Hyperphenylalaninemia due to tetrahydrobiopterin deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hyperphenylalaninemia due to tetrahydrobiopterin deficiency. ORPHA:238583

Hyperphenylalaninemia (HPA) due to tetrahydrobiopterin (BH4) deficiency, also known as malignant HPA is an amino acid disorder with neonatal onset that is clinically characterized by the classic manifestations of phenylketonuria (PKA; see this term) and that later on is clinically differentiated by neurologic symptoms such as microcephaly, intellectual disability, central hypotonia, delayed motor development, peripheral spasticity and seizures, that develop and persist despite an established metabolic control of plasma phenylalanine.