

NSPKU welcomes families living with Allied disorders such as BH4 deficiencies like Dihydropteridine Reductase Deficiency (DHPR deficiency)

DHPR deficiency is one of 6 different BH4 (tetrahydrobiopterin) deficiencies. It is a metabolic disorder affecting the nervous system and, like PKU, it is an autosomal recessive inherited disorder. The affected gene in DHPR deficiency is the gene for recycling BH4 in the body.

BH4 is responsible for making dopamine and serotonin which are two vital brain messengers. BH4 deficiency affects these brain messengers (neurotransmitters), and this can lead to low muscle tone, abnormal movements, swallowing difficulties, and problems with focus and learning. As with PKU, phe can build up in the body of the person affected.

Incidence: BH4 deficiencies are very rare. They comprise around 1-2% of people with raised Phe levels. DHPR is the second most common BH4 deficiency and people with DHPR make up one third of all the BH4 deficiencies.

Screening: DHPR deficiency is first detected through raised phenylalanine levels on newborn screening- and can be initially mistaken for Phenylketonuria (PKU). All patients who have raised phenylalanine on newborn screening are tested for BH4 deficiencies, the results of these tests are usually available in 2 weeks. If a BH4 deficiency is suspected, further testing is done.



Symptoms of DHPR deficiency can include:

- Low birth weight
- Restricted head growth
- Seizures
- Abnormal limb movements
- Developmental/cognitive impairment
- Emotional Lability
- Impaired speech development
- Excessive sweating / salivation
- Swallowing and feeding difficulties

Treatment

Early diagnosis is important as treatment in early infancy can lead to a good outcome.

- A low phenylalanine diet: to lower blood phenylalanine levels. The use of sapropterin may be considered.
- Regular medications (L-dopa and

5-hydroxytryptophan) to replace the neurotransmitters that cannot be made in the brain.

- Folinic acid supplementation, to help with maintaining folate levels

Families or individuals living with DHPR deficiency all need to monitor and control blood phe levels in the same way people with PKU do.

