Refsum disease

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Refsum disease. ORPHA:773

Refsum disease (RD) is a very rare, clinically variable, multisystemic metabolic disease, characterized by anosmia, early-onset retinitis pigmentosa and possible neurological manifestations, including neuropathy, and cerebellar ataxia, deafness, ichthyosis, skeletal abnormalities, and cardiac arrhythmia. It is characterized biochemically by accumulation of phytanic acid in plasma and tissues.