Open Peer Review on Qeios

Peroxisome Biogenesis Disorder 4B

National Cancer Institute

Source

National Cancer Institute. <u>Peroxisome Biogenesis Disorder 4B</u>. NCI Thesaurus. Code C155755.

An autosomal recessive condition caused by mutation(s) in the PEX6 gene, peroxisome assembly factor 2, Peroxisome biogenesis disorder 4B is characterized by overlapping phenotypes of neonatal adrenoleukodystrophy and infantile Refsum disease.