

Our metabolism is a complex process in which our bodies produce energy from the food and drink that we take in. This process occurs through a series of steps. Metabolism also involves the removal of waste that is produced during this process. Metabolic disorders refer to a group of genetic conditions in which there is an error somewhere in this process. Specifically, an individual is not able to produce or breakdown certain enzymes, proteins, and/or other substances needed by the body. For this reason, metabolic disorders are also called biochemical disorders or inborn errors of metabolism.

There are hundreds of different kinds of metabolic disorders. The disorders are categorized by the specific process or pathway that they affect. The primary categories include:

- Amino acid disorders
- Carbohydrate disorders
- Fatty acid oxidation disorders
- Lysosomal disorders (e.g. [Fabry disease](#))
- Mitochondrial disorders
- Organic acid disorders
- Peroxisomal disorders
- Urea cycle disorders (e.g. [OTC deficiency](#))
- Other:
  - Congenital disorders of creatine metabolism
  - Congenital disorders of glycosylation
  - Lipid metabolism disorders (e.g. [familial hypercholesterolemia](#))
  - Metal metabolism disorders (e.g. [Wilson disease](#))
  - Purine and pyrimidine metabolism disorders
  - Porphyrin metabolism disorder
  - Vitamin and cofactor metabolism disorder

Metabolic disorders may affect a single part of the body, or multiple body systems, depending on what specific pathway is involved. Individuals may have limited to no symptoms, or may present with life threatening episodes. In addition, symptoms may vary between infancy to childhood to adulthood, can be progressive, and at times life-limiting. A metabolic disorder may be suspected in people with developmental disabilities, seizures, poor growth, and periods of regression.

Diagnosis of a metabolic disorder is made through the careful evaluation of an individual's medical and family history, physical examination, and measurement of specific levels of certain substances in the blood and urine. In some cases, imaging studies, measurement of levels of certain substances in other body tissues (e.g. skin or muscle), and/or genetic

testing may be recommended.

For many metabolic disorders, there is specific management and treatment available. In general, treatment is focused on reducing and eliminating the buildup of these toxic substances in the body. This may be done through a specialized diet, taking supplements, or avoidance of triggers, and some metabolic conditions can now even be treated by replacing the missing enzymes.

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Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

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