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QUIZ SECTION

Recurring Axillary, Abdominal and Genitofemoral Nodules and Abscesses: A Quiz

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A 22-year-old woman was referred with the diagnosis hidradenitis suppurativa. The primary complaint was recurring painful and inflamed lesions of the axillae. Minor asymptomatic lesions had persisted from early childhood, but for a couple of years the condition had worsened with more frequent inflammation and pain. The patient's father and sister had experienced the same symptoms. In her siblings treatment with isotretinoin has been attempted with varying effect. Microbiological investigation of lesions initially showed no growth of bacteria or fungi.

On examination, the patient presented with tender nodules and suppurating abscesses in the groin, the axillae (Fig. 1), proximal extremities, the neck, on the abdomen and the scalp. When punctured elements oozed creamy/oily fluid.

A skin biopsy revealed cystic structures with squamous epithelium without granular layer. Sebaceous glands near the cyst wall. Cysts contained fragments of hair. There were no signs of malignancy.

What is your diagnosis? See next page for answer.

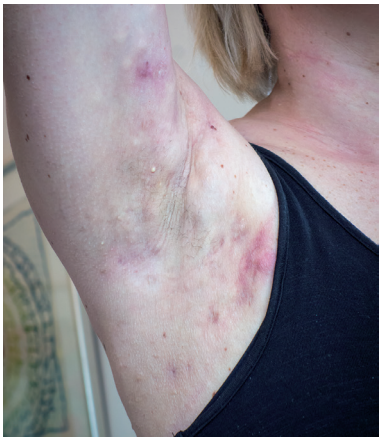

Fig. 1. Tender nodules and scar tissue in the axilla.

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ANSWERS TO QUIZ

Recurring Axillary, Abdominal and Genitofemoral Nodules and Abscesses: A Comment

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Diagnosis: **Hereditary steatocystoma multiplex**

First described by Jamieson (1), the dome shaped translucent yellow nodules in axillae, groin, on chest and abdomen are signs of the rare genetic disorder steatocystoma multiplex (SM) (2), ICD10: L72.2, ORPHA841. As the disease can develop with inflammation and suppuration due to rupture of the cysts (steatocystoma multiplex suppurutiva) it is a relevant differential diagnosis to hidradenitis suppurativa (HS). SM can present from early childhood, but most often only present with more serious symptoms in adolescence (3). An autosomal dominant (4) pattern of inheritance has been described.

Many physicians had seen the patient since her inflammatory lesions started in her early teens. Lesions were interpreted as boils and incised. At the age of 22 she was referred to a dermatological department with the diagnosis of HS. The location of lesions (involving neck, axillae and abdomen), the multiple monomorphic homogenous lesions and histology provided the correct diagnosis.

Multiple benign sebaceous gland tumours dominate the presentation of SM. The lesions are naevoid configurations of ineffective hair follicles located at the ducts of sebaceous glands. The endocrine stimulation of sebaceous glands triggered by puberty may be linked to the development of clinical disease (3). Keratin 17 abnormalities have been described in familial steatocystomas and in another rare genetic disorder, pachyonychia congenita type 2 (PC-2), thus linking the 2 disorders. However, PC-2 might involve additional mutations and it is therefore speculated that SM and PC-2 are different phenotypic expressions in a spectrum representing the same disorder (5).

Our patient had never had her DNA analysed.

The characteristic histological findings are patognomonic: thin cystic spaces with a corrugated luminal surface, eosinophilic stratum corneum and absence of granular layer. Cysts contain keratin, vellus hairs and sebum (Fig. 2). Cases of malignant degeneration have been described (2).

Palliative medical treatment shows varying rates of success and can involve tetracycline or isotretinoin. For localised inflamed lesions, fistulae, suppurating elements and problematic scar tissue excisional surgery is indicated. Cryo-therapy and CO_2-laser evaporation may be used to destroy non-inflamed lesions (6). Palliative treatment of localised inflamed lesions is possible with intralesional triamcinolone injections (3–5 mg/lesion).

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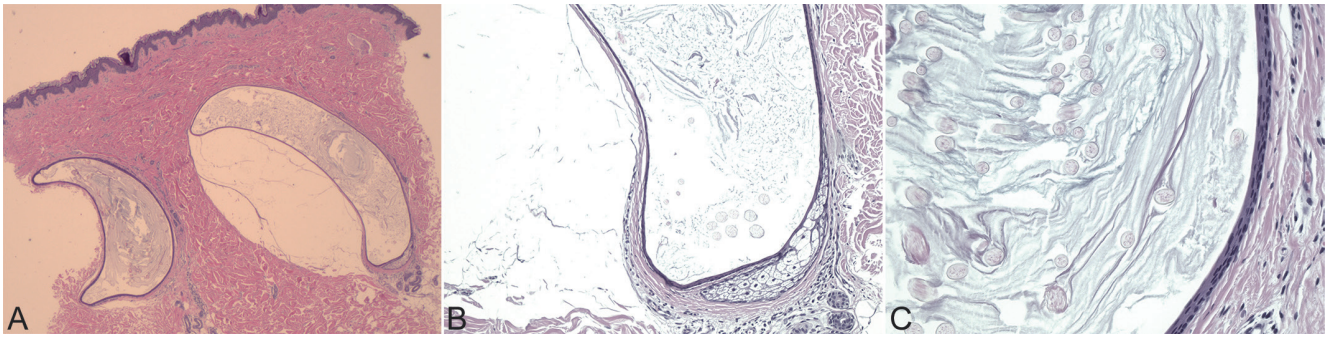

Fig. 2. Pathognomic histological findings: thin cystic spaces in the dermis. Histopathological examination (A, H&E stain, low power view). Eosinophilic stratum corneum and absence of granular layer. Sebaceous glands within the cyst wall (B, medium power view). Cysts contain keratin, vellus hairs and sebum (C, high power view). H&E-staining.