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URL of this page: https://medlineplus.gov/metabolicdisorders.html

Metabolic Disorders

Metabolism is the process your body uses to get or make energy from the food you eat. Food is made up of proteins, carbohydrates, and fats. Chemicals in your digestive system break the food parts down into sugars and acids, your body's fuel. Your body can use this fuel right away, or it can store the energy in your body tissues, such as your liver, muscles, and body fat.

A metabolic disorder occurs when abnormal chemical reactions in your body disrupt this process. When this happens, you might have too much of some substances or too little of other ones that you need to stay healthy. There are different groups of disorders. Some affect the breakdown of amino acids [https://medlineplus.gov/aminoacidmetabolismdisorders.html] , carbohydrates [https://medlineplus.gov/carbohydratemetabolismdisorders.html] , or lipids [https://medlineplus.gov/lipidmetabolismdisorders.html] . Another group, mitochondrial diseases [https://medlineplus.gov/mitochondrialdiseases.html] , affects the parts of the cells that produce the energy.

You can develop a metabolic disorder when some organs, such as your liver or pancreas, become diseased or do not function normally. Diabetes [https://medlineplus.gov/diabetes.html] is an example.

Start Here

- Metabolic Myopathies [https://rheumatology.org/patients/metabolic-myopathies] (American College of Rheumatology)
  Also in Spanish [https://rheumatology.org/patients/miopatias-metabolicas]
- Metabolism [https://kidshealth.org/en/parents/metabolism.html] (Nemours Foundation)
  Also in Spanish [https://kidshealth.org/es/parents/metabolism.html]
- Overview of Hereditary Metabolic Disorders [https://www.merckmanuals.com/home/children-s-health-issues/hereditary-metabolic-disorders/overview-of-hereditary-metabolic-disorders] (Merck & Co., Inc.)
  Also in Spanish [https://www.merckmanuals.com/es-us/hogar/salud-infantil/trastornos-metab%C3%B3licos-hereditarios/introducci%C3%B3n-a-los-trastornos-metab%C3%B3licos-hereditarios]

Diagnosis and Tests

- Comprehensive Metabolic Panel (CMP) [https://medlineplus.gov/lab-tests/comprehensive-metabolic-panel-cmp/]
  NIH (National Library of Medicine)
  Also in Spanish [https://medlineplus.gov/spanish/pruebas-de-laboratorio/panel-metabolico-completo-pmc/]
- Lactate Test [https://medlineplus.gov/lab-tests/lactate-test/] NIH (National Library of Medicine)
  Also in Spanish [https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-lactato/]
- Newborn Screening: MedlinePlus Health Topic [https://medlineplus.gov/newbornscreening.html]
  NIH (National Library of Medicine)
  Also in Spanish [https://medlineplus.gov/spanish/newbornscreening.html]

Specifics

- About Trimethylaminuria [https://www.genome.gov/Genetic-Disorders/Trimethylaminuria]
  NIH (National Human Genome Research Institute)

- Lesch-Nyhan Syndrome [https://rarediseases.info.nih.gov/diseases/7226/lesch-nyhan-syndrome] (Genetic and Rare Diseases Information Center)
- Overview of Urea Cycle Disorders [https://nucdf.org/about-ucd/what-is-a-ucd/] (National Urea Cycle Disorders Foundation)

Genetics

- Acatalasemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/acatalasemia] (National Library of Medicine)
- ALG1-congenital disorder of glycosylation: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/alg1-congenital-disorder-of-glycosylation] (National Library of Medicine)
- ALG12-congenital disorder of glycosylation: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/alg12-congenital-disorder-of-glycosylation] (National Library of Medicine)
- ALG6-congenital disorder of glycosylation: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/alg6-congenital-disorder-of-glycosylation] (National Library of Medicine)
- Aspartylglucosaminuria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/aspartylglucosaminuria] (National Library of Medicine)
- Beta-ureidopropionase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/beta-ureidopropionase-deficiency] (National Library of Medicine)
- Biotinidase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/biotinidase-deficiency] (National Library of Medicine)
- Carbonic anhydrase VA deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/carbonic-anhydrase-va-deficiency] (National Library of Medicine)
- Chylomicron retention disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/chylomicron-retention-disease] (National Library of Medicine)
- Citrullinemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/citrullinemia] (National Library of Medicine)
- Congenital hyperinsulinism: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/congenital-hyperinsulinism] (National Library of Medicine)
- Dihydropyrimidinase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/dihydropyrimidinase-deficiency] (National Library of Medicine)
- Dihydropyrimidine dehydrogenase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/dihydropyrimidine-dehydrogenase-deficiency] (National Library of Medicine)
- Fumarase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/fumarase-deficiency] (National Library of Medicine)
- Glutathione synthetase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glutathione-synthetase-deficiency] (National Library of Medicine)
- GM1 gangliosidosis: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/gm1-gangliosidosis] (National Library of Medicine)
- Hypophosphatasia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/hypophosphatasia] (National Library of Medicine)
- Molybdenum cofactor deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/molybdenum-cofactor-deficiency] (National Library of Medicine)
- Mucolipidosis II alpha/beta: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/mucolipidosis-ii-alpha-beta] (National Library of Medicine)
- Mucolipidosis III alpha/beta: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/mucolipidosis-iii-alpha-beta] (National Library of Medicine)
- Mucolipidosis III gamma: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/mucolipidosis-iii-gamma] (National Library of Medicine)

- Multiple sulfatase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/multiple-sulfatase-deficiency] NIH (National Library of Medicine)
- N-acetylglutamate synthase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/n-acetylglutamate-synthase-deficiency] NIH (National Library of Medicine)
- NGLY1-congenital disorder of deglycosylation: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/ngly1-congenital-disorder-of-deglycosylation] NIH (National Library of Medicine)
- Ornithine transcarbamylase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/ornithine-transcarbamylase-deficiency] NIH (National Library of Medicine)
- Phosphoribosylpyrophosphate synthetase superactivity: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/phosphoribosylpyrophosphate-synthetase-superactivity] NIH (National Library of Medicine)
- PMM2-congenital disorder of glycosylation: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/pmm2-congenital-disorder-of-glycosylation] NIH (National Library of Medicine)
- Pseudocholinesterase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/pseudocholinesterase-deficiency] NIH (National Library of Medicine)
- Short stature, hyperextensibility, hernia, ocular depression, Rieger anomaly, and teething delay: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/short-stature-hyperextensibility-hernia-ocular-depression-rieger-anomaly-and-teething-delay] NIH (National Library of Medicine)
- Sialic acid storage disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/free-sialic-acid-storage-disorder] NIH (National Library of Medicine)
- Sialidosis: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/sialidosis] NIH (National Library of Medicine)
- Sialuria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/sialuria] NIH (National Library of Medicine)
- SLC35A2-congenital disorder of glycosylation: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/slc35a2-congenital-disorder-of-glycosylation] NIH (National Library of Medicine)
- Trichothiodystrophy: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/trichothiodystrophy] NIH (National Library of Medicine)
- Trimethylaminuria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/trimethylaminuria] NIH (National Library of Medicine)

Clinical Trials

- ClinicalTrials.gov: Metabolic Diseases [https://clinicaltrials.gov/search?cond=%22Metabolic+Diseases%22&aggFilters=status:not%20rec] NIH (National Institutes of Health)
- ClinicalTrials.gov: Mucolipidoses [https://clinicaltrials.gov/search?cond=%22Mucolipidoses%22&aggFilters=status:not%20rec] NIH (National Institutes of Health)

Journal Articles

References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- Article: The role of irregular eating behaviors in metabolic dysfunction-associated steatotic liver... [https://www.ncbi.nlm.nih.gov/pubmed/40753232]
- Article: Myonectin and metabolic health: a systematic review. [https://www.ncbi.nlm.nih.gov/pubmed/40741175]
- Article: Possible Crosstalk and Alterations in Gut Bacteriome and Virome in HIV-1... [https://www.ncbi.nlm.nih.gov/pubmed/40733607]
- Metabolic Disorders -- see more articles [https://pubmed.ncbi.nlm.nih.gov/?term=%22Metabolic+Diseases%22%5Bmajr%3Anoexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bedat%5D+NOT+%28letter%5Bpt%5D+OR+case+reports%5Bpt%5D+OR+editorial%5Bpt%5D+OR+comment%5Bpt%5D%29+AND+free+full+text%5Bsb%5D+]

Find an Expert

- Genetic and Rare Diseases Information Center [https://rarediseases.info.nih.gov/] NIH
- National Institute of Diabetes and Digestive and Kidney Diseases [https://www.niddk.nih.gov] NIH

Teenagers

- Metabolism [https://kidshealth.org/en/teens/metabolism.html] (Nemours Foundation)

Patient Handouts

- Acidosis [https://medlineplus.gov/ency/article/001181.htm] (Medical Encyclopedia)
  Also in Spanish [https://medlineplus.gov/spanish/ency/article/001181.htm]
- Alkalosis [https://medlineplus.gov/ency/article/001183.htm] (Medical Encyclopedia)
  Also in Spanish [https://medlineplus.gov/spanish/ency/article/001183.htm]
- Lactic acid test [https://medlineplus.gov/ency/article/003507.htm] (Medical Encyclopedia)
  Also in Spanish [https://medlineplus.gov/spanish/ency/article/003507.htm]
- Metabolic acidosis [https://medlineplus.gov/ency/article/000335.htm] (Medical Encyclopedia)
  Also in Spanish [https://medlineplus.gov/spanish/ency/article/000335.htm]
- Metabolic neuropathies [https://medlineplus.gov/ency/article/001161.htm] (Medical Encyclopedia)
  Also in Spanish [https://medlineplus.gov/spanish/ency/article/001161.htm]
- Pseudohypoparathyroidism [https://medlineplus.gov/ency/article/000364.htm] (Medical Encyclopedia)
  Also in Spanish [https://medlineplus.gov/spanish/ency/article/000364.htm]

MEDICAL ENCYCLOPEDIA

Acidosis [https://medlineplus.gov/ency/article/001181.htm]

Alkalosis [https://medlineplus.gov/ency/article/001183.htm]

Blood gases [https://medlineplus.gov/ency/article/003855.htm]

Crigler-Najjar syndrome [https://medlineplus.gov/ency/article/001127.htm]

Dementia due to metabolic causes [https://medlineplus.gov/ency/article/000683.htm]

Hereditary urea cycle abnormality [https://medlineplus.gov/ency/article/000372.htm]

Homocystinuria [https://medlineplus.gov/ency/article/001199.htm]

Inborn errors of metabolism [https://medlineplus.gov/ency/article/002438.htm]

Lactic acid test [https://medlineplus.gov/ency/article/003507.htm]

Lesch-Nyhan syndrome [https://medlineplus.gov/ency/article/001655.htm]

Metabolic acidosis [https://medlineplus.gov/ency/article/000335.htm]

Metabolic neuropathies [https://medlineplus.gov/ency/article/001161.htm]

Pseudohypoparathyroidism [https://medlineplus.gov/ency/article/000364.htm]

Transient familial hyperbilirubinemia [https://medlineplus.gov/ency/article/001196.htm]

Urine odor [https://medlineplus.gov/ency/article/007298.htm]

Related Health Topics

Amino Acid Metabolism Disorders [https://medlineplus.gov/aminoacidmetabolismdisorders.html]

Amyloidosis [https://medlineplus.gov/amyloidosis.html]

Carbohydrate Metabolism Disorders [https://medlineplus.gov/carbohydratemetabolismdisorders.html]

G6PD Deficiency [https://medlineplus.gov/g6pddeficiency.html]

Gaucher Disease [https://medlineplus.gov/gaucherdisease.html]

Genetic Brain Disorders [https://medlineplus.gov/geneticbraindisorders.html]

Hemochromatosis [https://medlineplus.gov/hemochromatosis.html]

Lipid Metabolism Disorders [https://medlineplus.gov/lipidmetabolismdisorders.html]

Malabsorption Syndromes [https://medlineplus.gov/malabsorptionsyndromes.html]

Mitochondrial Diseases [https://medlineplus.gov/mitochondrialdiseases.html]

Phenylketonuria [https://medlineplus.gov/phenylketonuria.html]

Porphyria [https://medlineplus.gov/porphyria.html]

Rickets [https://medlineplus.gov/rickets.html]

Wilson Disease [https://medlineplus.gov/wilsondisease.html]

National Institutes of Health

The primary NIH organization for research on *Metabolic Disorders* is the National Institute of Diabetes and Digestive and Kidney Diseases [https://www.niddk.nih.gov]

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