Refsum disease is one of a family of genetic disorders known as the leukodystrophies in which, as a consequence of the disruption of lipid metabolism, the myelin sheath that insulates and protects the nerves of the brain fails to grow. It is inherited as an autosomal recessive trait. It is characterized by progressive loss of vision (retinitis pigmentosa); degenerative nerve disease (peripheral neuropathy); failure of muscle coordination (ataxia); and dry, rough, scaly skin (ichthyosis). The age of onset of Refsum disease varies greatly. It may occur at any time from early childhood until around 50 years of age, but in most symptoms will have appeared by age 20. Males and females are affected in equal numbers. The presence of phytanic acid in blood or urine samples is diagnostic.