Focal Dermal Hypoplasia Associated with Ichthyosis

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We report a case of focal dermal hypoplasia associated with ichthyosis in an 18-year-old Korean female. The patient showed a yellowish atrophic patch on the left side of her abdomen, abnormal skin pigmentation, nail dystrophy and ichthyosis since birth. We could also find skeletal and dental deformities.

The biopsy specimen obtained from the yellowish atrophic patch showed marked attenuation of collagen fibers and ectopic fatty tissue nearly reaching up to the upper dermis.

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Key Words: Focal dermal hypoplasia, Ichthyosis.

Focal dermal hypoplasia (Goltz's syndrome) is a rare mesodermal and ectodermal origin defect which was first described by Goltz et al[1] in 1962. The syndrome is predominantly seen in females, a fact possibly explained by an X-linked dominant inheritance. Almost all the cases of focal dermal hypoplasia syndrome reported in the literature have been in caucasian patients but cases in oriental patients have rarely been reported[2].

This rare condition is characterized by widespread dysplasia of mesoectodermal origin, those are dermal hypoplasia with herniation of the adipose tissue, skeletal defects, abnormal skin pigmentation, dental, ocular and nail deformities[1-6].

We report a case of Goltz's syndrome associated with ichthyosis.

REPORT OF A CASE

An 18-year-old Korean high school girl was seen in our department of dermatology in December 1991. The patient was born in normal delivery at full-term. Her mother had no experience of spontaneous miscarriage. Neither her ancestors nor relatives had been known to have unusual skin or musculoskeletal disorders. On physical examination the patient had left 2nd, 3rd and right 3rd, and 4th finger syndactyly (Fig. 1). A Hand operation had been done before she visited our hospital. The Right hand syndactyly was surgically corrected but her lobster-claw deformity was not repaired. The Left thumb and index finger nail were dystrophic (Fig. 1). A lower incisor was absent and all teeth showed hypoplastic changes (Fig. 2). The anterior aspect of the trunk showed marked hyper & hypopigmentations and yellowish atrophic patches with telangiectasia on the left side (Fig. 3A). On the left side of the lower abdomen, the skin showed atrophy and telangiectasis (Fig. 3B).

The posterior trunk revealed atrophy of the skin with mottled hyper-and hypo-pigmentation (Fig. 4). The extensor surface skin of both lower extremities showed ichthyosiform patches which developed a few months after birth (Fig. 5).

The biopsy specimen taken from the yellowish atrophic patches on the left side of abdomen showed focal epidermal acanthosis and marked attenuation of collagen fibers and fatty tissue nearly reaching up to the upper dermis without overlying elastic fibers (Fig. 6). A Skin biopsy specimen

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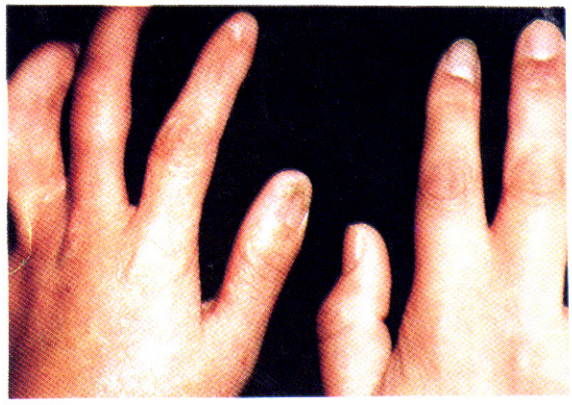

Fig. 1. The right 2nd and 3rd and the left 3rd and 4th fingers showed syndactyly and the left tumb and index finger nail showed deformities.

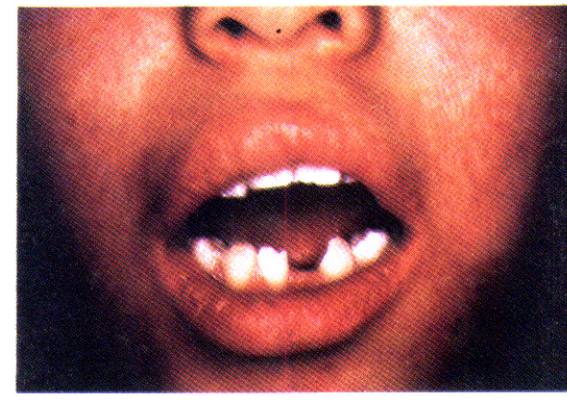

Fig. 2. A lower incisor was absent and all teeth showed hypoplastic changes.

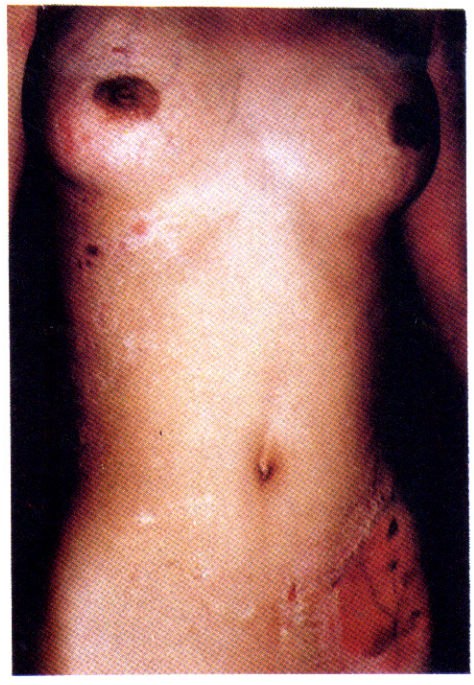

Fig. 3A. The anterior aspect of the trunk showed marked hyper & hypopigmentations and yellowish atrophic patches with telangiectasia on the left side.

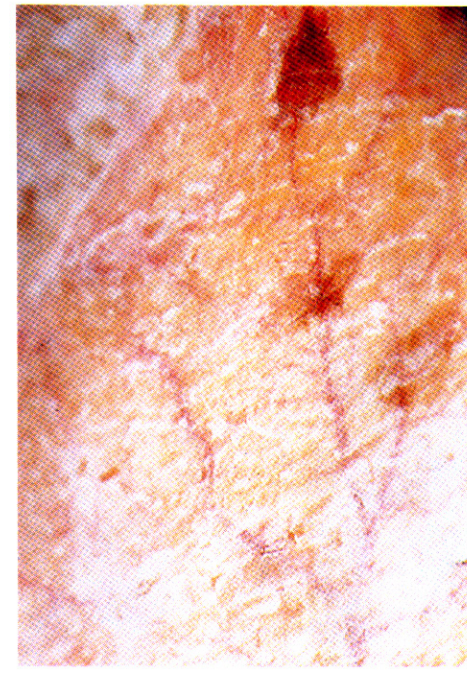

Fig. 3B. On the left side of the abdomen, the skin showed atrophic patches and telangiectasia.

from ichthyosiform patches of the right lower extremity showed hyperkeratosis without a granular layer (Fig. 7).

The laboratory examinations including CBC, X-ray of the chest, ECG as well as chromosomal study were normal or within normal limits. Her intellingence was normal.

DISCUSSION

Focal dermal hypoplasia syndrome is a congenital dysplasia derived from ectoderm and mesoderm. Attention was drawn by Goltz et al[1] and Gorlin et al[3] to a rare syndrome of multiple congenital defects affecting tissues of both mesodermal and ectodermal origin. Because of the dysplastic character of the syndrome Ishibashi and

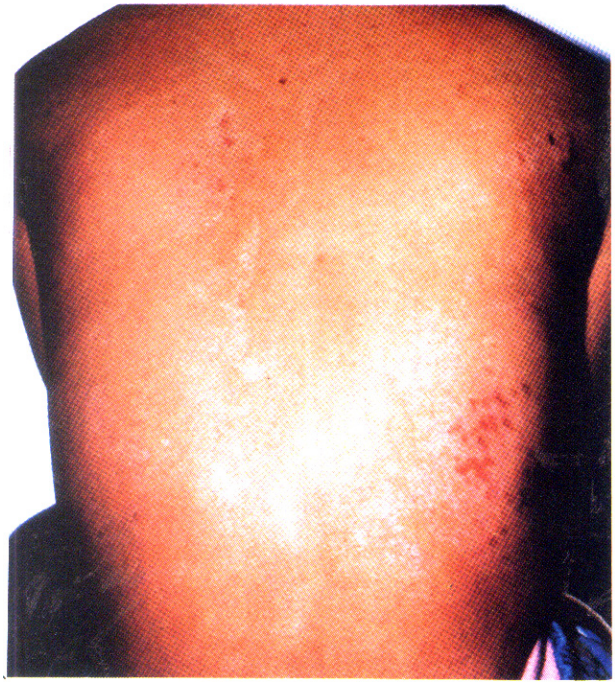

Fig. 4. The trunk revealed atrophy of the skin with hyperpigmentation and the same interspersed areas of hypopigmentation.

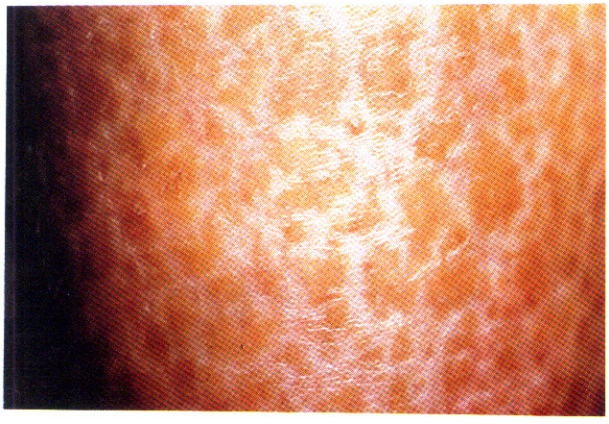

Fig. 5. The extensor surface skin of the lower extremities showed ichthyosiform patches.

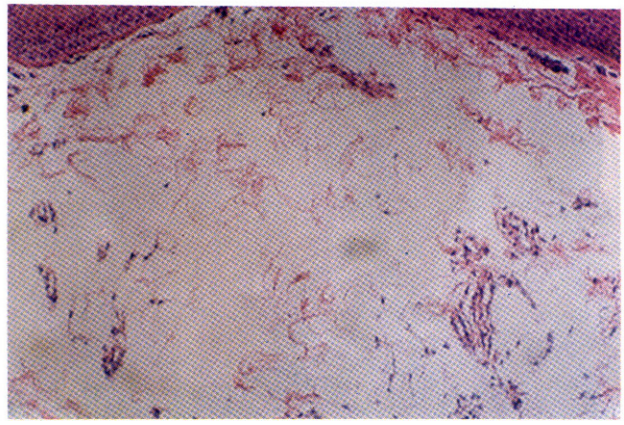

Fig. 6. A skin biopsy specimen from the yellowish atrophic patches on the left side of abdomen showed marked attenuation of collagen fibers and fatty tissue reaching up to the upper dermis without overlaying elastic fibers (H & E stain, ×200).

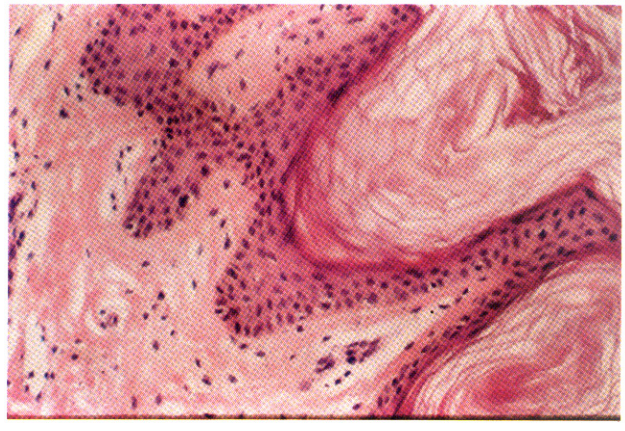

Fig. 7. A skin biopsy specimen from the ichthyosiform patches of the right lower extrimity showed hyperkeratosis without a granular layer (H & E stain, ×200).

Kurihara[10]prefer to name it dysplasia rather than hypoplasia. It is possible that this condition is inherited as an X-linked dominant trait, lethal in the male, and producing markedly reduced fertility in the female. Goltz et al[9] proposed that there is an autosomal dominant defect with sex limitation. So, it occurs primarily in females. To date, karyotype examination has provided no indication of damaged chromosomes[8]. There seemed to be a racial preference because most reported cases are in caucasians Hall and Terezhalmy[8] reviewed 125 cases of focal dermal hypoplasia and their distinct features included cutaneous abnormalities, skeletal defects, ocular abnormalities, oral and dental anomalies and soft tissue defects.

The skin changes are so striking that there is no confusion with other diseases. The main skin changes are linear or reticular areas of hypo-or hyper-pigmentation associated with dermal hypoplasia and herniations of the subcutaneous fat through this thinned dermis. So, the diagnosis is made primarily on the base of skin lesions and the characteristic histologic feature of super-

ficial fatty deposition in the skin. Next to skin manifestations skeletal defects are frequently seen.

The most common skeletal abnormalities are syndactyly, adactyly, clinodactyly, and hypoplasia of the digital limbs. Especially syndactyly between the second and third or third and fourth digits are relatively common findings. Our patient revealed left 3rd, 4th and right 2nd, 3rd syndactyly. Goltz et al[9] assumed that abnormal skeletal development occured by the 8th week of gestation, because the digits normally have elongated and seperated by that time. The factors of external environment in the literature correlated with this period[11].Developmental problems may affect the eruption and position of teeth, and enamel hypoplasia resulted in easy dental caries. Dystrophic nails are almost a constant feature. Our patient showed also left thumb and index finger nail deformities. Ocular lesions, especially colobomas[11], are frequently seen but our patient manifested normal findings.

For clinical or histologic diagnosis of the skin lesions in Goltz's syndrome we should consider congenital poikiloderma, connective tissue nevi, anhidrotic and hidrotic ectodermal dysplasia, incontinentia pigmenti, nevus lipomatosus superficialis and skin tag. But these were easily differentiated from focal dermal hypoplasia not only by its peculiar clinical manifestations but also distinctive histopathologic findings.

The relationship between the FDH and the ichthyosis has not been clearly elucidated but we could assume that ichthyosis resulred from an ectodermal defect the same as FDH, and there was indeed a case of FDH accompanying ichthyosis reported by Baden and Rex[7] in 1970.

We report a case of Goltz's syndrome accompanying ichthyosis showing typical clinicohistopathologic findings.

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