Phenyllactate
Phenylpyruvate
Phenylalanine
Tyrosine
Melanin
ALBINISM (see text)
PHENYLKETONURIA (see text)
Catecholamines
p-Hydroxyphenylpyruvate
Homogentisate
ALKAPTONURIA (see text)
Maleylacetoacetate
Fumarylacetoacetate
Acetoacetate
FUMARATE
Succinate
Malate
Purines
Creatine
Porphyrins
Conjugates
3-P-Glycerate
Glycine
Serine
Homocysteine
Cystathionine
α-Ketobutyrate
Cysteine
Asparagine
Aspartate
Purines
Pyrimidines
Alanine
PYRUVATE
ACETYL COA
OXALOACETATE
Citrate
α-KETOGLUTRATE
SUCCINYL COA
MAPLE SYRUP URINE DISEASE
(see "Valine" and "Isoleucine" below)
Leucine
α-Ketoisocaproate
Isovaleryl CoA
Threonine
ACETOACETYL COA
Alanine
Lysine
Tryptophan
NAD(H)
Serotonin
Creatine
Arginine
Urea
Ornithine
Glutamate
Proline
Glutamine
Urocanate
Histidine
Histamine
Purines
Pyrimidines
Methylmalonyl CoA
Methylmalonyl semialdehyde
Propionyl CoA
Acetyl CoA
Threonine
α-Ketobutyrate
Isobutyryl CoA
α-Methylbutyryl CoA
α-Ketoisovalerate
α-Keto-β-Methylvalerate
Valine
Isoleucine
Cysteine
Cystathionine
Serine
Homocysteine
S-Adenosylhomocysteine
S-Adenosylmethionine
Methionine

TYROSINEMIA TYPE I

- The disease is due to a deficiency in *fumaryl-acetoacetate hydrolase*.
- Fumarylacetoacetate and its metabolites, particularly succinyl-acetone, accumulate in the urine.
- Characteristic cabbage-like odor occurs.
- Liver failure and renal tubular acidosis result.
- Treatment includes dietary restriction of phenylalanine and tyrosine and substrate reduction therapy.

METHYLMALONYL CoA MUTASE DEFICIENCY

- The disease is due to a deficiency in *methylmalonyl CoA mutase*.
- Elevated levels of methylmalonic acid (methylmalonate) occur in blood.
- Metabolic acidosis and developmental problems occur.

HISTIDINEMIA

- The disease is due to a deficiency in *histidase*.
- Elevated levels of histidine occur in blood and urine.
- No clinical symptoms are present in most individuals.

CYSTATHIONINURIA

- Accumulation of cystathionine and its metabolites is due to a rare deficiency in *cystathionase*.
- No clinical symptoms are present.

MAPLE SYRUP URINE DISEASE

- The disease is due to a deficiency in *branched-chain α-keto acid dehydrogenase*.
- Levels of branched-chain α-amino acids and their α-keto analogs are elevated in plasma and urine.
- Neurologic problems are common. The disease has a high mortality rate.
- Treatment involves a restricted dietary intake of the branched-chain amino acids.

HOMOCYSTINURIA

- The disease is due to a deficiency in *cystathionine synthase*.
- Accumulation of homocysteine occurs in the urine.
- Methionine and its metabolites are elevated in the blood.
- Intellectual disability, osteoporosis, myocardial infarction, and a characteristic dislocation of the lens occur.

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