Chapter 18
Disorders of the Dermis and Subcutaneous Fat

The dermis and subcutaneous tissue hold and support the epidermis. If the tissue is injured, the entire structure of the skin may be greatly affected, even though the surface of the skin itself might show only minor changes. This chapter discusses diseases that predominantly affect the dermis and subcutaneous tissue.

Disorders of the dermis

A. Cutaneous atrophy

1. Striae

Synonyms: Striae distensae, Striae atrophicae

Outline
- Slightly concave, linear cutaneous atrophy follows the lines of cleavage (skin tension lines).
- The thighs and lower abdomen are most commonly affected.
- Oral steroids may be the inductive factor. Striae occur when skin undergoes rapid growth or stretching, especially in pregnancy (striae gravidarum) and adolescence.

Clinical features

Striae are slightly concave, several millimeters wide, and 10 cm or more long. They run roughly parallel each other. The color is rose pink in the early stages, becoming grayish white later on (Fig. 18.1). The long axis of the striae follows the lines of cleavage (skin tension lines) in most cases. The buttocks, lumbar region, thighs and popliteal fossae are often involved. Striae gravidarum is seen in more than 90% of all pregnancies. It occurs on the abdomen, breasts and groin from the 6 month of pregnancy to after delivery. Adolescent striae occur in the buttocks, lateral thighs, back and breasts.

Pathogenesis

Glucocorticoid inhibits fibroblast activity (collagen production), leading to reduction of connective tissue. Wound healing is impaired. If skin in this condition is subjected to external pressure or excessive extension, the connective tissue is destroyed, resulting in striae and atrophy in the skin. Striae tend to occur when glucocorticoid levels are elevated, such as in conditions that require oral administration of steroids, Cushing syndrome, severe infection and diabetes. It may occur in those who are preg-
nant or obese, or those in adolescence.

**Pathology**

Striae (distensae) are basically scars.

**Treatment, Prognosis**

No specific treatment is necessary. The disorder subsides with age; nonetheless, it is irreversible and does not disappear completely.

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**2. Solar elastosis**

**Outline**

- It is dermal degeneration caused by excessive exposure to sunlight.
- This is an aging change and atrophy of the skin.
- Cutis rhomboidalis nuchae, a specific subtype of this condition, is deep rhombus striae that occur in the nuchal region.

**Clinical features**

The onset of solar elastosis is in the fourth decade of life. The skin becomes thin and yellowish, and degeneration of dermal elastic fibers is observed histologically. The skin slackens on the whole body. Large folds of skin form on the face, neck and joints. Because of the functional reduction of sweat glands and seborrheic glands, the whole body skin becomes dry and rough, leading to scaling, a characteristically atopic gloss, and brownish color.

Solar elastosis is remarkable on sun-exposed areas. Outdoor workers show marked changes caused by solar elastosis. Deep cleavages are seen, particularly in the nuchal region (cutis rhomboidalis nuchae).

**Pathology**

Atrophy and thinning of the dermis occur. Reduction of collagen fibers is marked (Fig. 18.2). The elastic fibers are ruptured, and solar elastosis is observed by Elastica-Van Gieson method. The sweat glands and seborrheic glands decrease in size and number, and subcutaneous fat tissue decreases.

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**3. White fibrous papulosis of the neck** *(Shimizu)*

**Clinical features**

Small round or oval papules 2 mm to 4 mm in diameter and white to light yellow occur on the neck region of the elderly (Fig. 18.3). Sharply circumscribed eruptions occur on follicular and
non-follicular sites. They do not coalesce. Thickening of the collagen fibers is histopathologically observed in the upper dermal layer (Fig. 18.4). The pathogenesis is age-related dermal degeneration.

**Epidemiology**

White fibrous papulosis of the neck may occur in any race.

### 4. Lichen sclerosus et atrophicus (LSA)

**Clinical features**

White, flat-topped papules of 2 mm to 3 mm in diameter appear and aggregate, forming firm white plaques. Later, the plaques shrink and take on a crepe-like appearance (Fig. 18.5). Inflammation may occur, accompanied by itching and pain. Lichen sclerosus et atrophicus (LSA) is a chronic disorder with a predilection for the anogenital lesion and trunk of middle-aged and elderly women. In postmenopausal women, when the vulva is involved the condition may be accompanied by atrophy in the labia majora and clitoris (kraurosis vulvae). LSA in the male genitalia is called kraurosis penis or balanitis xerotica obliterans. Atrophy may result in urethral stricture in male patients.

**Pathogenesis**

The pathogenesis is unknown; however, hereditary factors, endocrine abnormality or immunological mechanisms may be involved. Autoantibodies against extracellular matrix 1 (ECM1) have been found in the patient’s serum.

**Pathology**

Hyperkeratosis epidermal atrophy and vacuolar degeneration are present. In the dermal upper layer, collagen fibers are homogeneous and edematous, leading to reduction of cellular components. As it progresses, band-like lymphatic infiltration is seen in the dermis. There is keratin proliferation and the formation of follicular keratin plugs in some cases (Fig. 18.6).

**Prognosis**

LSA progresses slowly and tends to be intractable; however, it may resolve spontaneously. When the genitalia are affected, it progresses to squamous cell carcinoma in several percent of all cases, after a long course. Topical steroids and tacrolimus ointment are applied.
18 Disorders of the Dermis and Subcutaneous Fat

Synonym: Adult progeria

It typifies diseases of premature aging. Aging occurs in systemic tissue at adolescence.

It is caused by mutation in RecQ DNA helicase gene, RECQL2. It is autosomal recessively inherited.

Clinical features

Premature aging begins in systemic organs around adolescence. Subcutaneous fat and muscle markedly atrophy and adhere to the subcutaneous layer. Scleroderma-like articular contracture and atrophic hardening of the skin occur. The nose becomes thin and pointy from atrophy, giving the face a bird-like appearance. Keratinization and ulceration on the soles, pigmentation on the whole body, telangiectasia, and subcutaneous calcinosis occur. Gray hair and alopecia often accompany these symptoms (Fig. 18.7). In organs other than the skin, osteoporosis, arterial sclerosis, cataracts, insulin-resistant diabetes and gonadal hypofunction are caused by premature aging. High-pitched voice and loss of hircus and pubes are present.

Pathogenesis

Werner’s syndrome is caused by mutation in RECQL2 encoding the DNA helicase on chromosome 8. The product of the RECQL2 gene is thought to repair genes that are damaged during DNA replication. The mechanism of premature aging is unknown; nonetheless, it is thought that chromosomal instability is increased by incapacitation of the repairing gene, resulting in the onset of Werner’s syndrome.

Differential diagnosis

Differential diagnosis from other premature aging syndromes (progeria and acrogeria), scleroderma, and Rothmund-Thomson syndrome must be made.

Prognosis

Most patients are short-lived, with an average age of 46 years, as a result of myocardial infarction, cerebral apoplexy and aggravated diabetes. The incidence of malignancy is high.

6. Rothmund-Thomson syndrome

It is autosomal recessive. The cause is genetic mutation. One of the causative genes is RECQL4 encoding the DNA helicase, on chromosome 8. In infancy and childhood, the skin atrophies, reticular or diffuse erythema occurs on the face, and juvenile cataract appears (Figs. 18.8-1 and 18.8-2). Photosensitivity is present in one third of cases. In adulthood, head and body hair...
becomes sparse, and keratinization occurs on sun-exposed areas. There is impaired development of nails. Internal malignant tumor accompanies roughly 30% of cases; tibial osteosarcoma and multicentric osteosarcoma have been reported. The prognosis is good in the absence of malignancy. Like Werner’s syndrome, Rothmund-Thomson syndrome may be categorized as a type of progeria.

### 7. Progeria

**Synonym: Hutchinson-Gilford syndrome**

This is a premature aging syndrome. Abnormality in the lamin A gene has been reported. The main symptoms are growth impairment, evidenced by short stature, low body weight and skin atrophy. Patients are characterized by bird-like facial features. Basic treatments for progeria have not been found; symptomatic therapies including administration of growth hormones and a high-calorie diet are performed.

### 8. Acrogeria

**Synonym: Gottron’s syndrome**

Onset is thought to have a genetic contribution; however, the details are unknown. Skin atrophy and loss of subcutaneous fat are observed in the fingers, toes, nasal apex and auriculae. Acrogeria is a premature aging syndrome. It occurs most commonly in women. Atrophy, shortening and thickening of the nail plates occur. There are no systemic symptoms, and the prognosis is good; there are no basic treatments for acrogeria.

## B. Dysplasia

### 1. Congenital ectodermal dysplasia

This term is a catchall for congenital diseases of the hair, teeth, nails and sweat glands that cause abnormal formation of ectodermal tissue. It is classified into more than 100 subtypes according to the combinations of dysplastic components. Mutation in p63 has been found and reported. The main diseases caused by congenital ectodermal dysplasia are listed below.

1) **Anhidrotic (hypohidrotic) ectodermal dysplasia**

The main symptoms are thinning of hair, anhidrosis and abnormality in dental formation (Fig. 18.9). It is autosomal recessively inherited or X-linked, and is caused by mutation in the ectodysplasin anhidrotic receptor gene (EDAR) or the ectodysplasin-A receptor (EDARAV1).