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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.
The Jadassohn-Lewandowsky Syndrome *

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In 1906, Jadassohn and Lewandowsy first described [2] a combination of symptoms of dominantly inherited ectodermal dysplasias [5]. The syndrome was later on named Jadassohn-Lewandowsky Syndrome (JLS). Common symptoms include pachyonychia, focal palmar and plantar hyperkeratoses, and follicular hyperkeratoses of the epidermis. These main symptoms often associate abnormalities such as leukokeratosis of the oral mucosa, blistering on various places on the body, hyperhidrosis, irregularities of hair growth and structure – most frequently reduced growth and dystrophy – and dental deficiencies, in rare cases absence of single teeth [8].

Case report

66-year-old female patient R. F. has suffered from increasing dryness of the oral mucosa, joined by intense burning of the tongue since fall 1971. The internist Prof. Dr. Kleinschmidt, Director of the 2. Medical Hospital of the LMA, performed an examination that provided no pathological explanation for the symptoms. Symptoms varied in the course of the disease and are therefore presented in chronological order:

Patient has presented deformed fingernails and brittle toenails since birth. As far as she remembers, she has suffered from epiphora on the left eye since childhood. The epiphora is caused by partial atresia of the left nasolacrimal canal. Surgical correction of the irregularity had been planned in earlier years but was never performed because the necessary surgical dilation of the narrow outer nasal ostia had not produced the expected result. At first, the narrow nasal ostia did not cause her any trouble. Only later on, when she also suffered from

* dedicated to the memory of Prof. H. Koehler on 23 October 1973
increasing dryness and burning of the oral mucosa, did she complain about impaired nasal breathing.

Since 1940, she has repeatedly presented lentil- to coin-sized hyperkeratoses on the pressure points of palms and soles. Some of these hyperkeratoses broke open and became inflamed. At the same time, blistering occurred on both lower legs. The resulting open sores heal very slowly. This process recurred in the following summers, but was limited to the distal third of the lower left leg.

Patient has always had sparse hair and teeth with a strong tendency towards caries. She lost all her teeth between age 20 and 40.

*Family anamnesis.* The father’s paternal grandfather is said to have had similarly deformed fingernails. The patient’s mother and maternal grandmother, the patient herself and her grandson suffered from epiphora. There is no indication that other family members had symptoms similar to the patient’s (Fig. 1).

*Findings.* On her admission on May 23, 1972, general and nutritional condition of the 66-year-old patient was reduced. No pathological findings after general clinical examination.

*Special findings.* All fingernails were considerably thickened from proximal to distal, rolled in laterally, lengthwise not curved, with slight yellowish discoloration, but transparent, lunules were absent. The nails of the first two toes were shaped like the fingernails, the other toes had no nails (Fig. 2).

The pressure points of palms and soles showed lentil- to coin-sized, very hard, flat, tubercular hyperkeratoses (Fig. 3).

Irregularly circumscribed palm-sized areas of brown-red colored atrophic skin on the distal third of both inner lower legs, on the left leg more pronounced than on the right.

Oral mucosa overall very dry; extensive rough hyperkeratoses on the tongue similar to a hairy tongue. Soft palate showed adherent, brown calluses. Dry lips with light fissures in the corners of the patient’s mouth. Indistinctly shaped, livid reddening around both eyes. Lacrimal fluid ran down left over the cheek. Lacrimal point present, lacrimal canal could not
be found. No other pathological findings on the outer and inner eye, and in particular no corneal abnormalities.

Sparse scalp hair, no areas of alopecia. Patient had lost all her teeth.

Nostrils were very narrow, nasal breathing was impaired and painful because of an inflammatory process in the left nasal cavity.

**Discussion**

Apart from the mentioned dysplasias, that appear to be frequent in JLS, some malformations of ectodermal and non-ectodermal origin are not so frequent: Changes of the cornea and lenses, intellectual deficiencies, retarded development of the genital organs and various, varying irregularities of the skeletal system [8]. In their two primary observations, Jadassohn and Lewandowsky described symptoms resembling granulosis rubra of the nose. According to Moldenhauer [8], only two more of the 93 cases she analyzed and evaluated were described in the following years.

Because of the wide variety of symptoms of JLS, Kumer and Loos [5] have classified the disease into three categories:

Type 1: Pachyonychia congenita with symmetrical hyperkeratoma and follicular hyperkeratoses on the epidermis,
Type 2: (also Riehl type): type 1 symptoms with leukokeratosis oris,
Type 3: type 1 symptoms with corneal abnormalities.

The classification of monosymptomatic forms proves difficult, in individual cases the family anamnesis permits greater differentiation.

With pachyonychia congenita and the insular hyperkeratoses on palms and soles, our patient showed two main symptoms of JLS. Sparse hair growth and the predisposition to bad teeth are indications of a syndrome of ectodermal malformations and are reported in several JLS cases [4, 8, 9].

The patient described recurring blistering on both lower legs. We only observed the scarry residues, but in all probability the blistering is part of JLS. Moldenhauer [8] found that in 35
of 93 cases mentioned in literature, patients repeatedly presented blisters on the most varied parts of the body. Very frequently, keratoses occurred on the soles and on other pressure points.

The course of the changes in oral mucosa that our patient suffered from indicates that these changes are not related to JLS. According to the anamnesis, such oral abnormalities had only occurred in the past two years. In 1958, the patient was examined in a hospital for dermatology because of the blistering on the lower legs. When a blister was found on the oral mucosa, she was suspected of having pemphigus vulgaris. Leiber-Olbrich [6] mentions blistering on the oral mucosa as one of the symptoms of JLS. The changes in oral mucosa that eventually led to her hospitalization in 1972 result from her continuous mouth breathing. Nasal breathing was almost completely impossible because the outer nasal ostia were very narrow. Nasal ostia were dilated in plastic surgery performed by Prof. Jatho, Medical Academy Lübeck Hospital for ENT, on 14 June 1972. Since then, the patient can breathe freely through the nose. During a follow-up examination in January 1973, oral mucosa presented well moistened and free of hyperkeratoses, and the tongue burning had disappeared.

The symptom of very narrow outer nasal ostia has not been described in any other JLS case [6,8], nor has it been observed in other syndromes of ectodermal abnormalities. The same applies to the partial atresia of the nasolacrimal canal that our patient presented.

Summarizing reports of nasal and ocular malformations [3, 7, 10] provided no description of these two symptoms occurring simultaneously. Thus, our theory on common pathogenetic features is not supported by similar observations. During the embryonal period, nasal ostia and nasolacrimal canal develop during a joint stage of cleavage, which includes the lateral nasal process and forms an ectodermal inversion [1].

Summary. Description of a JLS case. Apart from the typical combination of symptoms, the 66-year-old patient presented with partial atresia of the nasolacrimal canal and changes in oral mucosa. These changes resulted from impaired nasal breathing caused by narrow outer nasal ostia.
Figure 1. Family tree of the patient R. F.

- Patient
  - normal
  - JLS
  - x epiphora

Figure 2. Deformed finger nails

Figure 3. Hyperkeratoses on the soles
Bibliography


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