Maple Syrup Urine Disease (MSUD)

Maple Syrup Urine Disease is an autosomal recessive disorder caused by the inability to metabolize the amino acids leucine, isoleucine and valine. This disease is so named because the urine of affected people smells like maple syrup. Early neonatal symptoms include poor feeding, lethargy, seizures, coma and ketoacidosis as seen in the first week of life. Several different variants have been found with MSUD which differ in severity, age of onset, clinical symptoms and thiamine responsiveness.

Prevalence:	1: 290,000
Anaylte Measured:	Leucine and isoleucine
Reporting Ranges:	Elevated levels of leucine and isoleucine
Feeding Effect:	None
Timing Effect:	None
Confirmation:	Quantitative measurement of leucine, isoleucine and valine
Treatment:	Referral is made to a Metabolic Specialist. Correct any symptoms of dehydration, electrolyte imbalances and metabolic acidosis. A dietary consult is made for a special MSUD formula low in BCAA and a diet low in protein.
Comment:	MSUD is a chronic long-term disorder where the patient may decompensate when stressed and is at risk of mental and neurological deficits and sudden death. Strict compliance with treatment is necessary to prevent neurological damage. Affected people must stay on this special diet for life.

Maine Newborn Screening Program
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