Fabry disease (angiokeratoma corporis diffusum universale) is a rare, X chromosome-linked lysosomal storage disease. The deficient enzyme, alpha-galactosidase A (alpha-gal A), is responsible for the accumulation of neutral glycosphingolipids within vascular endothelial lysosomes of various organs, including skin, kidneys, heart, and brain. The disease manifests primarily in affected hemizygous men and to some extent in heterozygous women ('carriers'). The diagnosis of Fabry disease is made in hemizygous males after the detection of the presence of angiokeratomas, irregularities in sweating, edema, scant body hair, painful sensations, and of cardiovascular, gastrointestinal, renal, ophthalmologic, phlebologic, and respiratory involvement. A deficiency of alpha-gal A in serum, leukocytes, tears, tissue specimens, or cultured skin fibroblasts further supports the diagnosis in male patients. Since heterozygous women show angiokeratomas in only about 30% of cases and may have alpha-gal A levels within normal range, genetic analysis is recommended. Current treatment of angiokeratomas of Fabry disease is based mainly on the use of laser systems, including variable pulse width 532nm Neodymium:Yttrium-Aluminum-Garnet (Nd:YAG) laser, 578nm copper vapor laser, and flashlamp-pumped dye laser. When cutaneous and mucous glands are affected, restrictions may be required with regard to the time
spent in a warm climate and the amount time spent working or on sporting activities, and may necessitate the use of topical and systemic antiperspirant agents, and topical application of artificial lacrimal fluid and saliva, respectively. For the future, new treatment modalities, including enzyme replacement therapy, substrate deprivation strategies, and gene therapy offer extraordinary options for the cutaneous and visceral lesions in patients with Fabry disease.