Hyperphenylalaninemia, BH4-deficient C

National Cancer Institute

Source

National Cancer Institute. Hyperphenylalaninemia, BH4-deficient C. NCI Thesaurus. Code C138173.

An autosomal recessive condition caused by mutation(s) in the QDPR gene, encoding dihydropteridine reductase. It is characterized by BH4-defecient hyperphenylalanemia, depletion of dopamine and serotonin, and progressive cognitive and motor deficits.