Spontaneous Fading of Reticular Pigmentation in Naegeli-Franceschetti-Jadassohn Syndrome

Peter H. Itin¹, Bettina Burger²,³
Departments of ¹Dermatology and ²Biomedicine, University Hospital Basel, Basel, Switzerland

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Naegeli-Franceschetti-Jadassohn (NFJ) syndrome is a rare symptom complex out of the spectrum of ectodermal dysplasia that affects the skin and its pigmentation, sweat glands, nails, hair and teeth (OMIM 161000). NFJ syndrome is inherited in an autosomal dominant fashion and is caused by mutations in the KRT14 gene localized on chromosome 17q11.2–17q21. The disease is allelic to dermatopathia pigmentosa reticularis [1]. Prevalence is roughly estimated to be 1 in 3 million.

The main clinical findings are absence of dermatoglyphs, reticular or mottled hyperpigmentation, hypohidrosis with diminished sweat gland function and discomfort provoked by heat, nail dystrophy with congenital malalignment of the great toe nail, enamel defects of the second teeth with resulting early caries, and moderate hyperkeratosis of the palms and soles. Diffuse palmo-plantar keratoderma may coexist with punctate keratoses that are sometimes accentuated in the creases or exhibit a linear pattern [2].

Reticulate hyperpigmentation starts around the age of 2 years without a preceding inflammatory stage. The pigmentation is brown and gray-brown and is localized on the trunk, proximal extremities, axillae, groins and flexures as well as in the perioral and perioral regions. The pigmentation shows a gradual increase during the first 10 years of life, and fading starts around the age of 15 years. Patients with NFJ syndrome who are older than 70 years only have minimal or no pigmentation left [3].

Although patient history gave this information on fading, no photographic documentation of this natural course has previously been presented in the literature. We describe the natural course of hyperpigmentation in a patient of the original family with NFJ syndrome.

Our female patient was born in 1944 with NFJ syndrome. Her mutation in keratin 14 had been confirmed previously [1]. The patient had always had difficulties in hot temperatures because of her limited ability to sweat. A pilocarpine test in this patient had been performed by Franceschetti and Jadassohn and was only weakly positive [4]. She had impressive enamel defects and total prostheses were necessary at the age of 30 years. The patient had moderate diffuse palmo-plantar keratoderma but her nails were unaffected.

Fig. 1. At the age of 40 years, marked perioral pigmentation was present (previously shown and reprinted with permission from Elsevier).

Fig. 2. At the age of 65 years, complete resolution of the reticular perioral pigmentation was seen.
At the age of 40 years, the patient was documented in our clinic and still had marked hyperpigmentation especially around the mouth (fig. 1). In the year 2009, the patient was seen again in our department, and interestingly pigmentation around the mouth but also in other regions had faded completely without medical intervention (fig. 2). The patient had applied no camouflage or make-up. This is the first photographic documentation of total involution of hyperpigmentation in NFJ syndrome.

References


Prof. Peter Itin
Department of Dermatology, University Hospital Basel
CH–4031 Basel (Switzerland)
Tel. +41 61 265 4084, Fax +41 61 265 4200
E-Mail peter.itin@unibas.ch