Open Peer Review on Qeios

Peroxisome Biogenesis Disorder 3B

National Cancer Institute

Source

National Cancer Institute. <u>Peroxisome Biogenesis Disorder 3B</u>. NCI Thesaurus. Code C155753.

An autosomal recessive condition caused by mutation(s) in the PEX12 gene, encoding peroxisome assembly protein 12. Peroxisome biogenesis disorder 3B is characterized by overlapping phenotypes of neonatal adrenoleukodystrophy and infantile Refsum disease.