Two Cases of Goltz Syndrome

Sung Whan Cho, M.D., Wook Lew, M.D., Eung Ho Choi*, M.D.,
Seung Hun Lee*, M.D., Sung Nack Lee, M.D., Sang Heon Lee,** M.D.

Department of Dermatology, Yonsei University College of Medicine
Seoul, Korea

Departments of Dermatology and Plastic Surgery**, Yonsei University Wonju*
College of Medicine. Wonju, Korea

Goltz syndrome, also known as focal dermal hypoplasia syndrome, is a rare congenital mesoectodermal disorder. Two cases, which we experienced, showed erythematous, tan skin rashes and atrophic scars on the whole body, but there were some differences in clinical manifestations. Histopathologic findings in both cases showed diminution in the thickness of the dermis with subcutaneous fat extending upward to the epidermis. Therefore, we report a comparison of two cases of Goltz syndrome, especially with respect to clinical manifestations.

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Key Words: Focal dermal hypoplasia, Goltz syndrome

Goltz syndrome, also known as focal dermal hypoplasia syndrome, is a rare congenital mesoectodermal disorder which was first described in 1962[1]. This syndrome, which is characterized by widespread dysplasia of mesoectodermal structures including underdevelopment and maldevelopment of the dermis, is probably due to an X-linked dominant trait. The clinical manifestations of this syndrome include cutaneous defects (consisting of hypoplasia of the skin resembling striae distensae, herniations of adipose tissue in the form of yellowish papules, and abnormal skin pigmentation), and skeletal, dental, ocular, hair and nail deformities[2-6].

REPORT OF CASES

Case 1

A 16-year-old girl visited Dermatologic Department of Wonju Christian Hospital with erythematous, tan skin rashes over her whole body and syndactyly of the right foot since birth (Fig. 1). Asymmetric development of the face and extremities was also observed. She had reddish, tan scars, resembling striae distensae and atrophic scars associated with hypo-and hyperpigmentation over her entire body (Fig. 2), a walnut-sized alopecic patch on the occipital area, a pea-sized erythematous papule on the right margin of the lip, deformities of the finger and toe nails and hypoplasia of the teeth (Fig. 3.). No other persons in her family showed similar manifestation. There were no known family history of consanguinity. Past history were non-contributory.

Radiologic abnormalities included microcrania, hypoplasia of the left 12th rib (Fig. 4), kyphoscoliotic deformities, widening of the symphysis pubis and symphalangism of the right 3rd and 4th digits. Intelligence test using KWIS (Korean Wechsler Intelligence Scale) and chromosome studies were normal. There were no ocular lesions.

Microscopic examination of one of atrophic scars showed focal dermal hypoplasia characterized by a marked diminution in the thickness of the

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Reprint request to: Seung Hun Lee, M.D. Department of Dermatology, Yonsei University Wonju College of Medicine Wonju, Korea

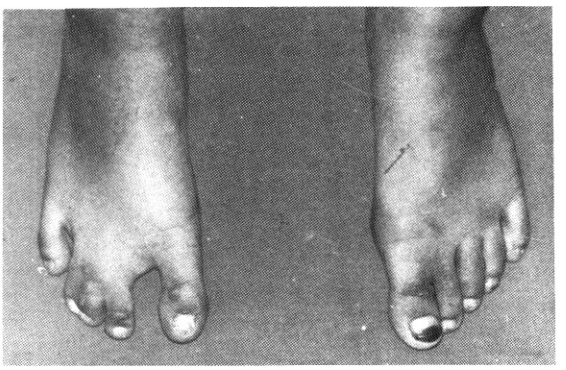

Fig. 1. Syndactyly on the right foot

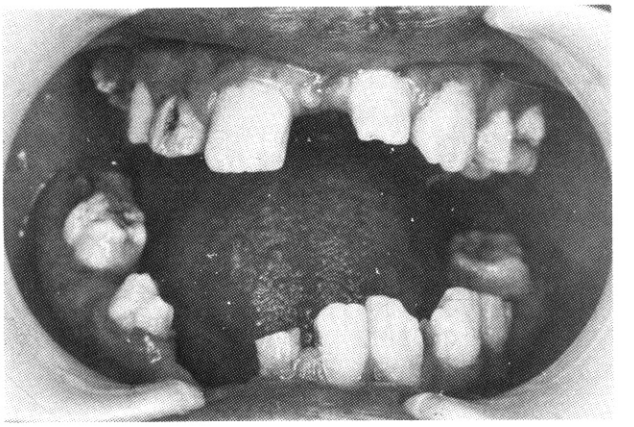

Fig. 3. Dental dysplasia of the teeth

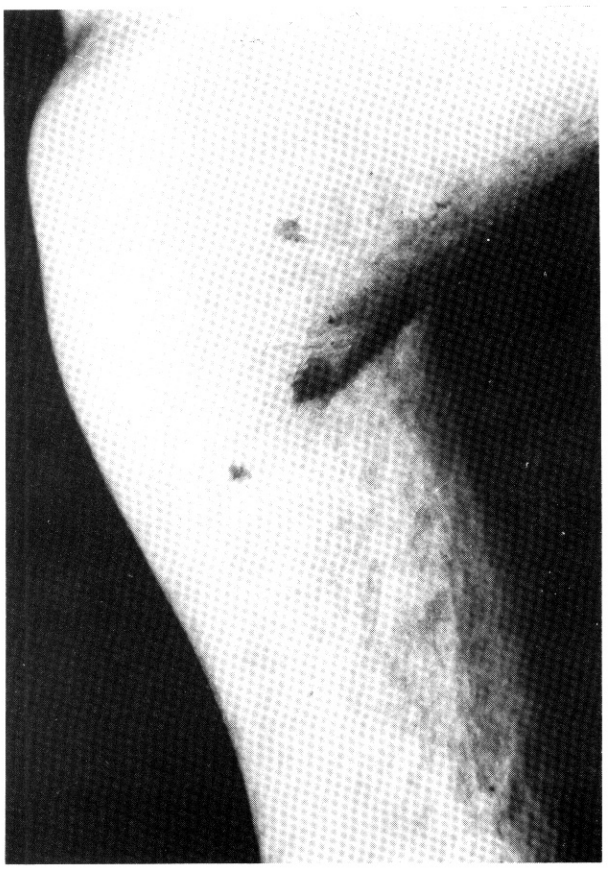

Fig. 2. Reddish-tan scars, resembling striae distensae and hypo-and hyperpigmented atrophic scars of the right arm

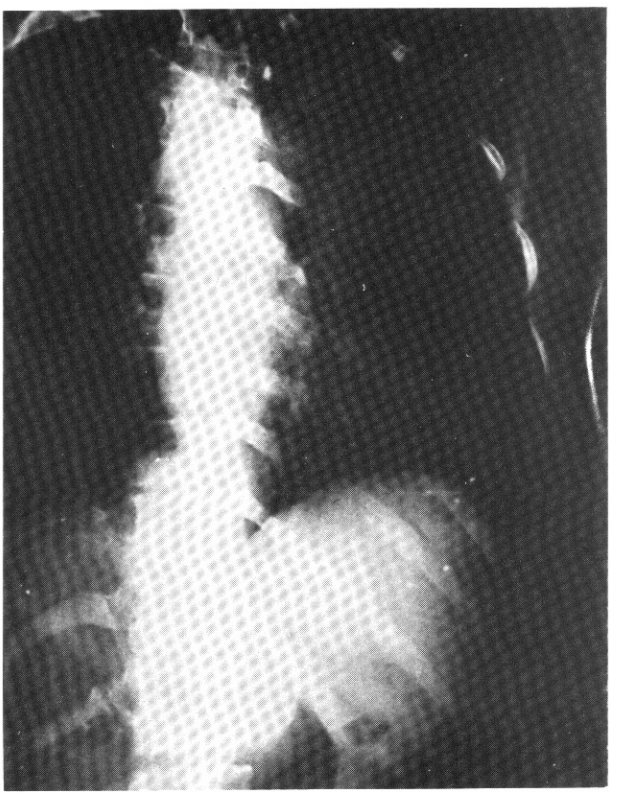

Fig. 4. Hypoplasia of the left 12th rib on the rib cage view

dermis, thinning of the collagen fibers and extension of the subcutaneous fat upward to the epidermis. To correct the syndactyly of the right foot, she underwent reconstructive surgery in the Department of Plastic Surgery.

Case 2

A 23-year-old female visited Dermatologic Department of Severance Hospital because of scar-like rashes over her whole body which had been present since childhood. Physical examination revealed reddish tan, cribriform, atrophic scars on the extremities and trunk, hypopigmented

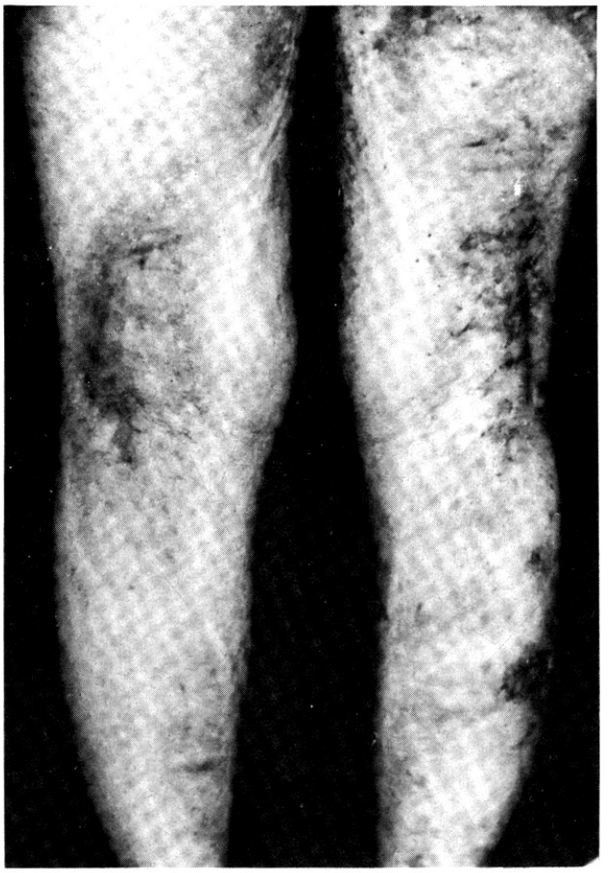

Fig. 5. Reddish-tan, cribriform, hypo-and hyperpigmented atrophic scars, and soft yellowish nodules

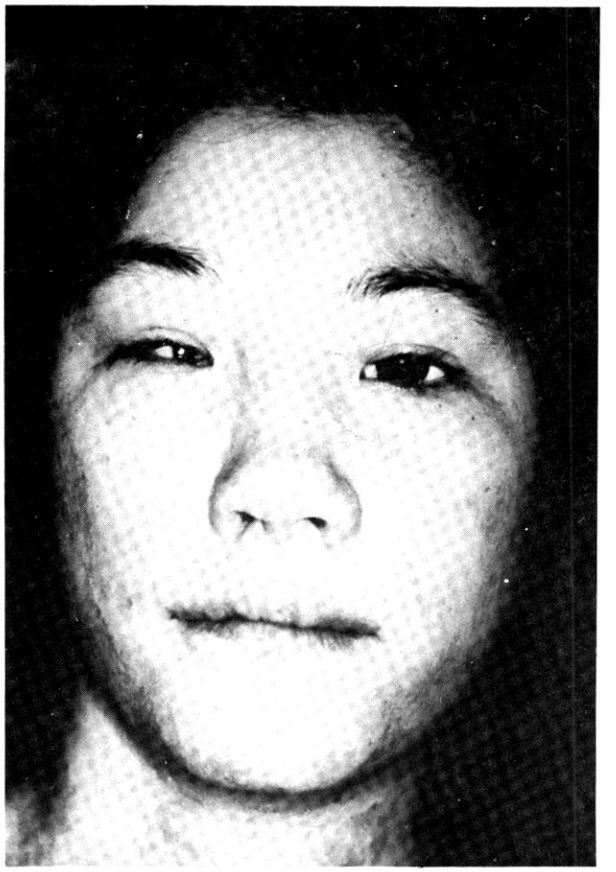

Fig. 6. Underdevelopment of the right side of the face ( ), papilloma on the right inner canthus, and strabismus

atrophic scars on both popliteal fossae, and soft protruding hyperpigmented nodules on the posterior thighs (Fig. 5). Other positive findings included underdevelopment of the right side of the face, bilateral blindness which had been present since high school, a papilloma on the right inner canthus, and strabismus of the right eye (Fig. 6). Three teeth were absent without any history of dental extraction or trauma, but no gross abnormality was noticed. Extremity examination revealed syndactyly (incomplete simple type), a shortened third phalanx and keratotic lesions of the left palm (Fig. 7). She had a past history of burn in her childhood, but no other persons in her family revealed similar manifestations.

Laboratory studies including serologic tests for syphilis were within normal limits or negative. Chest radiograph and abdominal ultrasound were normal. Neurologic studies and intelligence test were normal. Karyotype analysis revealed no chromosomal abnormalities. The skeletal radiograph series showed a shortened middle phalanx of the left third finger, osteoporosis of the pelvic bones, and tall vertebral bodies with osteoporotic changes. A deviated dorsum sella without asymmetrical bony underdevelopment was noted on skull radiographs. A skin biopsy of one of the posterior thigh lesions showed a marked diminution in the thickness of the dermis, thinning of the collagen fibers and extension of the subcutaneous fat upward to the epidermis, compatible with focal dermal hypoplasia (Fig. 8).

DISCUSSION

Goltz syndrome is an X-linked, dominantly inherited disorder with a pronounced lethality in

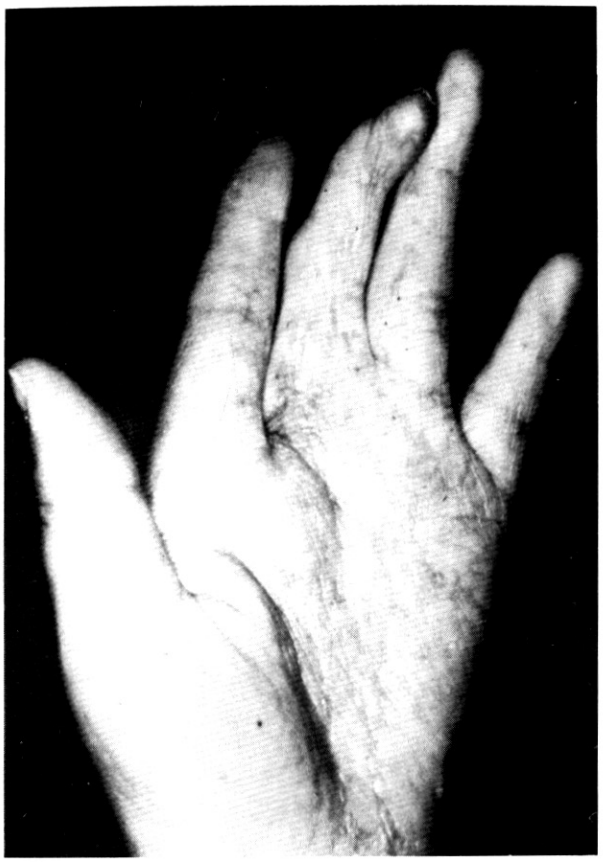

Fig. 7. Syndactyly (incomplete simple type), short 3rd phalange and keratotic lesions of the left hand

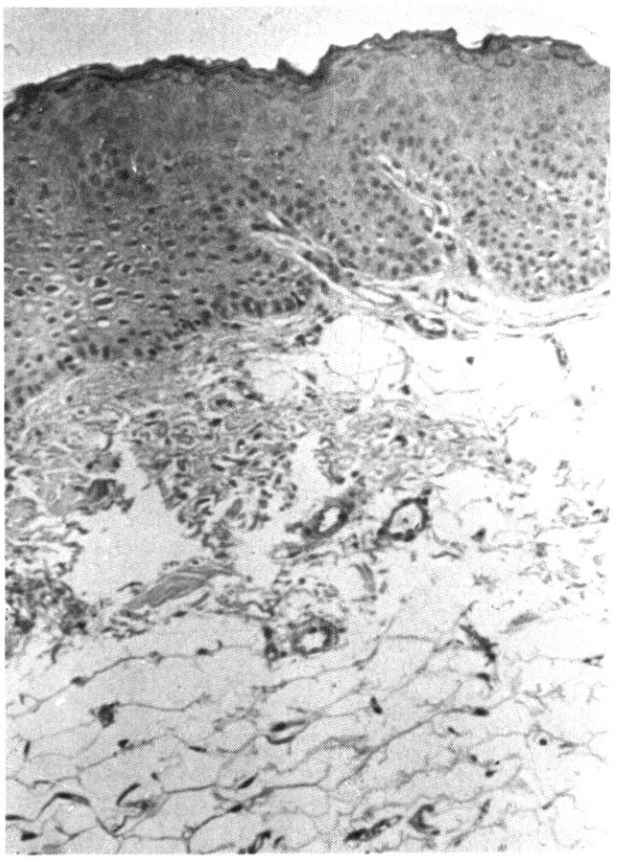

Fig. 8. Marked diminution in the thickness of the dermis and extended subcutaneous fat upward to the epidermis (H & E stain, ×40)

males[7-10]. Therefore, the syndrome occurs largely in females with only 12% of the reported cases in living males[4]. Some heredofamilial tendency has been reported[2], but the etiology and mode of transmission are unknown yet. Its occasional occurrence in males may be the result of a new mutation[11].

The clinical manifestations of the syndrome which have been previously described, include small stature, skeletal abnormalities, asymmetry of the face, trunk and extremities, mental deficiency, epileptiform seizures, and ocular, dental and cutaneous defects[1, 2, 12, 13]. In both our cases, the patients were young females with no family history. A comparison of clinical findings in the two cases is summarized in Table 1, 2, 3. The unique findings of case 1 were as follows: microcrania, kyphoscoliosis, symphalangism of the right 3rd and 4th digits, widening of the symphysis pubis, hypoplasia of the left 12th rib (Fig. 4) and nail deformities. In case 2, keratotic area of the left palm and short phalange of the left 3rd finger (Fig. 7), strabismus, blindness, osteoporotic changes in the vertebrae and extremities, and increased height of the vertebral bodies could be considered the unique findings. The protruding yellowish nodules (Fig. 5) due to herniated fat, hyper-or hypopigmented atrophic scars, papillomas of the lip or periorbital area (Fig. 6), telangiectasias, and alopecia of the scalp, pubis and axilla are cutaneous findings shared by both cases. Other shared findings are as follows: short stature, asymmetric development of the face and extremities (Fig. 1, 6, 7), syndactyly (Fig. 1, 7), normal intelligence, dysplasia (Fig. 3) or hypoplasia of teeth, normal karyotype.

The clinical or histopathologic differential diagnosis of the skin lesions in Goltz syndrome

Table 1. Comparison of cutaneous abnormalities of the two cases.

Items*	Case 1	Case 2
Area of underdevelopment and thickness of skin-reticular, vermiform, cribriform, frequently linear	Y	Y
Localized herniation of subcutaneous fat through the attenuated dermis	Y	Y
Linear or reticular areas of hyper- or hypopigmentation of the skin	Y	Y
Telangiectasia	Y	Y
Papilloma of the lip or periorbital skin	Y	Y
Keratotic lesions on palms and soles	N	Y
Hair lacking from small areas of scalp, pubis or axilla	Y	Y
Finger and toe nails absent, poorly developed, dystrophic, spooned, grooved, or hypopigmented	Y	N

*Modified from Ref. 2), Y: yes, N: no

Table 2. Comparison of skeletal defects of the two cases.

Items*	Case 1	Case 2
Microcrania	Y	N
Spinal anomalies		
Kyphoscoliosis	Y	N
Anomalies of vertebrae	N	Y
Anomalies of hands and feet		
Hypoplasia or absence of digits	N	Y
Syndactyly	Y	Y
Fusion of phalanges	Y	N
Rib anomalies	Y	N
Pelvic abnormalities	Y	N
Generalized osteoporosis	N	Y

*Modified from Ref. 2), Y: yes, N: no

Table 3. Comparison of other anomalies of the two cases.

Items*	Case 1	Case 2
Asymmetry of extremities and face, notching or underdevelopment of ala nasi	Y	Y
Short stature	Y	Y
Mental deficiency	N	N
Strabismus	N	Y
Blindness	N	Y
Dysplasia or hypoplasia of teeth	Y	Y
Chromosomal abnormalities	N	N

*Modified from Ref. 2), Y: yes, N: no

includes congenital poikiloderma, connective tissue nevi, anhidrotic and hidrotic ectodermal dysplasia, incontinentia pigmenti, nevus lipomatosus superficialis and skin tag.

In conclusion, we report two rare cases of Goltz syndrome showing typical dermatopathologic findings and clinical features.

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