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
Disorders of the Dermis and Subcutaneous Fat

18 Disorders of the Dermis and Subcutaneous Fat

2) Hidrotic ectodermal dysplasia (Clouston syndrome)

Deformity of nail plates, thinning of hair, and palmoplantar keratoderma are the three major symptoms. However, 30% of cases demonstrate only deformity of nail plates. Linear patterns form in the nail plates, which thicken and suffer from growth retardation. It is an autosomal dominant disease caused by mutation of the GJB6 gene, which codes for connexin 30 (Chapter 1). The prognosis is good.

2. Cutis verticis gyrata

Hyperplasia of the scalp results in skin folds at the top of the head. It occurs most commonly in boys. The folds are 1 cm to 2 cm wide, highly elastic and mobile. Normal hair growth is present in the groove portions, but not in the elevated portions (Fig. 18.10). Cutis verticis gyrata is classified into a primary form and a secondary form that accompanies nevoid abnormalities (e.g., nevus-cell nevus, connective tissue nevus) or systemic diseases (e.g., acromegaly). Plastic surgical repair may be conducted.

Pachydermoperiostosis (MIM, 167100) is a hereditary disease in which cutis verticis gyrata can be seen with clubbed fingers, osteohypertrophy and brawny skin change. It is autosomal dominant.

3. Chondrodermatitis nodularis chronica helicis

Painful keratotic nodules of 1 cm in diameter occur in the helices, particularly their upper parts (Fig. 18.11). These nodules result from chronic inflammatory reaction against collagen fibers that are degenerated by extrinsic stimulation such as of sunlight, injury or the cold. They occur most frequently in men of middle age and older. This disorder should be differentiated from seborrheic keratosis, basal cell carcinoma and squamous cell carcinoma. The main treatments are topical and local injection of steroids and surgical excision.