Carbohydrate 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. Chemicals in your digestive system (enzymes) 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. If you have a metabolic disorder [https://medlineplus.gov/metabolicdisorders.html] , something goes wrong with this process.

Carbohydrate metabolism disorders are a group of metabolic disorders. Normally your enzymes break carbohydrates [https://medlineplus.gov/carbohydrates.html] down into glucose (a type of sugar). If you have one of these disorders, you may not have enough enzymes to break down the carbohydrates. Or the enzymes may not work properly. This causes a harmful amount of sugar to build up in your body. That can lead to health problems, some of which can be serious. Some of the disorders are fatal.

These disorders are inherited. Newborn babies get screened [https://medlineplus.gov/newbornscreening.html] for many of them, using blood tests. If there is a family history of one of these disorders, parents can get genetic testing [https://medlineplus.gov/genetictesting.html] to see whether they carry the gene. Other genetic tests can tell whether the fetus has the disorder or carries the gene for the disorder.

Treatments may include special diets, supplements, and medicines. Some babies may also need additional treatments, if there are complications. For some disorders, there is no cure, but treatments may help with symptoms.

Specifics

- Diabetes: MedlinePlus Health Topic [https://medlineplus.gov/diabetes.html] NIH (National Library of Medicine)
  Also in Spanish [https://medlineplus.gov/spanish/diabetes.html]
- Galactosemia [https://liverfoundation.org/liver-diseases/pediatric-liver-information-center/pediatric-liver-disease/galactosemia/] (American Liver Foundation)
  Also in Spanish [https://liverfoundation.org/es/liver-diseases/pediatric-liver-information-center/pediatric-liver-disease/galactosemia/]
- Glycogen Storage Disease Type 1 (von Gierke) [https://liverfoundation.org/liver-diseases/pediatric-liver-information-center/pediatric-liver-disease/glycogen-storage-disease-type-1-von-gierke/] (American Liver Foundation)
  Also in Spanish [https://liverfoundation.org/es/enfermedades-del-HIGADO/centro-de-informaci%C3%B3n-hep%C3%A1tica-pedi%C3%A1trica/enfermedad-hep%C3%A1tica-pedi%C3%A1trica/enfermedad-por-almacenamiento-de-gluc%C3%B3geno-tipo-1-de-von-gierke/]
- Hurler Syndrome [https://www.nmdp.org/patients/understanding-transplant/diseases-treated-by-transplant/hurler-syndrome] (National Marrow Donor Program)
- Learn about MPS and ML [https://mpssociety.org/learn-about-mps/] (National MPS Society)
- Mucopolysaccharidoses [https://www.ninds.nih.gov/health-information/disorders/mucopolysaccharidoses] NIH (National Institute of Neurological Disorders and Stroke)

Genetics

- Alpha-mannosidosis: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/alpha-mannosidosis] NIH (National Library of Medicine)
- Beta-mannosidosis: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/beta-mannosidosis] NIH (National Library of Medicine)
- Essential pentosuria: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/essential-pentosuria] NIH (National Library of Medicine)
- Fucosidosis: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/fucosidosis] NIH (National Library of Medicine)
- Galactosemia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/galactosemia] NIH (National Library of Medicine)
- Galactosialidosis: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/galactosialidosis] NIH (National Library of Medicine)
- GLUT1 deficiency syndrome: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glut1-deficiency-syndrome] NIH (National Library of Medicine)
- Glycogen storage disease type 0: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glycogen-storage-disease-type-0] NIH (National Library of Medicine)
- Glycogen storage disease type I: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glycogen-storage-disease-type-i] NIH (National Library of Medicine)
- Glycogen storage disease type III: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glycogen-storage-disease-type-iii] NIH (National Library of Medicine)
- Glycogen storage disease type IV: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glycogen-storage-disease-type-iv] NIH (National Library of Medicine)
- Glycogen storage disease type IX: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glycogen-storage-disease-type-ix] NIH (National Library of Medicine)
- Glycogen storage disease type V: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glycogen-storage-disease-type-v] NIH (National Library of Medicine)
- Glycogen storage disease type VI: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glycogen-storage-disease-type-vi] NIH (National Library of Medicine)
- Glycogen storage disease type VII: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/glycogen-storage-disease-type-vii] NIH (National Library of Medicine)
- Hereditary fructose intolerance: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/hereditary-fructose-intolerance] NIH (National Library of Medicine)
- Lactate dehydrogenase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/lactate-dehydrogenase-deficiency] NIH (National Library of Medicine)
- Mucopolysaccharidosis type I: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-i] NIH (National Library of Medicine)
- Mucopolysaccharidosis type II: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-ii] NIH (National Library of Medicine)
- Mucopolysaccharidosis type III: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-iii] NIH (National Library of Medicine)
- Mucopolysaccharidosis type IV: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-iv] NIH (National Library of Medicine)
- Mucopolysaccharidosis type VI: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/mucopolysaccharidosis-type-vi] NIH (National Library of Medicine)

- Phosphoglycerate kinase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/phosphoglycerate-kinase-deficiency] NIH (National Library of Medicine)
- Phosphoglycerate mutase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/phosphoglycerate-mutase-deficiency] NIH (National Library of Medicine)
- Pompe disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/pompe-disease] NIH (National Library of Medicine)
- Pyruvate dehydrogenase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/pyruvate-dehydrogenase-deficiency] NIH (National Library of Medicine)
- Schindler disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/alpha-n-acetylgalactosaminidase-deficiency] NIH (National Library of Medicine)

Clinical Trials

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

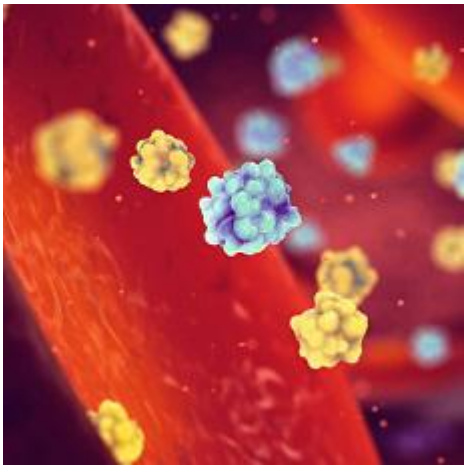
Journal Articles

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

- Article: Glut1 Deficiency Syndrome: Novel Pathomechanisms, Current Concepts, and Challenges. [https://www.ncbi.nlm.nih.gov/pubmed/40405536]
- Article: Seizure and redox rescue in a model of glucose transport deficiency. [https://www.ncbi.nlm.nih.gov/pubmed/40184423]
- Article: Clinical and genetic characteristics of glucose transporter 1 deficiency syndrome in... [https://www.ncbi.nlm.nih.gov/pubmed/40048124]
- Carbohydrate Metabolism Disorders -- see more articles [https://pubmed.ncbi.nlm.nih.gov/?term=%22Carbohydrate+Metabolism%2C+Inborn+Errors%22%5Bmajr%3Anoexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+2+Years%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+]

Children

- Sanfilippo Syndrome (For Parents) [https://kidshealth.org/en/parents/sanfilippo-syndrome.html] (Nemours Foundation)
  Also in Spanish [https://kidshealth.org/es/parents/sanfilippo-syndrome.html]



MEDICAL ENCYCLOPEDIA

Acid mucopolysaccharides [https://medlineplus.gov/ency/article/003368.htm]

Galactose-1-phospate uridyltransferase [https://medlineplus.gov/ency/article/003636.htm]

Galactosemia [https://medlineplus.gov/ency/article/000366.htm]

Hereditary fructose intolerance [https://medlineplus.gov/ency/article/000359.htm]

Hunter syndrome [https://medlineplus.gov/ency/article/001203.htm]

Mucopolysaccharidoses [https://medlineplus.gov/ency/article/001246.htm]

Mucopolysaccharidosis type I [https://medlineplus.gov/ency/article/001204.htm]

Mucopolysaccharidosis type III [https://medlineplus.gov/ency/article/001210.htm]

Mucopolysaccharidosis type IV [https://medlineplus.gov/ency/article/001206.htm]

Type V glycogen storage disease [https://medlineplus.gov/ency/article/000329.htm]

von Gierke disease [https://medlineplus.gov/ency/article/000338.htm]

Related Health Topics

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

Carbohydrates [https://medlineplus.gov/carbohydrates.html]

Lactose Intolerance [https://medlineplus.gov/lactoseintolerance.html]

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

Metabolic Disorders [https://medlineplus.gov/metabolicdisorders.html]