Climb™

Supporting those affected by Inherited Metabolic Disorders

Maple Syrup Urine Disease

This rare inherited metabolic disease has been named after the unusual odour of the urine, sweat and ear wax that arises in the untreated condition. Health problems occur because the body is unable to break down three amino acids (part of the 20 different building blocks of proteins) known as leucine, isoleucine and valine. The inability to break down these amino acids leads to a build up of acid levels in the blood that becomes toxic and can lead to damage of some tissues in the body, in particular the brain. There is wide variety in the severity. The most severely affected children develop symptoms soon after birth, whereas people with milder variants may present in later childhood.

MSUD is now routinely tested for shortly after birth as part of the Newborn Bloodspot Screening Programme. If MSUD is diagnosed, treatment is started immediately. Newborn screening and early management greatly improves outcomes and prognosis. Without lifelong treatment and management, severe and life-threatening symptoms can develop, including brain damage, seizures or coma.

Synonyms

Alternative names for this condition are:

- BCKD Deficiency
- Branched Chain Alpha-Ketoacid Dehydrogenase Deficiency
- MSUD
- MSD

Further information about this condition is available from Climb.

Disclaimer

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