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

Amino Acid Metabolism Disorders

Metabolism is the process your body uses to make energy from the food you eat. Food is made up of proteins, carbohydrates, and fats. Your digestive system breaks 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. If you have a metabolic disorder [https://medlineplus.gov/metabolicdisorders.html] , something goes wrong with this process.

One group of these disorders is amino acid metabolism disorders. They include phenylketonuria [https://medlineplus.gov/phenylketonuria.html] (PKU) and maple syrup urine disease. Amino acids are "building blocks" that join together to form proteins. If you have one of these disorders, your body may have trouble breaking down certain amino acids. Or there may be a problem getting the amino acids into your cells. These problems cause a buildup of harmful substances in your body. That can lead to serious, sometimes life-threatening, health problems.

These disorders are usually inherited. A baby who is born with one may not have any symptoms right away. Because the disorders can be so serious, early diagnosis and treatment are critical. Newborn babies get screened [https://medlineplus.gov/newbornscreening.html] for many of them, using blood tests.

Treatments may include special diets, medicines, and supplements. Some babies may also need additional treatments if there are complications.

Start Here

- Fatty Acid Oxidation Disorders [https://www.merckmanuals.com/home/children-s-health-issues/hereditary-metabolic-disorders/fatty-acid-oxidation-disorders] (Merck & Co., Inc.)
- Overview of Amino Acid Metabolism Disorders [https://www.merckmanuals.com/home/children-s-health-issues/hereditary-metabolic-disorders/overview-of-amino-acid-metabolism-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-del-metabolismo-de-los-amino%C3%A1cidos]

Diagnosis and Tests

- Methylmalonic Acid (MMA) Test [https://medlineplus.gov/lab-tests/methylmalonic-acid-mma-test/]
  NIH (National Library of Medicine)
  Also in Spanish [https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-acido-metilmalonico/]
- 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

- Understanding Hyperoxaluria [https://ohf.org/understanding-hyperoxaluria/] (Oxalosis and Hyperoxaluria Foundation)

Genetics

- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/3-hydroxy-3-methylglutaryl-coa-lyase-deficiency] NIH (National Library of Medicine)
- 3-methylcrotonyl-CoA carboxylase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/3-methylcrotonyl-coa-carboxylase-deficiency] NIH (National Library of Medicine)
- 3-methylglutaconyl-CoA hydratase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/3-methylglutaconyl-coa-hydratase-deficiency] NIH (National Library of Medicine)
- Alkaptonuria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/alkaptonuria] NIH (National Library of Medicine)
- Aminoacylase 1 deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/aminoacylase-1-deficiency] NIH (National Library of Medicine)
- Arginase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/arginase-deficiency] NIH (National Library of Medicine)
- Arginine:glycine amidinotransferase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/argininegIycine-amidinotransferase-deficiency] NIH (National Library of Medicine)
- Argininosuccinic aciduria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/argininosuccinic-aciduria] NIH (National Library of Medicine)
- Asparagine synthetase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/asparagine-synthetase-deficiency] NIH (National Library of Medicine)
- Beta-ketothiolase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/beta-ketothiolase-deficiency] NIH (National Library of Medicine)
- Dihydrolipoamide dehydrogenase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/dihydrolipoamide-dehydrogenase-deficiency] NIH (National Library of Medicine)
- Glutamate formiminotransferase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glutamate-formiminotransferase-deficiency] NIH (National Library of Medicine)
- Glutaric acidemia type I: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glutaric-acidemia-type-i] NIH (National Library of Medicine)
- Guanidinoacetate methyltransferase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/guanidinoacetate-methyltransferase-deficiency] NIH (National Library of Medicine)
- Hartnup disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/hartnup-disease] NIH (National Library of Medicine)
- Histidinemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/histidinemia] NIH (National Library of Medicine)
- Homocystinuria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/homocystinuria] NIH (National Library of Medicine)
- Hyperlysinemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/hyperlysinemia] NIH (National Library of Medicine)
- Hypermethioninemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/hypermethioninemia] NIH (National Library of Medicine)
- Hyperprolinemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/hyperprolinemia] NIH (National Library of Medicine)
- Isobutyryl-CoA dehydrogenase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/isobutyryl-coa-dehydrogenase-deficiency] NIH (National Library of Medicine)
- Isolated sulfite oxidase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/isolated-sulfite-oxidase-deficiency] NIH (National Library of Medicine)
- Isovaleric acidemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/isovaleric-acidemia] NIH (National Library of Medicine)

- Lesch-Nyhan syndrome: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/lesch-nyhan-syndrome] (National Library of Medicine)
- Maple syrup urine disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/maple-syrup-urine-disease] (National Library of Medicine)
- Methylmalonic acidemia with homocystinuria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/methylmalonic-acidemia-with-homocystinuria] (National Library of Medicine)
- Methylmalonic acidemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/methylmalonic-acidemia] (National Library of Medicine)
- Nonketotic hyperglycinemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/nonketotic-hyperglycinemia] (National Library of Medicine)
- Phosphoglycerate dehydrogenase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/phosphoglycerate-dehydrogenase-deficiency] (National Library of Medicine)
- Primary hyperoxaluria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/primary-hyperoxaluria/] (National Library of Medicine)
- Prolidase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/prolidase-deficiency] (National Library of Medicine)
- Short/branched chain acyl-CoA dehydrogenase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/short-branched-chain-acyl-coa-dehydrogenase-deficiency] (National Library of Medicine)
- Tyrosinemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/tyrosinemia] (National Library of Medicine)

Clinical Trials

- ClinicalTrials.gov: Amino Acid Metabolism, Inborn Errors [https://clinicaltrials.gov/search?cond=%22Amino+Acid+Metabolism,+Inborn+Errors%22&aggFilters=status:not%20rec] (National Institutes of Health)

Journal Articles

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

- Article: Usefulness of levels of 2-methylbutyrylglycine and 2-ethylhydracrylic acid in urine for... [https://www.ncbi.nlm.nih.gov/pubmed/40598537]
- Article: Molecular characterization, clinical phenotype, and neurological outcome of twelve Palestinian children... [https://www.ncbi.nlm.nih.gov/pubmed/40598206]
- Article: The value of CSF neurotransmitter monitoring in the outcome of gene... [https://www.ncbi.nlm.nih.gov/pubmed/40460520]
- Amino Acid Metabolism Disorders -- see more articles [https://pubmed.ncbi.nlm.nih.gov/?term=%22Amino+Acid+Metabolism%2C+Inborn+Errors%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

- Oxalosis and Hyperoxaluria Foundation [https://ohf.org]

Children

- Maple Syrup Urine Disease (For Parents) [https://kidshealth.org/en/parents/msud.html] (Nemours Foundation)
  Also in Spanish [https://kidshealth.org/es/parents/msud.html]

Women

- Phenylketonuria (PKU) (In Pregnancy or While Breastfeeding) [https://mothertobaby.org/fact-sheets/maternal-pku-pregnancy/] (Organization of Teratology Information Specialists)

MEDICAL ENCYCLOPEDIA

- Alkaptonuria [https://medlineplus.gov/ency/article/001200.htm]
- Amino acids [https://medlineplus.gov/ency/article/002222.htm]
- Hartnup disorder [https://medlineplus.gov/ency/article/001201.htm]
- Methylmalonic acid blood test [https://medlineplus.gov/ency/article/003565.htm]
- Methylmalonic acidemia [https://medlineplus.gov/ency/article/001162.htm]
- Plasma amino acids [https://medlineplus.gov/ency/article/003361.htm]

Related Health Topics

- Carbohydrate Metabolism Disorders [https://medlineplus.gov/carbohydratemetabolismdisorders.html]
- Dietary Proteins [https://medlineplus.gov/dietaryproteins.html]
- Lipid Metabolism Disorders [https://medlineplus.gov/lipidmetabolismdisorders.html]
- Metabolic Disorders [https://medlineplus.gov/metabolicdisorders.html]