

Maple Syrup Urine Disease

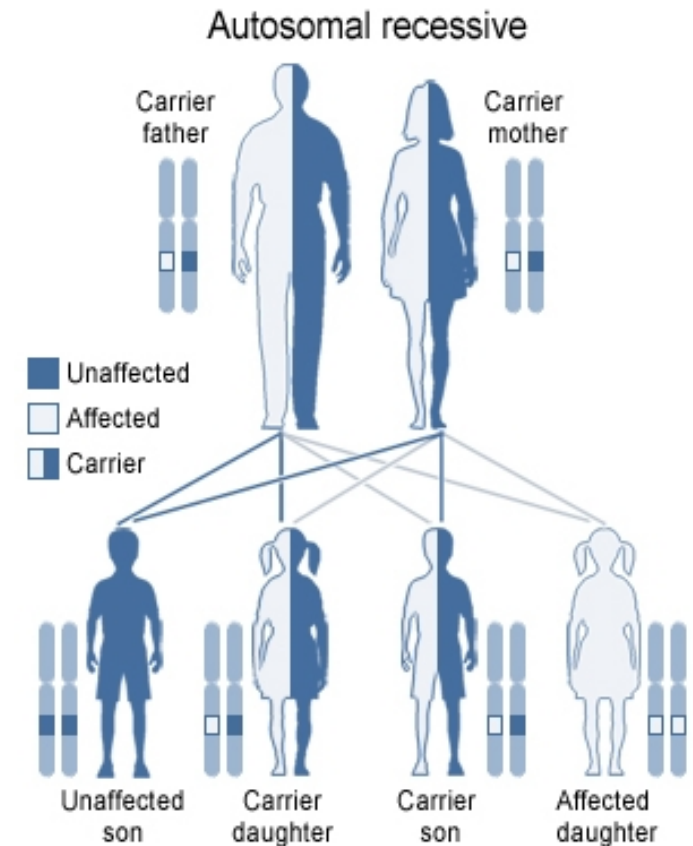
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BIOC118, Winter 2011

Professor Doug Brutlag

Inheritance / Prevalence

- Autosomal recessive
- About 1 in 185,000 children
- More common among Mennonites



U.S. National Library of Medicine

The Genotype to Phenotype of MSUD



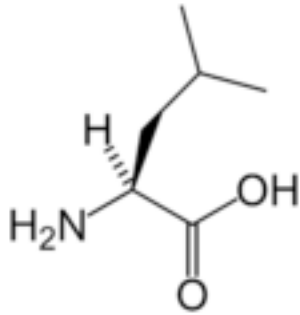
Mutated BCDKHA/B, DBT, or
DLD genes inherited

The Genotype to Phenotype of MSUD

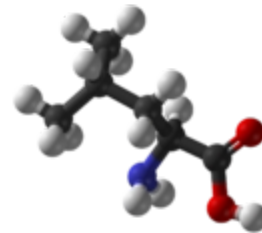


Unable to produce enzymes
in digestive complex

The Genotype to Phenotype of MSUD



Unable to decarboxylate
branched chain amino-acids



The Genotype to Phenotype of MSUD



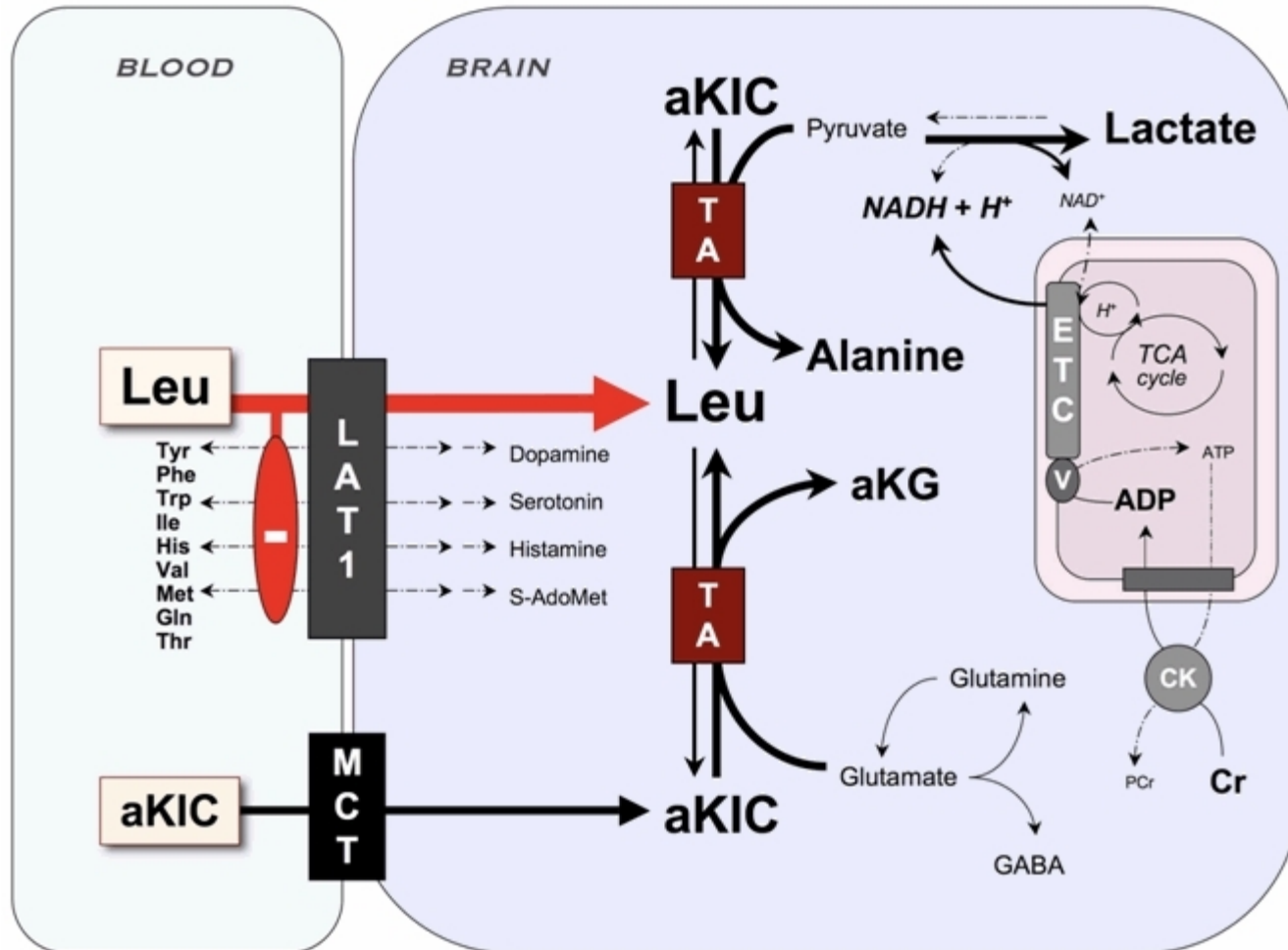
BCAA build up

The Genotype to Phenotype of MSUD



First week: Sweet urine
smell

The Genotype to Phenotype of MSUD



The Genotype to Phenotype of MSUD

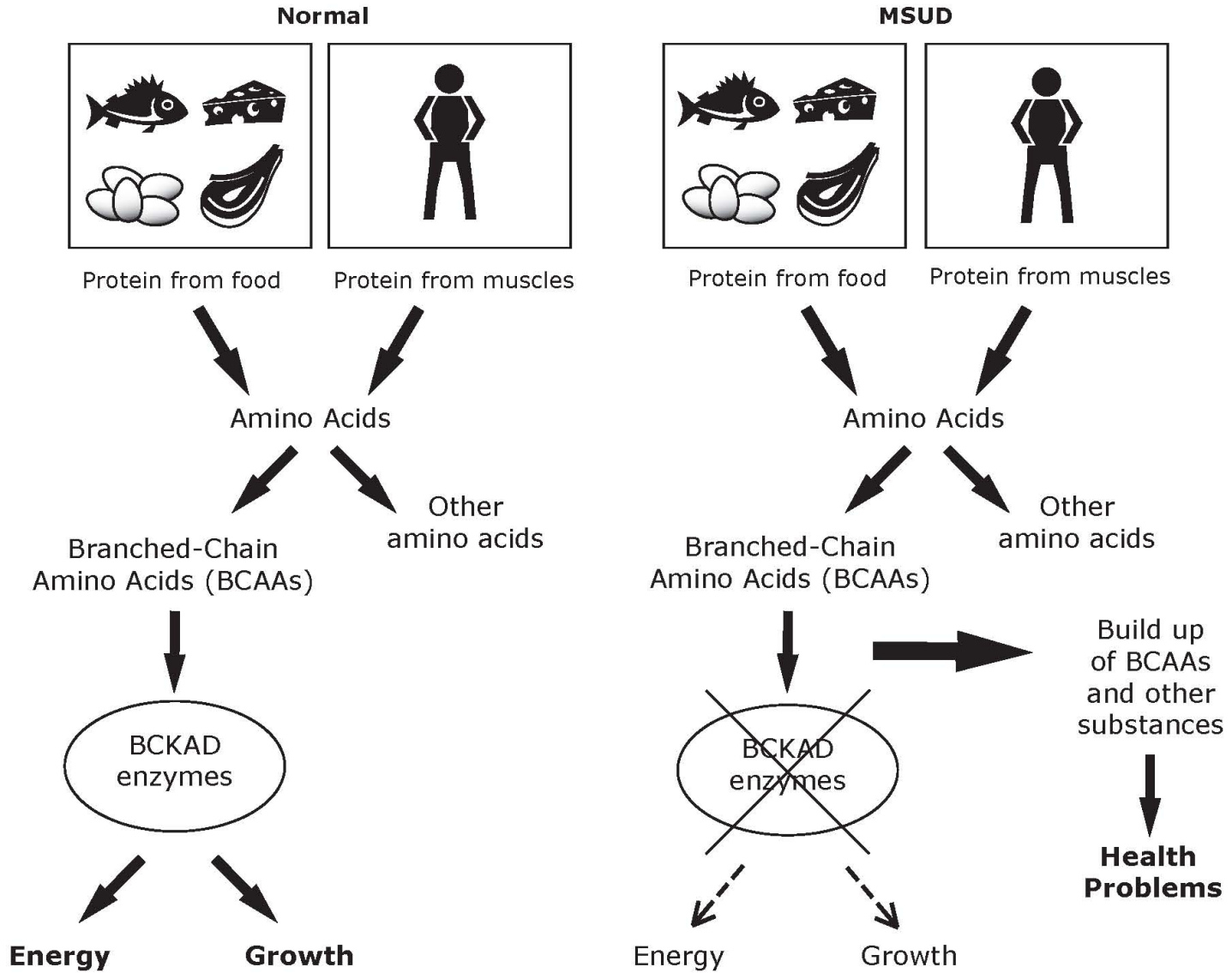


First month: Developmental
damage and seizures

The Genotype to Phenotype of MSUD

< 1 year: Death

Maple Syrup Urine Disease MSUD



Diagnoses since ~1955

CLASSICAL

Smell of urine

Elevated BCAA levels



MODERN

Gene Sequencing

Treatments

CLASSICAL

Attempted protein restriction



MODERN

Formula diet

Liver transplants

Bibliography

- <http://www.ncbi.nlm.nih.gov/books/NBK1319/>
- <http://www.ncbi.nlm.nih.gov/omim/248600>
- <http://ghr.nlm.nih.gov/condition/maple-syrup-urine>
- http://www.msud-support.org/index.php?option=com_content