Pityriasis Rubra Pilaris in Down Syndrome

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A 23-year-old woman with Down syndrome presented erythematous keratotic plaques with whitish scales on the elbows, forearm, knee and leg. Histopathological findings revealed acanthosis with broad and short rete ridges, alternating orthokeratosis and parakeratosis oriented in both vertical and horizontal directions, and dermal superficial perivascular lymphocytic infiltration, compatible with pityriasis rubra pilaris.
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Key Words : Down syndrome, Pityriasis rubra pilaris

Down syndrome is the most common abnormality of chromosomal number, featuring mental deficiency, protruding tongue, upward-slanting palpebral fissures, epicanthal folds, short, broad neck, and simian crease[1].

A large number of abnormal skin features have been reported in Down syndrome[2]. They include protruding and/or fissured lower lip, enlarged tongue, acrocyanosis, increased number of creases on palms and soles, xerosis, ichthyosis and keratosis palmaris et plantaris[3,4].

Rarely, pityriasis rubra pilaris (PRP) was reported in association with Down syndrome as first by Hazini et al.[5] and subsequently by Holden and Curley[6]. Herein we report a Korean case of pityriasis rubra pilaris in Down syndrome.

CASE REPORT

A 23-year-old woman visited our clinic with erythematous keratotic plaques with whitish scales on the elbow, forearm, knee, and leg. The bean-sized erythematous keratotic skin lesions had been noted on both knees 10 years ago. The skin lesions had been spreading progressively to other regions such as both elbows, right-side arm, dorsal hand, leg, and foot. Systemic disorders such as diabetes mellitus, heart, lung, or kidney disease were not recognized. Her family history was not contributory.

On physical examination, the typical Mongoloid features of Down syndrome such as short stature, epicanthal fold, dryness of the lips, enlarged tongue, short phalanges, and simian lines of the palms were present (Fig. 1). She had well-demarcated erythematous follicular plaques with scales on the both elbows and knees, right-side arm, dorsal hand, leg and foot (Fig. 2). The routine chromosomal analysis revealed trisomy 21 (Fig. 3).

Histopathological examination of the follicular plaques on the knees showed acanthosis with broad and short rete ridges, slight spongiosis, thick suprapapillary plates, focal hypergranulosis, alternating orthokeratosis and parakeratosis oriented in either vertical or horizontal directions. In the dermis, there is a mild superficial perivascular lymphocytic infiltrate and moderately dilated blood vessels without any connective tissue changes (Fig. 4). A diagnosis of pityriasis rubra pilaris was made upon clinical and histopathological findings.

The patient was treated with topical 0.25 % prednicarbate and oral etretinate for 6 months, and the lesions were much improved.

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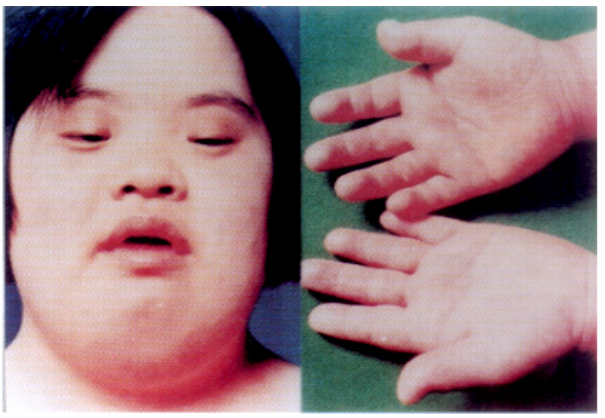

Fig. 1. Typical features of Down syndrome including epicanthal folds, short phalanges, and simian lines of the palms were shown.

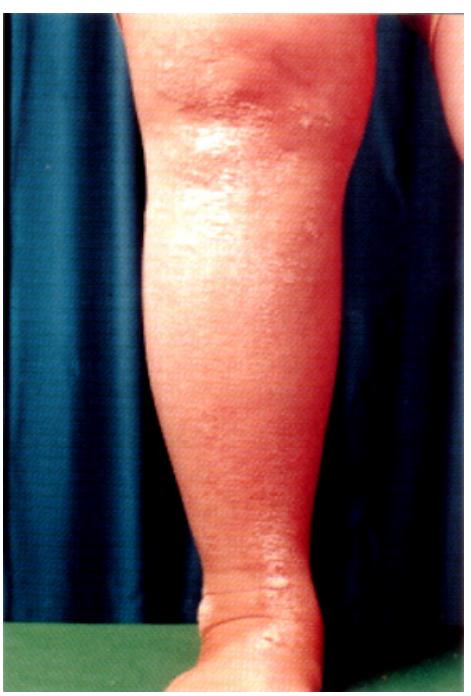

Fig. 2. Erythematous follicular plaques with scales on the right knee and ankle.

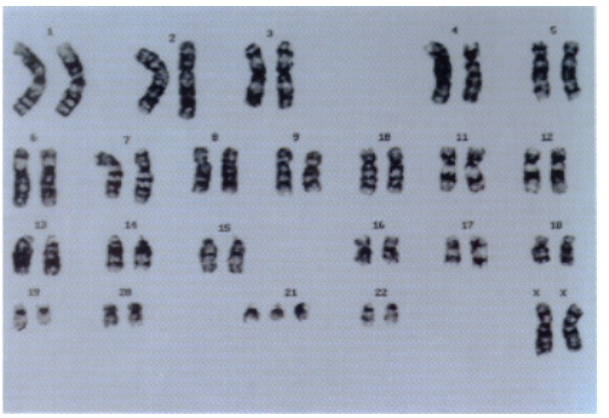

Fig. 3. The routine chromosomal analysis revealed trisomy 21.

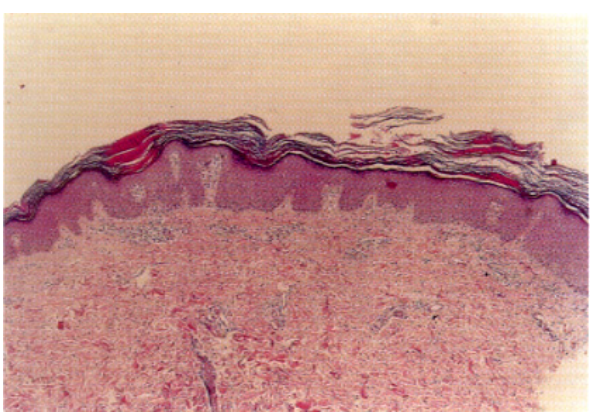

Fig. 4. Histopathological examination presented acanthosis with broad and short rete ridges, slight spongiosis, thick suprapapillary plates, focal hypergranulosis with alternating orthokeratosis and parakeratosis oriented in both vertical and horizontal directions. (H & E stain, × 100).

DISCUSSION

Down syndrome is a multisystem disorder that occurs approximately once in 600 to 700 live births[7]. Most commonly it is a consequence of faulty distribution of chromosomes resulting in an extra chromosome 21 (trisomy 21). Characteristic clinical findings include hypotonia, flat face, upward and slanted palpebral fissures, epicanthic folds, speckled irises, varying degrees of mental and growth retardation, cardiac malformation, simian crease, short, broad hands, and hypoplasia of middle phalanx of 5th finger[8].

Various cutaneous changes have been reported in Down syndrome[9]. These include xerosis or ichthyosis[3,4,10], atopic dermatitis[10-12], seborrheic dermatitis[10-12], folliculitis[13], follicular dermatosis[2,10], lichen simplex chronicus[4], elastosis perforans serpiginosa[10,11,13-16], keratoderma palmaris et plantaris[3,10], actinic sequellae-lentigines and atrophy[3],

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Table 1. Cutaneous manifestations of Down syndrome

Features	
Keratinization abnormalities	Others
Xerosis or ichthyosis	Atopic dermatitits
Keratoderma and Keratosis pilaris	Seborrheic dermatitis
Keratoderma palmaris et plantaris	Lichen simplex chronicus
Generalized connective tissue nevi	Cheilitis
Psoriasis	Elastosis perforans serpiginosa
Pityriasis rubra pilaris	Milialike idiopathic calcinosis cutis
	Reactive perforative collagenosis
	Actinic sequellae
	(lentigines and atrophy)
	Dermatophytes infection
	Norwegian scabies
	Folliculitis
	Alopecia areata
	Syringoma
	Vitiligo
	Acrocyanosis
	Cutis marmorata
	Acral lentiginous melanoma

dermatophytes infection[10-12], cutis marmorata[10-12], alopecia areata[2,10-12], syringoma[9-11,17], vitiligo[10,11], milialike idiopathic calcinosis cutis[9,18], reactive perforative collagenosis[19], generalized connective tissue nevi[20], acral lentiginous melanoma[21], cheilitis[10], keratoderma and keratosis pilaris[5,10]. (Table 1)

PRP in association with Down syndrome was first reported by Hazini et al.[5] in 1988. They described a 30-year-old woman with PRP and vitiligo in Down syndrome where suggested that the development of pityriasis rubra pilaris in Down syndrome was likely to be related with high incidence of keratinizing disorders, such as hyperkeratosis, xerosis, keratoderma, and keratosis pilaris. The second case was reported by Holden and Curley as well[6]. Previous reports and the present case share some features of similarity as follows : All affect young women and the skin lesions are erythematous plaques with scales on the elbows and knees, compatible with pityriasis rubra pilaris.

The relationship between Down syndrome and keratinization disorders is not clarified. Ercis et al.[22] suggested that vitamin A level seemed to be rather insignificant in the etiology of the keratinization disorders in Down syndrome than the genetic defect in the utilization of vitamin A at cellular level.

We report a 23-year-old woman with pityriasis rubra pilaris in Down syndrome, which is the first case in Korea. The pathogenesis of PRP in Down syndrome is still unknown. The pathogenic relationship between keratinizing disorders in Down syndrome and the role of vitamin A should be further evaluated in a number of cases.

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