

Inborn Errors of Metabolism

GLYCOGEN STORAGE DISEASES

MUSCLE GLYCOGENOSES

Muscle glycogen phosphorylase (2.4.1.1)
GSD Type V McArdle Disease
 MIM 232600

LIVER GLYCOGENOSES

Amylo-1,6-glucosidase (3.2.1.33)
 Glucanotransferase (2.4.1.25)
GSD Type III Cori Disease
 MIM 232400

Glucose-6-phosphatase (3.1.3.9)
GSD Type I von Gierke Disease
 MIM 232200

MONO- & DI-SACCHARIDES

Sucrase-isomaltase (3.2.1.10)
Sucrose-Isomaltase Malabsorption
 MIM 609845

Lactase (3.2.1.23) (3.2.1.108)
Lactose Intolerance
 MIM 223000

Galactokinase (2.7.1.6)
Galactose Kinase Deficiency
 MIM 230200

Galactose-1-phosphate uridyl transferase (2.7.7.12)
Classical Galactosaemia
 MIM 606999

PEROXISOMAL DEFECTS

Alanine-glyoxylate aminotransferase (2.7.1.44)
Hyperoxaluria Type I
 MIM 259900

LIPID METABOLISM

Hydroxymethylglutaryl CoA lyase (4.1.3.4)
Ketone Synthesis Defect
 MIM 246450

Medium chain acyl CoA dehydrogenase (1.3.99.3)
MCAD Deficiency
 MIM 607008

SPHINGOLIPIDOSES

Acid-β-glucosidase (3.2.1.45)
Gaucher Disease
 MIM 230800

Hexosaminidase A (3.2.1.52)
Tay Sachs Disease
 MIM 272800

Sphingomyelinase (3.1.4.12)
Niemann-Pick Disease
 MIM 257200

RESPIRATORY CHAIN DEFECTS

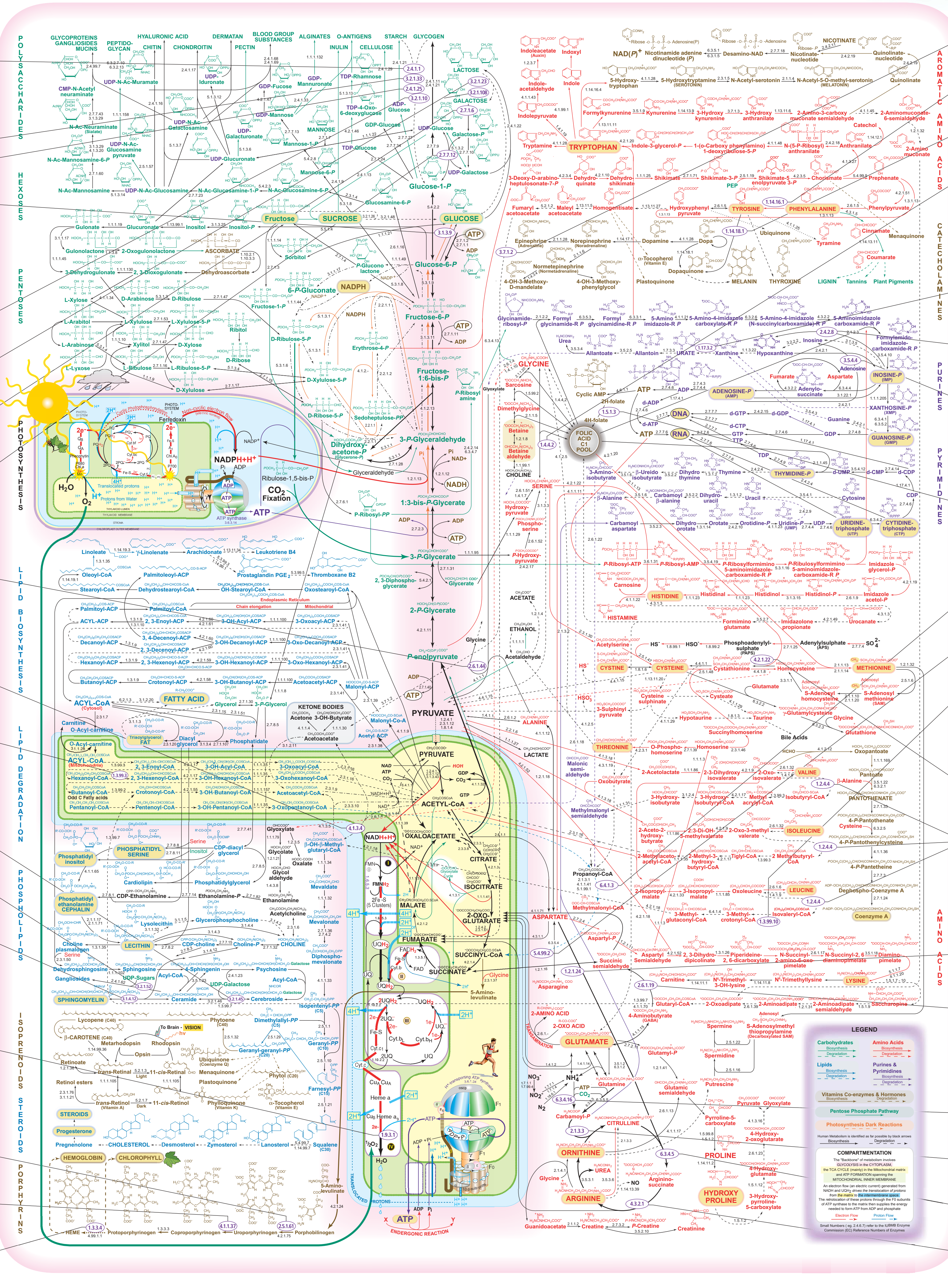
Cytochrome c oxidase (1.9.3.1)
Complex IV
 MIM 220110

PORPHYRIAS

Porphobilinogen deaminase (2.5.1.61)
Acute Intermittent Porphyria
 MIM 176000

Uroporphyrinogen decarboxylase (4.1.1.37)
Porphyria Cutanea Tarda
 MIM 176100

Protoporphyrinogen oxidase (1.3.3.4)
Variegate Porphyria
 MIM 176200



AMINO ACID METABOLISM

Fumarylacetoacetase (3.7.1.2)
Tyrosinaemia Type I
 MIM 276700

Phenylalanine hydroxylase (1.14.16.1)
Phenylketonuria (PKU)
 MIM 261600

Tyrosinase (1.14.18.1)
Tyrosine Negative Albism
 MIM 203100

Glycine dehydrogenase (1.4.4.2)
Non Ketotic Hyperglycinaemia (NKH)
 MIM 238300

PURINE & PYRIMIDINE METABOLISM

Xanthine oxidase (1.17.3.2)
Xanthinuria
 MIM 278300

Hypoxanthine guanine phosphoribosyl transferase (2.4.2.8)
HGPRT Deficiency, Primary Gout, Lesch-Nyhan Syndrome
 MIM 308000

Adenosine deaminase (3.5.4.4)
Severe Combined Immuno-Deficiency (SCID)
 MIM 102700

FOLIC ACID

Dihydrofolate reductase (1.5.1.3)
Megaloblastic Anaemia
 MIM 126060

ORGANIC ACIDURIAS

Propionyl-CoA carboxylase (6.4.1.3)
Propionic Acidemia
 MIM 606054

Methylmalonyl-CoA mutase (5.4.99.2)
Methylmalonic Acidemia
 MIM 251000

Isovaleryl-CoA dehydrogenase (1.3.99.10)
Isovaleric Acidemia
 MIM 243500

AMINO ACID METABOLISM

Cystathionine synthetase (4.2.1.22)
Homocystinuria
 MIM 236200

Branched chain ketoacid decarboxylase (1.2.4.4)
Maple Syrup Urine Disease
 MIM 248600

GABA METABOLISM NEUROTRANSMITTER DEFECTS

Succinic semialdehyde dehydrogenase (1.2.1.24)
4-Hydroxybutyric Aciduria
 MIM 271980

4-Aminobutyrate aminotransferase (2.6.1.19)
GABA Transfere Deficiency
 MIM 137150

UREA CYCLE DEFECTS (HYPERAMMONAEMIA)

Carbamoyl phosphate synthase (6.3.4.16)
CPS Deficiency
 MIM 237300

Ornithine carbamoyl transferase (2.1.3.3)
OCT Deficiency
 MIM 311250

Argininosuccinate synthase (6.3.4.5)
Citrullinaemia
 MIM 215700

Argininosuccinate lyase (4.3.2.1)
Argininosuccinate Aciduria
 MIM 207900