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| Hereditary metabolic disorders (DMPs) | |
|---------------------------------------|--|
| In general | DMP of complex molecules • DMP of small molecules • Neonatal screening • Screening of hereditary diseases • Examination methods at DMP |
| DMP amino acids | Alkaptonuria |
| Organic aciduria | - |
| DMP urea cycle | Alcaptonuria • Ornithine transcarbamylase deficiency • Prolidase deficiency • Phenylketonuria • Glutaric aciduria • Hyperphenylalaninemia • Hyperornithinemia • Isovaleric aciduria • Leucinosi • Non-ketotic hyperglycemia • Cystinosis • Tyrosinemia |
| DMP propionate, biotin and cobalamin | Biotinidase deficiency • Methylmalonic acidemia • Propionic acidemia |
| DMP purines and pyrimidines | Liver porphyria • Skin porphyria • Mitochondrial neurogastrointestinal encephalomyopathy |
| DMP sugars | Glycogenoses • Fructosealdolase deficiency • Fructose-1,6-bisphosphatase deficiency • Essential fructosuria • Galactokinase deficiency • Galactose-1-phosphate uridylyltransferase deficiency |
| DMP mitochondria | Phosphoenolcarboxykinase Deficiency • LCHAD Deficiency • MCAD Deficiency • Pyruvate Dehydrogenase Deficiency • Pyruvate Carboxylase Deficiency • SCAD Deficiency • Chronic Progressive External Ophthalmoplegia • Leber's Hereditary Optic Neuropathy • Leigh Syndrome • Maternally Inherited Diabetes and Deafness • SLC26A5 Syndrome |
| DMP peroxisomes | Neonatal adrenodystrophy • Refsum's disease • Rhizomelic chondrodysplasia punctata • X-linked adrenoleukodystrophy • Zellweger syndrome |
| DMP of lysosomes | Fabry disease • Gaucher disease • Krabbe disease • Danon's disease • Mucopolysaccharidosis II • Metachromatic leukodystrophy • Mucopolysaccharidosis III • Niemann-Pick disease • Cystinosis • Tay-Sachs disease |
| Portal: Pathobiochemistry | |


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Table of hereditary metabolic disorders (DMPs)

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- Examination methods in DMP

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