Polygenic Heredity of Porphyria cutanea tarda

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Sir,

We read with great interest the paper by Gandolfo et al. [1] published in your journal. We fully agree with the authors that their findings of normal erythrocytic urodecarboxylase (UD) activity in patients with familial porphyria cutanea tarda (PCT) and the recently published analogous cases [2] contradict the currently widely accepted view, which strictly separates the sporadic form of PCT from the familial form on the basis of the UD deficiency [4]. Held et al. [3] pointed out this contradiction too on the basis of their finding that in 5 out of 32 PCT patients without a positive family history of PCT the UD activity was approximately half the normal level. Further, symptom-free persons with a significantly decreased UD activity are also known. These cases lead us to another conception which predicts polygenic heredity for the disease and supposes that the sporadic and familial forms of PCT are the two end-poles of one and the same disease, which differ from each other only with regard to the severity of the genetic defect [5].

Polygenic heredity is characterized by genetic defects which, together with environmental factors, are responsible for the disease. The genetic background involves several genes and the alteration of a certain number of this set of genes (limit point of genetic determination) results in the genetic determination of the disease. The main characteristics of polygenic heredity were described by Carter [6].

In our investigations of the role of genetic and hepatotoxic factors in the development of PCT in 51 patients we have found that the rules of polygenetic heredity fit this disease well [5]. The present findings

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regarding the UD deficiency also correlate well with this conception. Accordingly, we presume that PCT belongs among the diseases of poly-genic heredity, where the gene coding UD is only one member of the PCT-responsible gene set. Thus, a genetic predisposition for PCT can occur both with and without the defect of the UD gene. The repeated appearance of the disease within a family will depend on the degree of the genetic alteration, and thus both UD-altered and UD-unaltered cases can occur in a familial form. Since PCT occurs more frequently in the same family in UD-deficient cases, it may be supposed that the gene coding UD is of unique and outstanding importance among the PCT-responsible genes. Thus, a defect of the UD gene results in a situation where it is much easier to reach the PCT-determining limit point of the genetic

alteration. We believe that the hypothesis of polygenic heredity of PCT gives further possibilities for the understanding of this disease.

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Koebner Phenomenon in Lichen sclerosus et atrophicus

We examined a woman with lichen sclerosus et atrophicus (LSA) with manifestations in two scars. As only a few reports on the appearance of Koebner phenomenon (KP) in LSA [1,2] have been published upon yet, we should like to inform you about our case.

Fig. 1. LSA in scar after cholecystectomy.

A 55-year-old woman had an appendectomy in 1964 and a cholecystectomy in 1976. The skin affection appeared in 1985 simultaneously on the inner aspects of both wrists, then in a recent scar after an incised wound (3 months old), and in an older scar in the abdominal wall after cholecystectomy (9 years old, fig. 1). The affection consisted of whitish, flat papules, 1-4 mm in diameter, with a coarse surface, which on the wrists merged into sharply demarcated irregular lesions. At the scar sites their configuration was linear, and on the abdomen, with accessory punctated lesions in the scars after stitches as well as outside them. The papules itched, and after dissemination showed no progression. There were no genital lesions. Histological findings were typical for LSA (fig. 2).

The etiology of LSA is unknown, lesions usually appearing spontaneously without any precipitating factor. A preceding infection is supposed to play a provocative or localizing role in some cases of the genital affection. LSA has been reported in a vaccination site [1] and following trauma [2]. We have diagnosed LSA in two scars.

Scars are sites of localization to psoriasis vulgaris, lichen planus and vitiligo. Other affections are localized there less frequently: mastocytosis [3] and erythema multiforme [4]. Probably not the localization but the age of the scar is of importance. In recent scars KP is more frequent.