Chapter 24
Diseases of the Lymphatics

The diseases of the lymphatics are confined almost exclusively to the lower limbs and are far less common in the upper limbs, the trunk, and the head. The visible evidence of lymphatic damage is edema, i.e., lymphedema. It is possible to subdivide lymphedema into primary and secondary lymphedema.

**Definition.** A lymphedema arises when the transport capacity of the lymphatic vascular system is reduced to a level at which reabsorption and transport of the lymphatic protein load is impossible or inadequate.

**Primary Lymphedema.** This may be genetically determined or acquired. It is relatively uncommon and may develop uni- or bilaterally. The structural abnormalities of the lymph vessels are usually manifest even in young patients.

**Secondary Lymphedema.** This is not of genetic origin. It, too, can occur uni- or bilaterally, is relatively common, and affects mainly elderly persons. There are many different causes of secondary lymphedema: damage following surgery or trauma, tumors, inflammations of the lymphatic and venous systems, and cutaneous or subcutaneous infections with bacteria, treponemes, fungi, and worms.

**Clinical Findings.** An important clinical feature in chronic lymphedema is increased girth of the affected part of the body. First the swellings pit on pressure; later they are consolidated by reactive fibrosis, and the skin cannot be elevated (Stemmer sign over the toes). Other signs of chronic lymphedema are congestive papillomatosis, verruciform epidermal hyperplasia, hyperpigmentations, and secondary changes of the nails, e.g., onychodystrophy, onychogryphosis, onychodyschromasia, and slow nail growth. Usually, the regional lymph nodes are not involved.

**Primary Lymphedema**

**Hereditary Primary Lymphedema**

Hereditary primary lymphedema can appear unilaterally or bilaterally.

**Lymphedema, Nonne-Milroy Type** [Nonne 1891, Milroy 1892]

**Synonyms.** Hereditary congenital lymphedema, elephantiasis congenita hereditaria, trophedema.

**Definition.** Hereditary lymphedema of the leg.

**Occurrence.** Very rare. Inheritance is probably autosomal dominant. The disease appears congenitally. Girls are affected more often than boys. A dysplasia of the lymphatics is the subject of debate.

**Clinical Findings.** They occur symmetrically or only unilaterally on the feet and the lower legs. First there are painless doughy swellings which are initially produced by an infiltrating edema and are later consolidated by reactive tissue fibrosis. Therefore the swellings do not pit on pressure. Typically, edema does not transgress the groin.

**Histopathology.** Uncharacteristic. In histological sections, after routine embedding and staining, the lymphatic vessels are very difficult to find. In severe lymphedema there is cellular fibrosis.

**Symptoms.** Feeling of heaviness, restricted mobility and cosmetic problems.

**Course.** A deterioration often accompanies puberty.

**Treatment.** Symptomatic. Trial of the higher classes of compression bandages (III and IV).

**Lymphedema, Meige Type** [Meige 1898]

**Definition.** A clinical picture similar to that of the Nonne-Milroy type of lymphedema but with late onset.

**Occurrence.** The lymphedema first appears at puberty and is almost twice as common in girls than in boys.

**Etiology.** Unknown. Familial cases with autosomal dominant and autosomal recessive inheritance have been reported.

**Clinical Findings.** In addition to lymphedema as in the Nonne-Milroy type, associated features are stunted growth, hypogenitalism, microencephaly, mental retardation, obesity of the hips and thighs,
alterations of the blood vessels up to hemangiomas, yellow or dystrophic nails (possibly associated with the yellow-nail syndrome), ptosis of the eyelids, distichiasis, and recurrent intrahepatic cholestasis. In the presence of cholestatic features, tooth enamel defects and tooth discoloration can also develop. The cholestasis causes severe pruritus; in attacks the skin is jaundiced.

**Course.** Chronic-recurrent without healing of the lymphedema.

**Treatment.** As for Nonne-Milroy lymphedema.

### Ullrich-Turner Syndrome

[Ullrich 1930, Turner 1938]

This rare syndrome develops in chromatin-negative patients lacking a sex chromosome (45 + X0). The patients are of small size, with an adult height less than 145 cm. The phenotype is feminine and characterized by sexual infantilism and primary amenorrhea. Clinically striking is the pterygium (winged skin) in the neck region. The syndrome is often associated with congenital lymphedema of the dorsum of hands and feet. In contrast to the other primary lymphedemas, this lymphedema is peculiar in that it regresses in early adult life. Other features include shield-like chest with wide-spaced nipples, cubitus valgus, short fourth metacarpals, nail dystrophy, increased numbers of pigmented nevi, renal anomalies, hyposiderinemia, and hearing loss.

### Nonhereditary Primary Lymphedema

#### Essential Congenital Lymphedema

**Definition.** Nonfamilial form of congenital lymphedema.

**Etiology.** Unknown.

**Clinical Findings.** One or both legs can be affected. From birth, the extremity is swollen and indurated. In some cases swelling is localized to the feet or the calf, but usually the whole leg is enlarged.

#### Nonhereditary Idiopathic Lymphedema, Noncongenital

**Definition.** All forms of lymphedema of unknown etiology that are not hereditary and not congenital.

**Occurrence.** Most common form of lymphedema. In 65% of the cases the disease begins between 10 and 24 years of age (*lymphedema praecox*), and it is less common in older adults (*lymphedema tardum*). Women are more often affected than men.

**Clinical Findings.** Initially patients observe a swelling of the foot or of the ankle which increases in the summer, prior to menstruation, or in pregnancy. The swelling proceeds from distal to proximal within a matter of days, months, or even years. In later stages, lymphedema does not decrease after elevation of the limb. The course of the disease is often complicated by erysipelas. Later, secondary alterations of the skin occur. As in the essential congenital form, lymphedema is associated with other deformities, especially of the vascular system.

**Diagnosis.** This is a clinical diagnosis. Lymphangiography shows aplasia of major lymphatic vessels.

**Treatment.** Symptomatic.

**Systemic.** Diuretics.

**Topical.** Compression bandages, classes III–IV. Erysipelas prophylaxis.

### Secondary Lymphedema

The origin of secondary lymphedema may be inflammatory or noninflammatory, due to congestion. A guide to differential diagnosis is given in Table 24.1.

### Acute Lymphangitis

Acute inflammation of the lymphatics is shown by a reddened, often tender streaking of the skin along the lymph pathways. It occurs in numerous bacterial skin infections and is closely associated with the underlying infectious dermatosis. Treatment is according to the type of infection.

### Elephantiasis

**Definition.** Elephantiasis is a shapeless swelling of parts of the body due to chronic lymphatic obstruction, occlusion of the lymph vessels, and reactive fibrosing inflammation with connective tissue proliferation (fibrosis, induration, dermatisclerosis).

**Clinical Findings.** Initially the swellings pit on pressure, but with increasing fibrosis and formation of collagen fibers the skin becomes solid (*pachyderm*). The swellings increase continuously until they finally become monstrous. Elephantiasis is seen most frequently in the lower legs. Other skin areas, however, may be affected, including the genitalia (scrotum, penis, or labia), lips, ears, nose, eyelids, or backs of the hands. Function deteriorates with increasing degree of elephantiasis, e.g., the ability to walk if the legs are affected. In extreme cases a leg altered by elephantiasis can become so heavy and deformed that walking is impossible. The skin color is initially whitish-yellow, then bluish-red, and later dirty brown. The ini-