Chapter 12
Atopic Dermatitis, Hidradenitis Suppurativa and Poikiloderma

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A seven-year-old girl presented with classil atopic dermatitis (eczema on the upper and lower limb folds, xerosis, history of wool intolerance and high temperatures, elevated IgE, allergic rhino-conjuntivitis) since six months of age. Hidradenitis suppurativa (acne inversa) had also appeared on the perigenital and groin areas for a year. This was the main reason for the dermatological consultation.

The patient was born at full term but with low weight. Slow weight gain, short stature, and teeth retardation were reported by her parents. Worsening of the facial erythema following sun exposure was also noticed.

The physical examination confirmed the reported dermatological diagnoses (Figs.12.1 and 12.2) but we also notited atrophic and dischromic lesions on the cheeks and extensor regions of the limbs (Figs. 12.3 and 12.4).

The examination of the oral cavity highlighted dental abnormalies referable to micro and hypodontia (Fig. 12.5). Laboratory tests documented only high level of IgE.

Based on the Clinical History and the Photographs Which Is Your Diagnosis?

1. Lupus erythematosus (LE)
2. Rhotmund Thomson syndrome
3. Kindler syndrome
4. Dyskeratosis congenital

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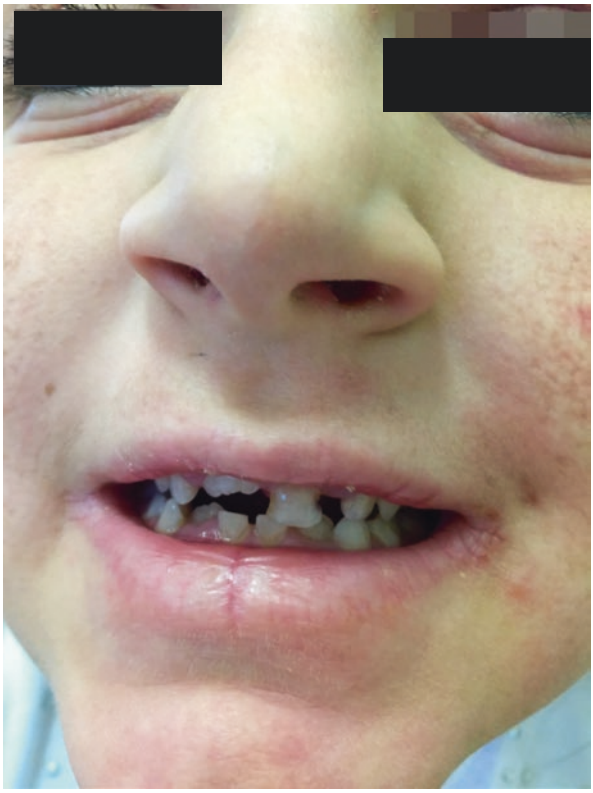

Fig. 12.1 Dennie-Morgan folds on lower eyelids (diagnostic marker of atopic dermatitis), cheilitis and perioral dermatitis

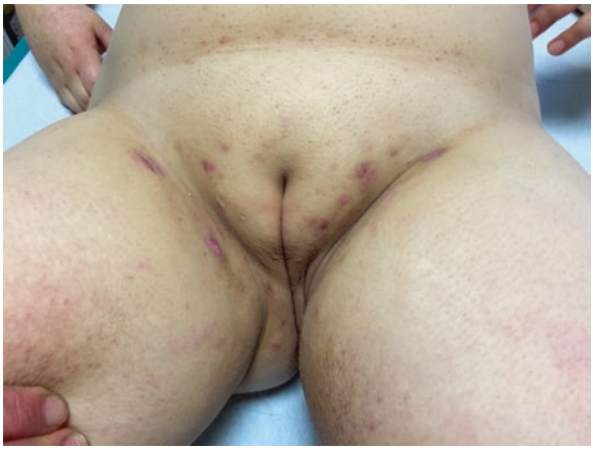

Fig. 12.2 Hidradenitis suppurativa (acne inversa). Blackheads, papules, nodules and scarring on the perigenital areas and groin

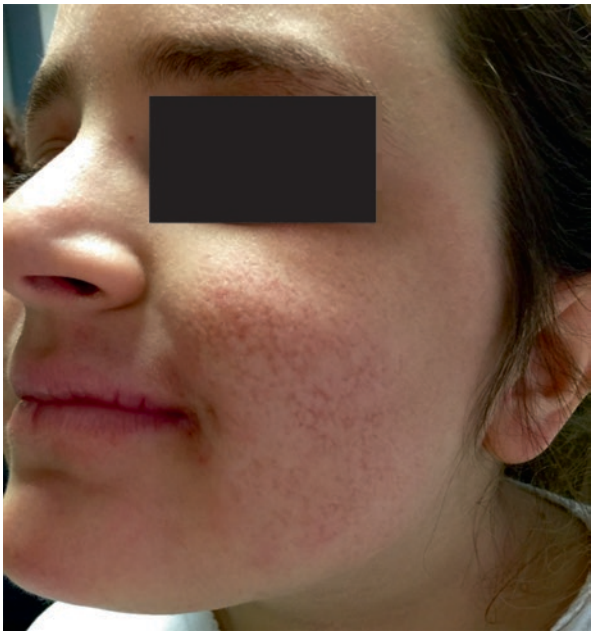

Fig. 12.3 Reticulated atrophy and punctate scars with dischromic appearance on the face

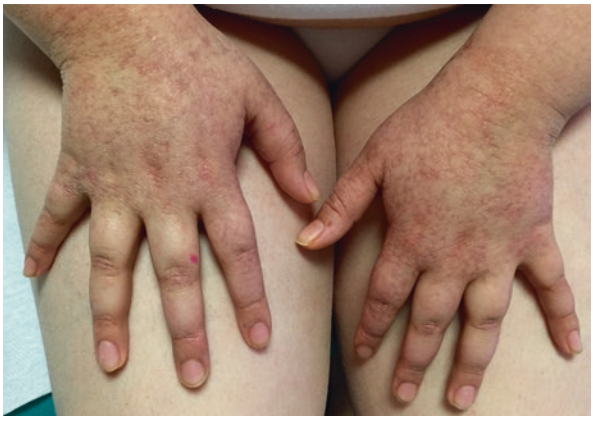

Fig. 12.4 Reticulated atrophy and punctate scars with dischromic appearance on the dorsal regions of the wrists and hands

Diagnosis

Rhotmund Thomson Syndrome (RTS).

Discussion

Rothmund-Thomson syndrome (RTS) is a rare autosomal recessive disorder characterized by: a facial rash, growth retardation, sparse or absent hair on the scalp,

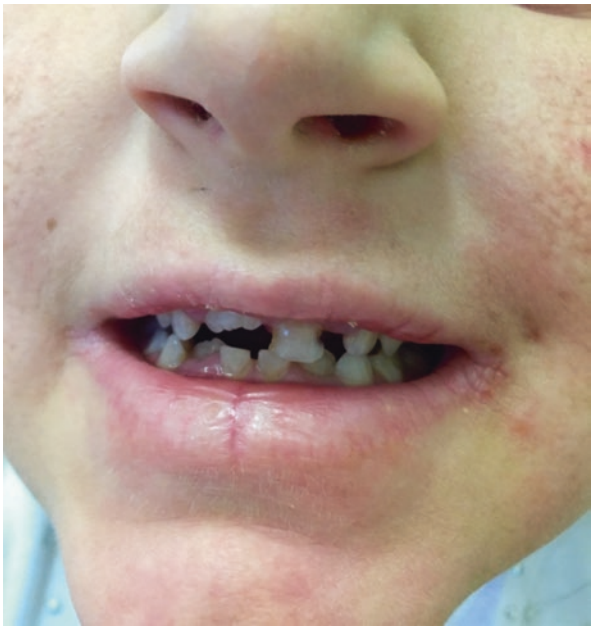

Fig. 12.5 Dental abnormalities

eyebrows and eyelashes, juvenile cataracts, skeletal abnormalities and a predisposition to early onset of cancer. The diagnostic hallmark is facial erythema, which appeares between three and six month, then extending to the extremities with the exception of the trunk. The evolution is towards punctate atrophy with reticular hypo- and hyper-pigmentation having the typical appearance of poikiloderma that persists throughout life [1].

There are two clinically defined forms of RTS: type I, characterized by poikiloderma, ectodermal dysplasia and juvenile cataracts with unknown etiology, and type II characterized by poikiloderma, congenital bone defects, an increased frequency of osteosarcoma and other skin cancers, caused by RECQL4 (8q24.3) mutation [1, 2]. The prevalence of RTS is unknown, about 300 cases have been reported in the literature. Type 2 RTS accounts for almost two-thirds of cases [1].

Patients may display few or many of the associated clinical features. The severity of each sign can also vary. Although some clinical signs suggest precocious aging, life expectancy is not impaired in patients who do not develop cancer [3].

The diagnosis is based on clinical findings (primarily at the age of onset, spreading and appearance of the poikiloderma) and molecular analysis for RECQL4 mutation.

Our patient had same of the clinical signs of RTS as poikiloderma, small stature, dental abnormality. Nevertheless her hair, eyelashes and eyebrows were normal, no skeletal abnormalities were clinically or radiologically detected. No cataracts and cancer have been found so far. She had atopic dermatitis and hidradenitis suppurativa, never priviously reported in association with RTS. The diagnosis of RTS was made according to typical lesions and mutation of RECQL4 gene. The patient was

advised to avoid sun exposure and undergo annual checkups for teeth, eyes, skin, and bones.

Poikiloderma occurs in a number of genodermatoses and other syndromes but with a subtly different pattern [3].

Lupus erythematosus. Poikiloderma has been reported as a feature of LE, but it does not appear early and is mainly expressed in the advanced phases of the disease.

Kindler syndrome is a hereditary and bullous syndrome, where poikiloderma arises generally at the age of two – three years. Blistering is induced by trauma and photosensitivity usually starts in early infancy. The patient commonly shows acral bullae, since birth or the first few days of life. The disease resembles epidermolysis bullosa. The blistering improves with age giving place to poikiloderma, mainly involving the face but also spreading to other sites. Additional features include syndactyly, dental abnormalities, stenosis of the oesophagus and anus, ectropion, chronic inflammation of oral mucosa and anhidrosis.

Dyskeratosis congenita is characterised by the tried: abnormal skin pigmentation, nail dystrophy and leukoplakia. The poikiloderma involves the face, neck, trunk and thighs. Its onset - like other clinical manifestations - occurs generally during childhood, though later than in RTS. The nail dystrophies are severe and are the first appearance, before the poikiloderma. Mental retardation is more common than in RTS. There is no photosensitivity.

Key Points

- The diagnosis of RTS is very probable when poikiloderma on the face, spreading to extremities, appeared in early infancy
- Poikiloderma is also a symptom of many systemic diseases, such as Lupus erythematosus, Kindler syndrome, dyskeratosis congenita and other
- Recognition of the age of onset and evolution of poikiloderma may be very useful to differentiate RTS from other syndromes
- The diagnosis of RTS should be made according to typical lesions and mutation of RECQL4 gene.
- The result of genetic testing is instructive and useful for a definitive diagnosis and future procreation guidance for the patient's family
- Genetic counselling should be provided for RTS type II patients and their families, together with a recommendation for cancer surveillance

References

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