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OPINION ARTICLE

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Signs and Symptoms of Maple Syrup Urine Disease

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Description

Branched-chain amino acids are impacted by Maple Syrup Urine Disease (MSUD), an autosomal recessive metabolic illness. This particular case of organic academia, named after the unique sweet smell of the affected infants' urine and earwax, especially before diagnosis and during periods of acute sickness. The indications and symptoms of maple syrup urine disease can be categorised, as well as the hereditary aetiology. The classic form of this illness is the most prevalent and severe type; it manifests shortly after birth and, if left untreated, results in symptoms that worsen over time and never go away. Variant forms of the disorder may not manifest until later in infancy or youth, and they often have milder symptoms that may only manifest during periods of fasting, stress, or illness. However, if ignored, these forms of the disorder can still cause mental and physical difficulties [1,2].

Signs and symptoms

The condition is so named because, when a person enters a metabolic crisis, their urine has a sweet, maple syrup-like odour. During a metabolic crisis, the stench can also be detected in an affected person's ear wax. The aroma of maple syrup can be compared to that of fenugreek in communities where that herb is unfamiliar, and fenugreek consumption can cause urine to take on that aroma. The degree of remaining enzyme activity has a significant impact on the symptoms of MSUD, which vary widely amongst patients [3-5].

Classic MSUD

Within the first 24 to 48 hours, infants with classic MSUD will start to exhibit mild symptoms. Lethargy, impatience, and poor bottle- or breast-feeding are all subtle indications. The baby will thereafter display more pronounced focal neurologic symptoms. These neurological symptoms, which cause convulsions and unconsciousness, include athetoid, hypertonia, spasticity, and opisthotonus. Untreated MSUD will result in central neurologic dysfunction, respiratory failure, and death. The risks of metabolic decompensation and bone mass loss, which can result in osteoporosis, pancreatitis, and intracranial hypertension, persist even after MSUD has been controlled. Intellectual disability and behavioural difficulties are additional classic MSUD symptoms that may be present [6].

Intermediate MSUD

Compared to classic MSUD, intermediate MSUD has higher levels of lingering enzyme activity. Most kids with intermediate MSUD receive their diagnosis between the ages of 5 months and 7 years. Intermediate MSUD also exhibits symptoms that are similar to those of classic MSUD.

Intermittent MSUD

Individuals with intermittent MSUD will experience normal growth and intellectual development in contrast to those with classic and intermediate MSUD. When a person is under stress, doesn't eat, or is sick, lethargic symptoms and a distinctive maple syrup odour appear. There is still a chance of a metabolic crisis that results in seizures, coma, and brain damage [7,8].

Thiamine-response MSUD

The signs and symptoms of intermediate MSUD are similar to those of thiamine-responsive MSUD. Rarely do newborns exhibit symptoms.

Later onset

Depending on how severe the condition is, MSUD symptoms may also develop over time. Untreated symptoms of the condition include extreme or erratic behaviour and moods, hallucinations, lack of appetite, weight loss, anaemia, diarrhoea, vomiting, dehydration, lethargy, oscillating hypertonia and hypotonia, ataxia, seizures,

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hypoglycemia, ketoacidosis, opisthotonus, pancreatitis, rapid neurological decline, and coma in older people and during times of metabolic crisis. If there is no therapy, cerebral edema-related death will probably happen. Additionally, MSUD patients undergo atypical disease progression in mild illnesses that might cause long-term harm.

Prevention

Infants with two faulty copies of the BCKD gene cannot be treated to stop the pathology of MSUD from showing up. To screen for the condition, however, couples may speak with genetic counsellors and undergo DNA testing. The condition can also be detected in an unborn kid in the pregnancy *via* DNA testing.

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