Inheritance / Prevalence

- Autosomal recessive
- About 1 in 185,000 children
- More common among Mennonites
The Genotype to Phenotype of MSUD

Mutated BCDKHA/B, DBT, or DLD genes inherited
The Genotype to Phenotype of MSUD

Unable to produce enzymes in digestive complex
The Genotype to Phenotype of MSUD

Unable to decarboxylate branched chain amino-acids
The Genotype to Phenotype of MSUD

BCAA build up
The Genotype to Phenotype of MSUD

First week: Sweet urine smell
The Genotype to Phenotype of MSUD

The Genotype to Phenotype of MSUD

First month: Developmental damage and seizures
The Genotype to Phenotype of MSUD

< 1 year: Death
Maple Syrup Urine Disease
MSUD

Normal

Protein from food
Protein from muscles

Amino Acids

Branched-Chain Amino Acids (BCAAs)

BCKAD enzymes

Energy
Growth

MSUD

Protein from food
Protein from muscles

Amino Acids

Branched-Chain Amino Acids (BCAAs)

BCKAD enzymes

Build up of BCAAs and other substances

Health Problems

Energy
Growth

http://www.newbornscreening.info/tools/GraphicsLib.html
Diagnoses since ~1955

CLASSICAL

Smell of urine  Elevated BCAA levels

MODERN

Gene Sequencing
Treatments

CLASSICAL

Attempted protein restriction

MODERN

Formula diet | Liver transplants
Bibliography