(2)

(*) Maple Syrup Urine disease. (MSUD)

→ MSUD also called Branched Chain Ketonuria

→ Incidence is 1 per 1 lakh births

→ Characteristic smell of urine similar to burnt sugar or maple sugar due to excretion of branched chain keto acids.

→ Biochemical defect – Deficient decarboxylation of Branched Chain keto acid.

→ Oxidative decarboxylation with the help of CoA, NAD+, and branched chain

alpha - keto acid dehydrogenase

↓

Lacking In Maple Syrup Urine disease

which convert

Valine – Isobutyryl - CoA
Leucine – Isovaleryl - CoA
Isoleucine – Alpha - methylbutyryl - CoA

→ **Clinical finding**

- disease start in the first week of life
- Convulsions, severe mental Retardation Vomiting, Acidosis, coma
- Death within first year of life.
- fatal Ketoacidosis

Laboratory finding

- Plasma and Urinary level of leucine, Isoleucine, Valine and their alpha keto acid are elevated,
- Rothera's test is Positive
- Diagnosis depend on enzyme analysis in cells.
- Diagnosis should be done prior to 1 week after birth.

MSUD

Normal

Protein from food → Branched-Chain Amino Acid (BCAAs)

Protein from Muscle → Branched-Chain Amino Acid (BCAAs)

Branched Chain Keto acid Dehydrogenase (BCKAD)

↓

Energy

↓

Growth

MSUD

Protein from food → BCAAs (Branched-Chain Amino Acids)

Protein from Muscles → BCAAs (Branched-Chain Amino Acids)

BCKAD enzyme ✗ deficient

↓

Build up of BCAAs and other Substances

↓

Acidosis & Mental Retardation

Treatment

→ Low protein diet, low quantity of branched chain amino acid.

→ Mild Variant is called Intermittent branched chain ketonuria, this will respond to high doses of thiamine

→ Because decarboxylation of BKA requires thiamine.

→ LIVER transplantation successfully tried in some cases of MSUD.