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Case Report
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A New Isolated Genetic Syndrome with Ichthyosis: Congenital Ichthyosis, Photosensitivity, Poikiloderma, Frontal Bossing, and Hypodontia

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Abstract

Observation: The ichthyosis, a member of a heterogenous group of diseases characterized by abnormal cornification of the epidermis, is distinguished clinically by generalized scaling. Some variants of ichthyosis are characterized by additional manifestations such as hair shaft abnormality, hypogonadism, keratitis, deafness, skeletal changes, and mental retardation. We report a 14-year-old boy with congenital ichthyosis, photosensitivity, poikiloderma, frontal bossing, and hypodontia. On dermatological examination, there was a diffuse ichthyosiform scaling but the great flexures and face were spared as seen in ichthyosis vulgaris. Photosensitivity and reticulate pigmentation on the face were prominent. We excluded all the described syndromes associated with these findings. There was no positive family history. To our knowledge, our patient is the first report on this new genodermatosis which may be inherited in an autosomal recessive pattern.

Introduction

The ichthyoses are a heterogeneous group of skin disorders of epidermal differentiation, with both inherited and acquired forms. This cornification disorder may be found isolated or in association with other genetic defects [**1, 2**]. We report a 10-year-old Turkish patient with ichthyosis vulgaris, photosensitivity, poikiloderma, frontal bossing and hypodontia as a first report on this syndrome.

Case Report

A 10-year-old boy was admitted to our hospital because of photosensitivity, and a very dry skin. He was born at term after an uncomplicated pregnancy. Ichtyosis was present at birth, but there was no history of a collodion baby. In early childhood period, photosensitivity and poikiloderma had appeared on the face.

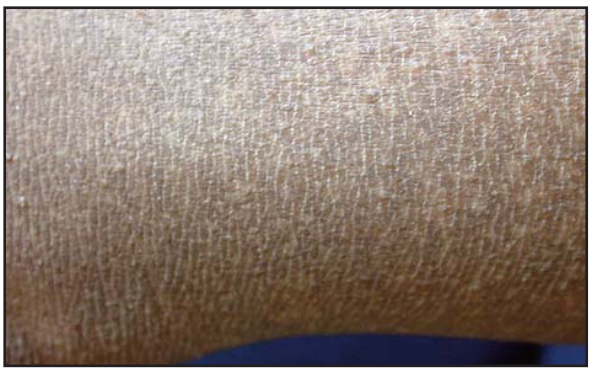

Figure 1. Ichthyosis on the forearm

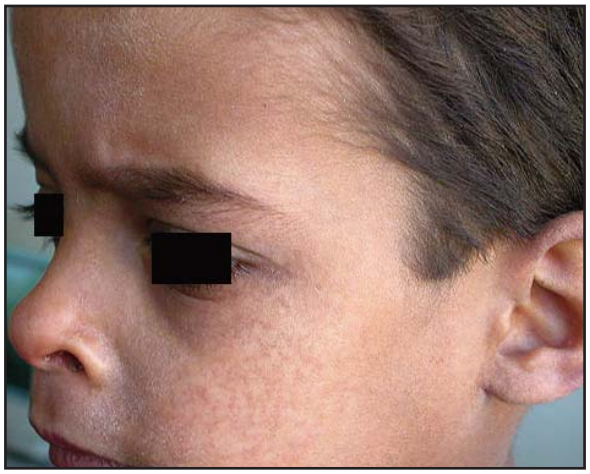

Figure 2. Marked telengiectasia, hipopigmentation and hiperpigmentation on the face and frontal bossing

On dermatological examination, there was a diffuse ichthyosiform scaling but the great flexures and face were uninvolved as seen in ichthyosis vulgaris (**Figure 1**). Sweating was evaluated clinically and pronounced hypohydrosis was detected. Marked telengiectasia, hipopigmentation and hiperpigmentation were observed on the face, which was consistent with poikiloderma (**Figure 2**). We observed tooth abnormalities such as tooth decay and raw tooth, especially involving upper and lower incisors. (**Figure 3**). Nail dystrophy, palmoplantar keratoderma, ectropion, eclabium, mucosa, hair abnormalities, and limb anomalies were not observed. He was mentally normal and of normal stature according to age. Hair examination was microscopically normal. There was no history of atopy. The patient was otherwise healthy and detailed ophthalmologic, urologic, neurologic and audiometric examinations were normal.

He had a negative family history of skin diseases. His parents were consanguineous, they were first cousins and their dermatological examinations were normal. There was no history of a drug intake of her mother along the pregnancy period. He had three unaffected sisters and four brothers.

In laboratory examination, the results of complete blood cell count, urinalysis, chemistry group, immunoglobulins including IgE and erythrocyte sedimentation rate were within normal limits. Serum levels of triiodo-thyronine, thyroxine, thyroid stimulating hormone were normal. No remarkable findings were noted on roentgenograms of chest, skull, mandible and long bones except for frontal bossing.

A biopsy specimen was obtained from the ichthyotic skin. The histopathological examination of the

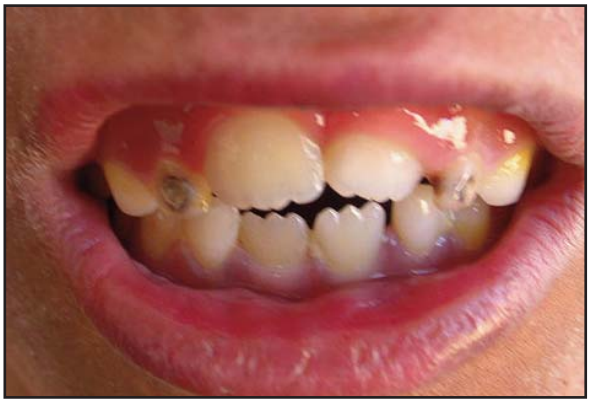

Figure 3. Teeth abnormalities

biopsy specimen revealed a moderate degree of laminated orthokeratosis. The granular layer was thin or absent in focal areas.

Based on clinical and laboratory findings a diagnosis of ichthyosis vulgaris associated with photosensitivity, poikiloderma, hypodontia and frontal bossing were made. The patient was prescribed 10% urea cream and salicylic acid ointments.

Discussion

Ichthyosis is a feature of several genetic disorders. These are rare disorders and the associated ichthyosis may be mild [**1**]. Syndromes associated with ichthyosis such as *Happle*'s syndrome (*Conradi-Hunermann* syndrome), *Sjögren-Larsson* syndrome, *Rud*'s syndrome, *Netherton*'s syndrome, Hystrix-like ichtyosis with deafness (HID) syndrome, PIBIDS (ichthyosis associated with photosensitivity, brittle hair, impaired intelligence, decreased fertility and short stature) syndrome, keratosis, ichthyosis, deafness (KID) syndrome, *Dorfmann* syndrome, ichthyosis follicularis with atrichia and photophobia (IFAP) syndrome, congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD) syndrome may be considered in the differential diagnosis of our patient.

Happle's syndrome is an X-linked dominant type of ichthyosis, characterized by severe erythrodermic ichthyosis at birth evolving into streaky hyperkeratosis in later life, patchy cicatricial alopecia, and cataracts. In older children generalized atrophoderma mainly involving the hair follicles and pigmentary disturbances is noted. Other features include stippled calcifications of the area of encondral bone formation, and asymmetric

shortening of legs [**3, 4**]. Major findings of this syndrome were absent in our patient.

Bazex-Dupre-Christol syndrome is characterized by follicular atrophoderma, multiple basal cell carcinomas, hypotrichosis, milia and localized hypohydrosis. Inheritance pattern of this syndrome is X-linked dominant [**5, 6**]. We excluded this syndrome because of the absence of characteristic findings such as milia, basal cell carcinomas and the pattern of inheritance. We ruled out *Comel-Netherton* syndrome as peculiar hair abnormality (trichorexis invaginata), generalized exfoliative erythroderma and atopy were not present [**7**]. The major features of the other syndromes including KID, CHILD, HID, *Sjögren-Larsson, Dorfmann*, IFAP syndrome were not noted in our patient [**8, 9**].

Trichothiodystrophy is a rare neuroectodermal disorder of autosomal recessive inheritance that is characterized by brittle hair, nail dysplasia, ichthyosis, mental retardation, and gonadal failure [**10**]. We ruled out this syndrome since there were not characteristic manifestations.

IBIDS is a syndrome characterized by ichthyosis, brittle hair, impaired intelligence, decreased fertility, and short stature, but unassociated with skeletal lesions [**11**]. These findings were not present in our patient.

Photosensitivity and poikiloderma may be found isolated or in association with other genetic defects. Syndromes such as *Rothmund-Thomson, Kindler, Smith-Lemli-Opitz* syndromes, porphyria, xeroderma pigmentosum, trichothiodystrophy, PIBIDS can be associated with photosensitivity and poikiloderma [**4**]. We also ruled out these syndromes.

Syndromes associated with hypodontia such as hereditary ectodermal dysplasia, *Rothmund-Thomson* syndrome, segmental odontomaxillary dysplasia, dyskeratosis congenita, *Kabuki* syndrome, *Papillon-Lefevre* syndrome, focal dermal hypoplasia (*Goltz*'s syndrome), *Hallerman-Streiff* syndrome, incontinentia pigmenti might be considered in the differential diagnosis [**9**]. However, major findings of these syndromes were absent in our patient. The presence of small, notched, peg-shaped incisors is also part of the late congenital syphilis triad. Syphilis serologic tests were negative in our patient.

Our patient also had frontal bossing. Frontal bossing can be seen in several genetic disorders such as *Hutchinson-Gilford* progeria, hypohidrotic ectodermal dysplasia, oral-facial-digital syndrome type I, cutis marmorata telangiectatica congenita, X-linked dominant *Conradi-Hunermann* syndrome, and neurofibromatosis [**9**]. Manifestations of these syndromes were absent in our case. We also observed hypohidrosis in our patient. Hyperkeratosis can interfere with normal sweat gland functions, resulting in hypohydrosis. Hypohydrosis is an expected consequence of ichtyosis, therefore we believe that it must not be considered as an additional characteristic finding of this new genodermatosis with ichtiyosis.

Considering our case, the inheritance may be in an autosomal resessive pattern but we could not exlude sporadic fashion since there was no sign of this syndrome in any of the patient's family members.

We conclude that this syndrome is a new genodermatosis with its characteristic features and this is the first report on this congenital ichthyosis, photosensitivity, poikiloderma and hypodontia syndrome. We believe that reports of new cases will help to clarify the clinical features and the certain inheritance pattern of this syndrome.

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