Climb™

Supporting those affected by Inherited Metabolic Disorders

6-Pyruvoyl-Tetrahydrobiopterin Synthase Deficiency

6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency is a cause of malignant hyperphenylalaninaemia caused by tetrahydrobiopterin (BH4) deficiency. Hyperphenylalaninaemia is characterised by elevated concentrations of the amino acid phenylalanine (Phe) in the blood. If left untreated, PTPS deficiency causes neurological problems within the first 6 months of life, although signs of the condition are often obvious from birth. This condition should be suspected in all infants with a positive neonatal screening test for phenylketonuria.

Synonyms

Alternative names for this condition are:

- 6PTS Deficiency
- Hyperphenylalaninemia, BH4-deficient
- Hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency
- PTPS Deficiency
- PTS Deficiency

Further information about this condition is available from Climb.

Disclaimer

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