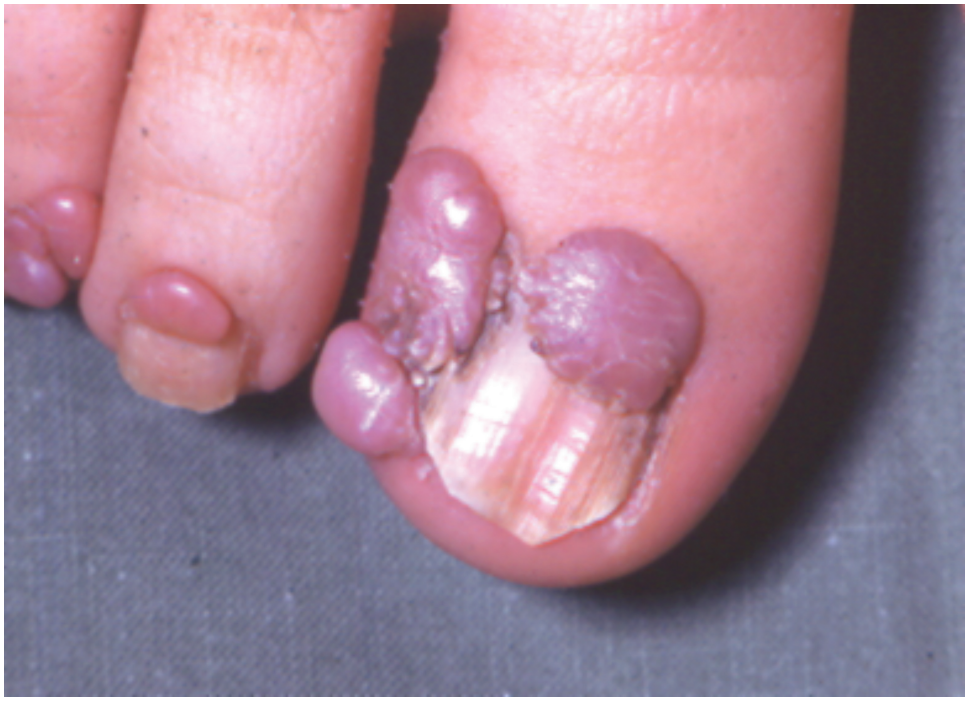

Fig. 3. Periungual Koenen's tumours (feet).

Return to the case

The patient's son, aged 8 years, was examined and had some minor skin hamartomas, indicating that this lady suffered from the genetic form of the disease. She was submitted to bilateral nephrectomy and successfully transplanted.

Conflict of interest statement. None declared.

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