Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Cerebral Folate Transport Deficiency

Cerebral Folate Transport Deficiency is caused by a defect in the FOLR1 gene which is needed to produce the folate receptor alpha protein. This protein is needed to help folate (vitamin B9) to be brought from the blood stream into the fluid which surrounds the brain and spinal cord (CSF) enabling it to enter the brain cells. The brain needs folate to make neurotransmitters and myelin (a fatty substance covering the nerve fibres, making up the brains white matter). These are both needed to send messages across the nervous system. A deficiency of the protein causes many processes in the brain to be impaired due to a lack of folate. The deficiency also causes Leukodystrophy which is a loss of the white matter and further contributes to the neurological symptoms in this condition.

Signs and symptoms present in late infancy and include a loss of skills, movement problems, intellectual problems, speech problems and seizures.

Synonyms

Alternative names for this condition are:

- Cerebral Folate Deficiency
- FOLR1 Deficiency
- Neurodegeneration due to Cerebral Folate Transport Deficiency

Further information about this condition is available from Climb.

Disclaimer

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