Hyperphenylalaninemia, BH4-deficient A

National Cancer Institute

Source

National Cancer Institute. Hyperphenylalaninemia, BH4-deficient A. NCI Thesaurus. Code C138171.

An autosomal recessive condition caused by mutation(s) in the PTS gene, encoding 6-pyruvoyl tetrahydrobiopterin synthase. It is characterized by BH4-defecient hyperphenylalanemia, depletion of dopamine and serotonin, and progressive cognitive and motor deficits.