Refsum Disease

National Cancer Institute

Source

National Cancer Institute. Refsum Disease. NCI Thesaurus. Code C85043.

A rare autosomal recessive condition caused by mutation(s) in the PHYH gene, encoding phytanoyl-CoA dioxygenase, peroxisomal. It is characterized by abnormalities in the breakdown of phytanic acid and impaired growth of myelin sheaths. Signs and symptoms include neurologic damage, cerebellar degeneration, and neuropathy.