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Punctate Palmoplantar Keratoderma (Brauer-Buschke Fischer Syndrome) and Pituitary Adenoma

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Abstract The type 1 punctate palmoplantar keratoderma/ Brauer-Buschke-Fischer syndrome (PKK 1) is a rare genodermatosis with a prevalence estimated at 1.17/100000. We report the case of a 70-year old patient, with type 2 diabetes, who presents a PPK 1 since the age of 30. Examination revealed an important family inbreeding, a significant family history as 38 members were affected by this disease: it was associated with different tumors (colorectal cancer, hepatocellular cancer and melanoma). Clinical examination showed multiple papular hyperkeratotic lesions with variable diameter between 2 and 10 mm on the palms and the soles. The histology confirmed the diagnosis of punctate palmo-plantar keratoderma. This patient presented at the age of 65 an acute adrenal insufficiency. Different explorations showed a non-secreting necrotic pituitary adenoma. In our knowledge, it is the first observation of association of PKK1 and pituitary adenoma. Its pathophysiological mechanism is still unclear. More studies are needed to will have to be clarify it.

Keywords: *punctate palmoplantar keratoderma (Brauer-Buschke-Fischer syndrome), pituitary adenoma*

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1. Introduction

The type 1 punctate palmo-plantar keratoderma type 1 (KPP1), also called Buschke-Fischer-Brauersyndrome is a rare hereditary skin disorder. It is marked by impaired epidermal differentiation leading to hyperkeratosis on the palms and the soles. It has autosomal dominant pattern of inheritance with variable penetration [1]. Its prevalence is estimated at 1.17/100000 [1]. Its age of onset varies from 12 to 30 years. It is marked by an excessive thickening of the epidermis of the palms and the soles. This lesion can touch a part or the whole of the palmoplantar surface. The KPP1 can be isolated or associated with other skin or extra-cutaneous manifestations essentially neoplasm lesions.

2. Observation

We report the observation of 70 – year old patient. He has diabetes since 7 years. He was initially treated by on metformin and glibenclamide. Because of its hepatotoxicity, glibenclamide was replaced by insulin. He hasalso moderate chronic renal failure (creatinine 50 clearance ml/min) complicating bilateral nephrolithiasis.

Table 1. The results of the patient hormonal tests

Hormonal Tests	Results	Range
Synacthene ®1 µg Test: Cortisol T0 Cortisol T60	 28,7 ng/ml 129 ng/ml	 70-250
Free T4 TSH	9 pmol/l 2.8 µU/ml	9,6-29 0.15-5.0
FSH LH Testosterone	7.6 UI/L 5.9 UI/L 2.8 ng/ml	1.4 - 18.1 1-7 2-10
Prolactin	3.2 ng/ml	2-15
IGF1	20 µg/ml	70-300

He was admitted in intensive care unit because a severe hypovolemic shock after gastroenteritis. He had an arterialpressure at 80 mmHg/ 60 mmHg with moderate regular tachycardia up to 100 pulse per minute. The examination showed notableskin and mucosal pallor, macroglossia and diffuse cutaneous infiltration. He had received insulin supplementation, hydration and adequate antibiotics. After several days of treatment, his arterial pressure had remained low under 100 mmHg/ 60 mmHg, with hyponatremia and normal kaliemia: hypothyroidism and adrenocortical insufficiency were suspected.

Later, his status had improved dramatically. After resuscitation, the hormonal tests had revealed multiple pituitary hormone deficiency, including corticotroph, thyrotroph, somatotroph, and gonadotroph deficiencies

(Table 1). The MRI exploration, looking for pituitary lesions, had shown the presence of a necrotic pituitary macroadenoma, the cavernous sinus and the pituitary stalk were of normal appearance (Figure 1 and Figure 2).

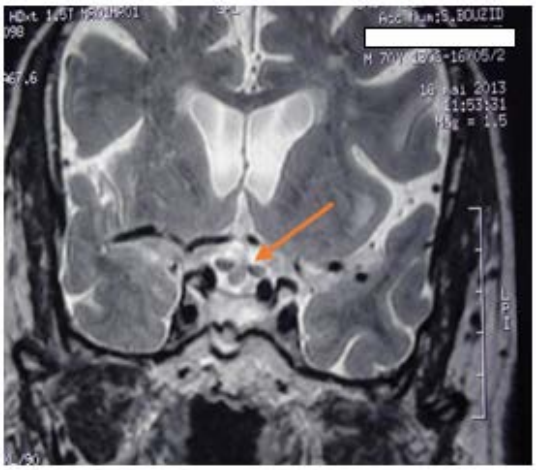

Figure 1. Pituitary MRI showing a pituitary macroadenoma

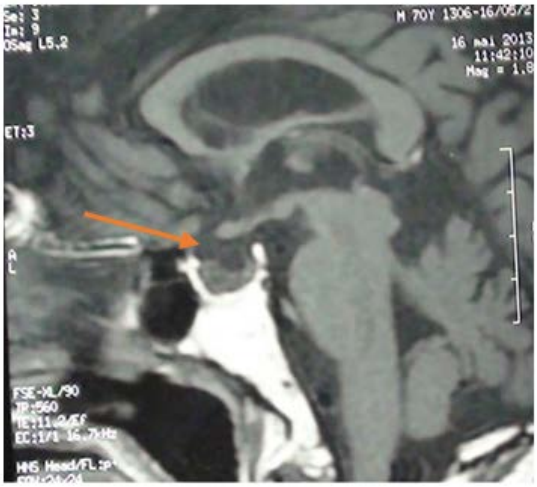

Figure 2. Pituitary MRI showing a pituitary macroadenoma

Clinical examination has also shown the presence of palmoplantar hyperkeratosis: multiple popular hyperkeratotic lesions with variable diameter between 2 and 10 mm (Figure 3 and Figure 4). These lesions had appeared at the age of 35 years and aggravated by manual labor. No exploration or treatment was carried out. The research of similar cases in the family had revealed the presence of the KPP 1 in many other family members over four generations. It was associated with colorectal cancer, hepatocellular carcinoma and melanoma in different family members (Figure 5). The skin biopsy with histological study showed considerable hyperkeratosis, with acanthosis and thickening of the granular layer without any sign of malignancy (Figure 6 and Figure 7).

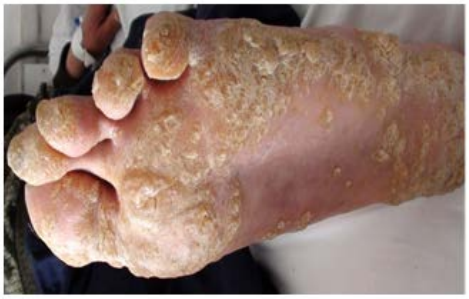

Figure 3. Plantar hyperkeratosis

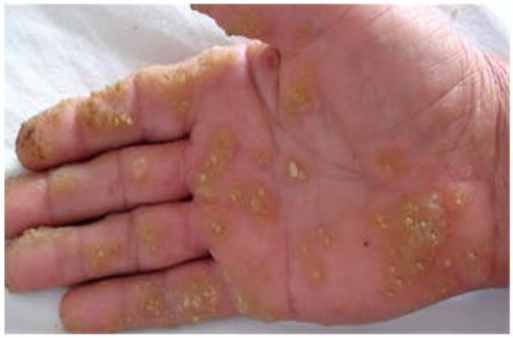

Figure 4. Palmar hyperkeratosis

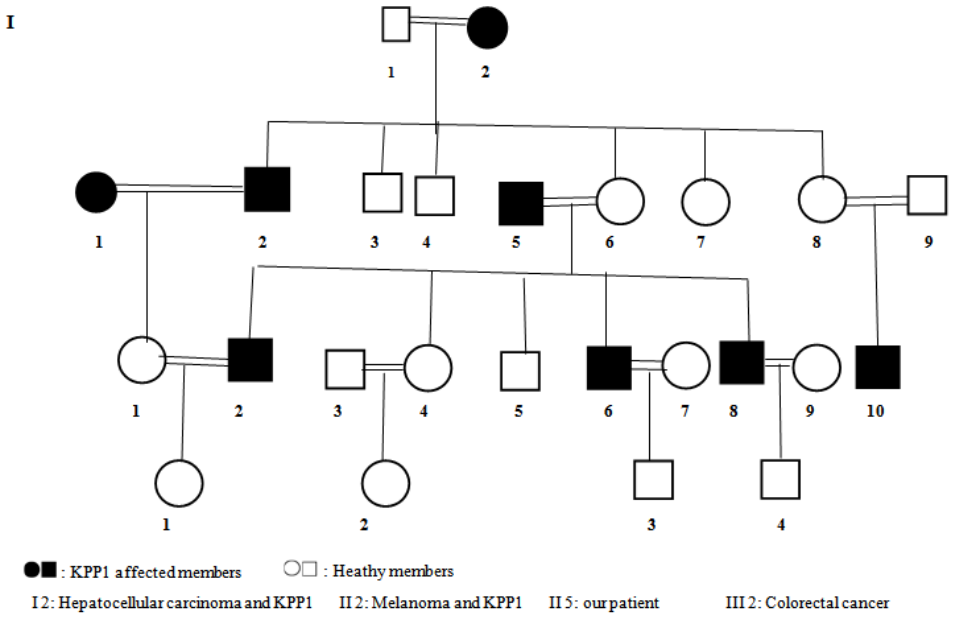

Figure 5. Family tree showing affected members in different generations

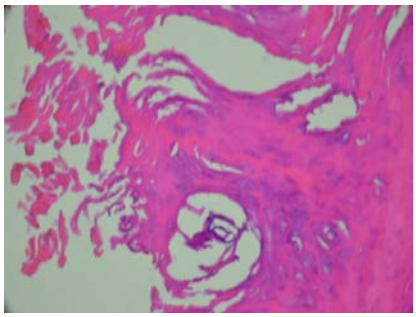

Figure 6. Histological sections of the skin biopsy of the lesions of palmar and plantar hyperkeratosis

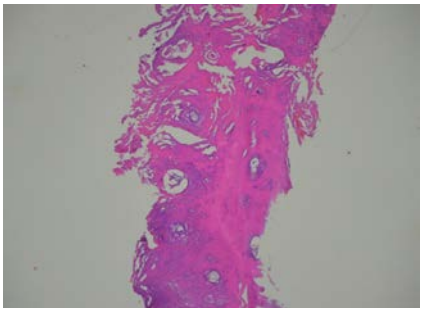

Figure 7. Histological sections of the skin biopsy of the lesions of palmar and plantar hyperkeratosis

3. Discussion

The KPP1 is a rare hereditary skin disease. It is characterized by epidermal differentiation impairment causing the appearance of multiple hyperkeratotic papules distributed irregularly on the palms and the soles [2,3,4].

Its pathophysiology is still unclear. However the combination of impairment of genetic and environmental factors is most likely. The transmission is often autosomal dominant, rarely autosomal recessive. [5,6,7]. This is the case of our patient.The aggravation by manual work is well demonstrated [8].

Giehl et al. reported in 2012, for the first time the exact pathophysiological mechanism of the disease: two heterozygous nonsense mutation of the AAGAB gene located on chromosome 15 were found in all affected individuals of the study [2].

In histology, the dermis is often normal without inflammatory reaction. On the other hand, there is marked hyperkeratosis, parakeratosis, and mild acanthosis (Figure 5), which is consistent with our results [9]. The age of onset varies from 12 to 30 years which is the case of our patient and the various members of his family [10]. Indeed since 1980, many families with a KPP1 have been reported (Table 2) [5,6,8,11,12,13].

Table 2. Different cases of KPP 1 in the literature

Author	Year	Transmission	Cases number	Associated diseases
Powell FC, et al. [11]	1983	Autosomal dominant	10 patients	spastic paralysis
Bennion SD, et al. [12]	1984	Autosomal dominant	8 patients	Adenocarcinoma of colon and pancreas
Nielson PG [8]	1988	Autosomal dominant	4 patients	Morbus Bechterew et HLA B 27 positive
Steven HP, et al. [13]	1996	Autosomal dominant	49patients	Hodgkin lymphoma, Adenocarcinoma of colon and pancreas, Breast and kidney cancer
Emmert S, et al. [5]	2003	Autosomal dominant	47 patients	Palmoplantar Hyperhidrosis
Guptal R, et al [6]	2003	Autosomal dominant	17 patients	Bilateral dystrophy of the big toes

Data from the literature reported the association of this form with neurological or rheumatologicaldisorders (spastic paralysis, ankylosing spondylitis) and other dermatoses (Sebaceous Glands Hyperplasia, nails disorders…) [1,6]. The association with malignant tumors is frequently reported. It is mainly represented by gastrointestinal tumors (colon, pancreas, esophagus...), lung tumors, lymphoma [2,6,11]. Systematic screening of these diseases is indicated for all members of the family with the disease. In our knowledge, the association of KPP1 with a pituitary adenoma is reported for the first time.

4. Conclusion

The KPP 1 is a rare inherited skin disease. Its association with an increased risk of tumors, neurological, rheumatological and other cutaneous disorders is known. We report in this study, for the first time, the association of the KPP1 a pituitary adenoma. Subsequent work its necessary to detail the pathophysiologic mechanism of this association.

Conflict of Interest

None declared.

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