Pachyonychia Congenita Project

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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.

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Dermatological Research

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Pachyonychia congenita (Jadassohn–Lewandowsky syndrome) – evaluation of symptoms in 36 patients

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Since hereditary palmoplantar keratoderma (PPK) of the Jadassohn–Lewandowsky type (HPPK–JL, pachyonychia congenita) was first described by Müller in 1904 [5] and later by Wilson in 1905 [8] as well as by Jadassohn and Lewandowsky in 1906 [3], approximately 180 cases have been reported. Even assuming that a certain number of patients are going undetected, the conclusion is justified that HPPK–JL is a rather rare pathological condition. In sharp contrast to this situation is the fact that this condition has been relatively frequently observed in Slovenian [2] and Croatian [7] population. In Slovenia, with a total of 1.86 million inhabitants, 11 cases have been observed giving a prevalence of 0.7 cases per 100000 inhabitants, while in Croatia, with 4.7 million inhabitants, 25 cases give a prevalence of 0.53 per 100000.

As a result of these observations an attempt was made to analyse and discuss the different clinical symptoms characteristic of HPPK–JL

Our extensive clinical studies have confirmed the observation that HPPK–JL is not a completely unique hereditary disease, but rather a syndrome comprising a spectrum of clinical symptoms that are not all displayed in any one patient: thickening of the nail plates, hyperkeratosis of palms and soles, follicular hyperkeratosis, hyperhidrosis, blisters, oral leucokeratosis, corneal changes, premature dentition, laryngeal lesions, hoarseness, hair abnormalities and cicatricial alopecia. This kind of approach is reflected in the classifications of HPPK–JL proposed by various authors, for example Kumer and Loos [4], Schönfeld [6] and Feinstein et al. [1].

The data used for our analysis were the most frequent clinical symptoms as recorded in the medical records of

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Table I. Review of symptoms in 36 patients with HPPK–JL in Slovenia and Croatia

Family	Generation	PPK	P	FK	HY	LK	B
1	I	+	+	+	0	0	0
	II	+	+	+	+	+	+
2	I	+	0	+	0	0	0
	II	+	+	+	+	0	+
	II	+	+	+	+	0	+
	II	0	+	+	0	0	0
3	II	+	+	+	0	+	0
	III	+	+	+	0	+	0
	III	+	+	+	+	+	0
4	I	+	+	+	+	+	+
5	III	+	+	0	+	0	0
6	I	0	0	+	0	0	0
	I	+	0	0	0	0	0
	I	+	0	0	0	0	0
	II	+	+	+	+	+	+
7	I	+	+	+	+	+	0
	II	+	+	+	+	+	0
	III	+	+	+	+	+	+
8	I	+	+	0	+	0	0
	II	+	+	0	+	0	0
	III	+	+	0	+	0	0
	III	+	+	0	+	0	0
9	II	+	+	+	+	+	+
10	I	+	0	0	0	0	0
	I	+	+	+	+	0	0
	II	+	0	0	0	0	0
	II	+	0	0	0	0	0
	II	+	+	+	+	0	0
	III	0	+	0	0	0	0
11	II	+	+	+	+	+	0
	III	+	+	+	0	0	0
12	I	+	+	0	0	0	0
	II	+	+	0	0	0	0
	II	0	+	0	0	0	0
	III	+	+	+	+	0	0
	IV	0	0	+	+	0	0

PPK, palmoplanar keratoderma; P, pachyonychia; FK, follicular hyperkeratosis; HY, hyperhidrosis; LK, leucokeratosis; B, blisters

11 Slovenian and 25 Croatian patients: PPK pachyonychia, follicular hyperkeratosis, hyperhidrosis, leucokeratosis and blisters. These 36 patients belonged to 12 families, five in Slovenia and seven in Croatia. A statistical evaluation of various combinations of symptoms was carried out.

The 36 cases of HPPK–JL represent a fairly large number of patients sufficient for statistical analysis. The six most frequently observed symptoms were selected for evaluation. A review of the symptoms is presented in Table 1. The most frequently appearing symptom was PPK, followed by pachyonychia, follicular hyperkeratosis, hyperhidrosis, leucokeratosis and blisters. These results are presented in detail in Table 2. The appearance of other symptoms was related to the presence of PPK (as the most constantly present symptom) and the conditional probabilities were calculated. Pachyonychia and PPK together were present in 78.07% of patients, this was followed by follicular hyperkeratosis and hyperhidrosis. The details are given in Table 3. A majority of the families show a dominant transmission of the trait, but from Table 1 it can be seen that, even in the same family, there is considerable clinical variability of disease expression.

Table 2. Frequency of symptoms in 36 patients with HPPK–JL

PPK	32/36
Pachyonychia	25/36
Follicular hyperkeratosis	19/36
Hyperhidrosis	19/36
Leucokeratosis	11/36
Blisters	7/36

Table 3. Conditional probabilities of appearance of certain symptoms if PPK present

Pachyonychia	78.07%
Follicular hyperkeratosis	59.39%
Hyperhidrosis	59.39%
Leucokeratosis	34.42%
Blisters	21.82%

From this rather large series of cases (36 patients) it is possible to conclude that the symptoms in HPPK–JL can be classified into major ones and minor ones. The major symptoms are PPK, pachyonychia, follicular hyperkeratosis and hyperhidrosis. The minor symptoms are leucokeratosis and blisters. Hoarseness was observed in only one patient and cicatricial alopecia in one patient; these two symtoms were not statistically evaluated, but they deserve to be included in the minor group. The results obtained in the present clinico-genetic study do not favour previous attempts at classification, e.g. that of Feinstein et al. [1], so we suggest that the classification into major and minor symptoms is more useful for clinical needs.

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