Climb™

Supporting those affected by Inherited Metabolic Disorders

Sepiapterin Reductase Deficiency

Sepiapterin Reductase Deficiency is a rare disorder that is caused by a defect in the SPR gene which provides instructions for the sepiapterin reductase enzyme. This enzyme is involved in the production of tetrahydropterin (BH4) which processes specific amino acids and helps in the production of neurotransmitters which are vital chemicals that enable the nerve cells to communicate with each other. Because of the lack of BH4 in the brain, not enough dopamine and serotonin is produced which are needed for different functions including physical movement and regulating appetite, sleep and mood.

This condition usually presents in the first few weeks of life. Severity varies greatly and can range from an often misdiagnosed or undiagnosed mild movement disorder to severe progressive neurological disease. Over 40 cases have been reported so far, however the true incidence is unknown due to the mild cases being misdiagnosed or undiagnosed.

Synonyms

Alternative names for this condition are:

- Dopa-responsive dystonia due to sepiapterin reductase deficiency
- SRD
- SPR Deficiency

Further information about this condition is available from Climb.

Disclaimer

Updated 06/04/17

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