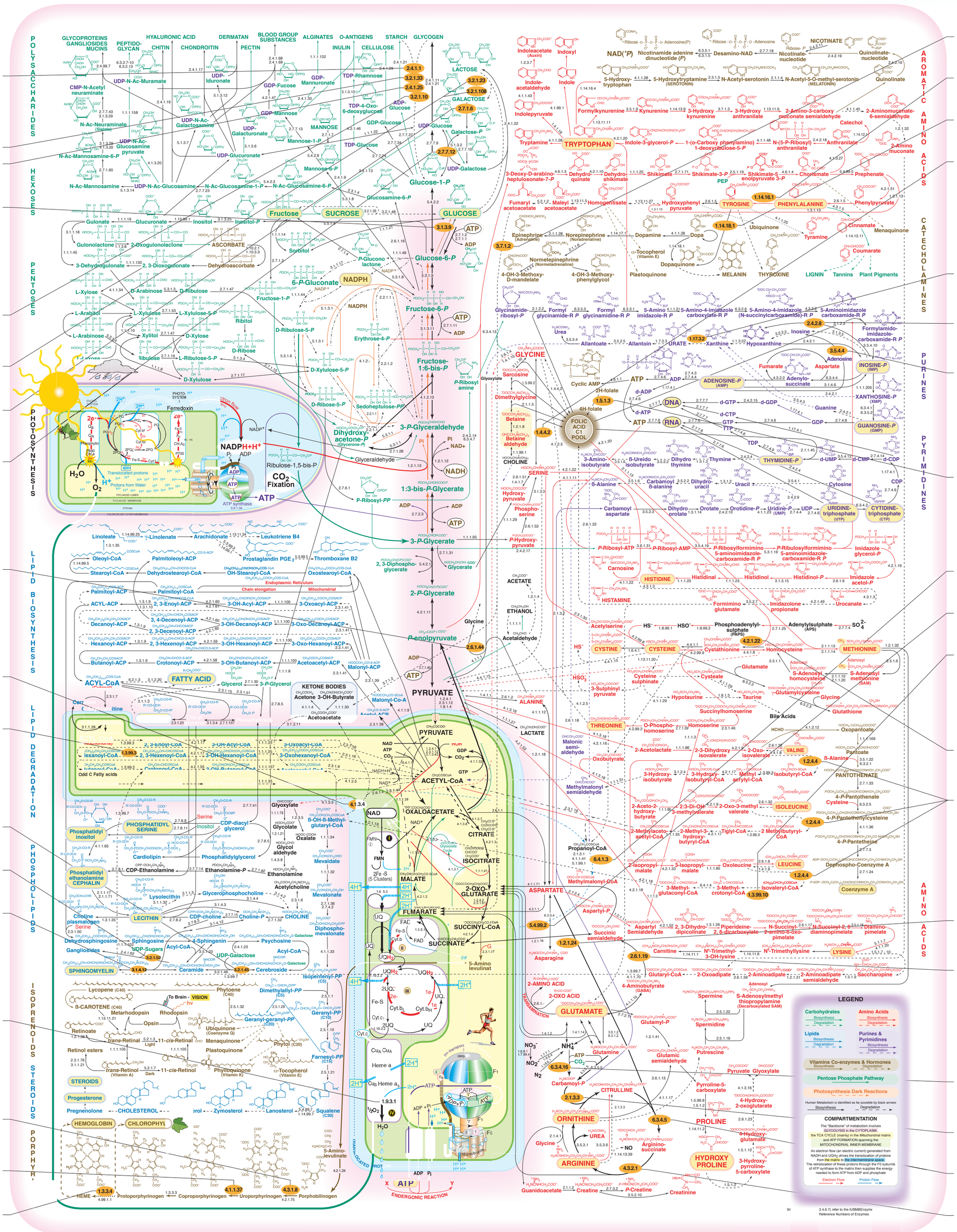
Inborn Errors of Metabolism



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-Isomaltose 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 (2.6.1.44)
Hyperoxaluria Type 1
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-ß-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 Albinism
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 phospho-ribosyl 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 Acidaemia
MIM 606054

Methylmalonyl-CoA mutase (5.4.99.2)
Methylmalonic Acidaemia
MIM 251000

Isovaleryl-CoA dehydrogenase (1.3.99.10)
Isovaleric Acidaemia
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 Transferase 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 300161

Argininosuccinate synthase (6.3.4.5)
Citrullinaemia
MIM 215700

Argininosuccinate lyase (4.3.2.1)
Argininosuccinate Aciduria
MIM 207900

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Designed by Donald E.Nicholson,The University of Leeds,England – and Sigma-Aldrich in collaboration with Mick Henderson,St.James' Hospital,Leeds – and the UK MetBioNet Training Group www/metbio.net.
For further information about metabolic disorders visit the OMIM (Online Mendelian Inheritance in Main) website, hosted by the Johns Hopkins University – www.ncbi.nlm.gov/entrez/query/fcgi?db=OMIM.

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GDS