



METABOLIC DISORDERS

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MEDICAL FOCUS

METABOLISM

Metabolism is the process used to maintain life and produce energy. For example, through metabolism, the body converts the food we eat into energy. Metabolism is the total of all the chemical processes that occur in the body each day. These chemical reactions break down food or chemicals for use or storage. Other chemical processes break down substances that are no longer needed or make substances that are lacking in the body.

METABOLISM HAS THREE MAIN PURPOSES:

1. Conversion of food to energy to power cells
2. Conversion of food/fuel to build or synthesize proteins, lipids, nucleic acids and carbohydrates
3. Elimination of nitrogenous wastes

DEFINITION

A metabolic disorder occurs when these processes do not work properly due to a hormone or enzyme deficiency. Metabolic disorders are categorized by whether a specific substance builds-up in harmful amounts, is too low or is missing.

Metabolic disorders are sometimes classified as disorders of carbohydrate, amino acid or organic acid metabolism or lysosomal storage diseases. More recently, other classifications have emerged including disorders of the urea cycle, fatty acid oxidation, mitochondrial function and peroxisomal function, as well as disorders of mitochondrial, porphyrin, purine, pyrimidine or steroid metabolism. Many metabolic disorders do not fall into any of these categories.

Examples of metabolic disorders include glycogen storage disease, phenylketonuria, glutaric acidemia, porphyria, Lesch-Nyhan syndrome and Zellweger syndrome.

ETIOLOGY

Metabolic disorders are caused by inherited genetic defects, typically from both parents. There are hundreds of inherited metabolic disorders. Some metabolic disorders are part of newborn routine screening, such of phenylketonuria (PKU), and others are identified once symptoms emerge. One study estimated that metabolic disorders occur in 1 of 1,400 births.

TREATMENT

Treatment varies by the specific metabolic disorder and may include limiting the diet to avoid chemicals that an individual cannot appropriately metabolize, supplementing the diet to compensate for the metabolic issue, or replacing missing enzymes. If the metabolic disorder has led to other medical conditions, each condition will also require treatment.

PROGNOSIS

Prognosis also varies by specific metabolic disorder; however, some disorders can lead to significant medical complications and functional deficits. In some cases, seating and wheeled mobility, as well as other assistive technologies, may be indicated.

Health care professionals treating a patient with a metabolic disorder can often find detailed information about the specific disorder through credible internet sites. The patient and caregivers are frequently very knowledgeable about the condition, as well, and can be an invaluable source of information.

CONTACT THE AUTHOR

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REFERENCES

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