Mild Non-BH4-Deficient Hyperphenylalaninemia

National Cancer Institute

Source

National Cancer Institute. Mild Non-BH4-Deficient Hyperphenylalaninemia. NCI Thesaurus. Code C159654.

An autosomal recessive condition caused by mutation(s) in the DNAJC12 gene, encoding dnaJ homolog subfamily C member 12. It is characterized by increased serum phenylalanine concentrations resulting in variable neurologic defects, including movement defects and intellectual disability. BH4 metabolism is normal.