Peroxisome Biogenesis Disorder 2B

National Cancer Institute

Source

National Cancer Institute. Peroxisome Biogenesis Disorder 2B. NCI Thesaurus. Code C155751.

An autosomal recessive condition caused by mutation(s) in the PEX5 gene, encoding peroxisomal targeting signal 1 receptor. Peroxisome biogenesis disorder 2B is characterized by overlapping phenotypes of neonatal adrenoleukodystrophy and infantile Refsum disease.