

# METABOLIC DISORDERS

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## SYMPTOMS OR BEHAVIORS

Metabolic disorders do not have unique symptoms, but rather may affect various organ systems of the body.

- Neurological/cognitive impairment
- Growth impairment
- Loss of muscle control
- Muscle weakness and pain
- Gastro-intestinal disorders
- Swallowing difficulties
- Cardiac disease
- Liver disease
- Diabetes
- Respiratory complications
- Seizures
- Visual/hearing problems
- Lactic acidosis
- Migraines
- Stroke
- Susceptibility to infection
- Failure to gain weight
- Fatigue



## ABOUT THE DISORDER

Metabolism is the sum of all the chemical reactions involved in the continual process of breakdown and renewal in the body. Metabolic processes result in growth, produce energy, eliminate wastes, and control other body functions which distribute nutrients in the blood after food is digested. A metabolic disorder is anything that interferes with how food is built up or broken down in the body to keep it healthy.

There are hundreds of different metabolic diseases; the most common including phenylketonuria (PKU), mitochondrial disease, maple syrup disease, and organic disorders. Each disease is a block in the body's ability to change one chemical to another. The impact on the person with one of these defects varies widely. There may be no symptoms at all, or there may be significant effects such as neurological impairment, cognitive impairment, and impaired growth.

Mitochondrial disease results when the mitochondria, the part of the cell that produces energy, is defective. Diseases of the mitochondria appear to cause the most damage to the cells of the brain, heart, liver, skeletal muscles, kidney, and the endocrine and respiratory systems. Metabolic disorders cannot be cured; however many effects can be prevented if treatment is begun in infancy.

Metabolic disorders are rare, and even the most prevalent, PKU, affects fewer than 1 in 12,000 individuals. Most metabolic disorders are inherited as autosomal recessive traits. This means that two copies (one from each parent) of the defective gene are needed for the disorder to be expressed. These genes are non-sex chromosomes, meaning that males and females are affected equally.

Most metabolic disorders can be diagnosed using a blood test or an examination of a tissue sample to determine whether a specific enzyme is deficient or missing. The PKU screening for newborn infants, in which a blood sample is collected from the baby's heel, is now routine in all states. When a baby's screening shows signs of a metabolic disorder, additional tests are performed to confirm and classify the disorder, since treatment is extremely specific.

Treatment depends on the diagnosis, and may consist of a special diet; megadoses of a vitamin, an "orphan" drug; or a combination of these approaches. The most frequent treatment is a special diet which is low in the offending chemical. Often diet is the sole treatment, and, when begun in infancy, may mean the difference between normal functioning and cognitive deficiency. In all disorders, consistent dietary intervention is critical for normal physical and cognitive development. With controlled diet, children with metabolic disorders may lead normal and healthy lives.

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## EDUCATIONAL IMPLICATIONS

In many instances metabolic disorders affect cognitive ability. School personnel should have resources about the effects of these disorders on the student's individual needs. The student may need support in dealing with self-image and adjustment problems as well as acceptance by peers. In instances of growth/physical limitations, a modified physical education program may be beneficial as well as environment adaptations and/or adaptive equipment.

## INSTRUCTIONAL STRATEGIES AND CLASSROOM ACCOMMODATIONS

- Curriculum modifications (extra time for assignments, every other problem, no time limits, alternative ways to test and get information, and support in resource room)
- Depending on cognitive ability, special education instruction and paraprofessional support may be necessary.
- Growth reduction and delayed puberty may create emotional/social problems. It may be necessary to include counseling (school counselor, social worker, or school psychologist) in the student's educational plan.
- An Individual Health Care Plan may be developed by the school nurse to meet the health needs of the student.

## RESOURCES

Metabolic Information Network  
P.O. Box 670847  
Dallas, TX 75367-0847  
(214) 696-2188

Association for Neuro-metabolic Disorders  
5223 Brockfield Lane  
Sylvania, OH 43560-1809  
(419) 885-1497  
volk4olks@aol.com

National Phenylketonuria Foundation  
6301 Tejas Drive  
Pasadena, TX 77503  
(713) 487-4802

Children's PKU Network (CPN)  
1520 State St., Suite #240  
San Diego, CA 92101  
(619) 233-3202  
[pkunetwork@aol.com](mailto:pkunetwork@aol.com)  
Resources including video and pamphlet for children

PKU Parents  
8 Myrtle Lane  
San Anselmo CA 94960  
(415) 457-4632

The United Mitochondrial Disease Foundation  
8085 Saltsburg Road, Suite 201  
Pittsburg, PA 15239  
(412) 793-8077

CLIMB Theater  
6415 Carmen Ave E  
Inver Grove Heights, MN 55076  
(800) 767-9600  
[mail@climb.org](mailto:mail@climb.org)  
[www.climb.org](http://www.climb.org)

School Nurse's Source Book of Individualized Health Care Plans – Volume II  
Sunrise River Press  
c. 2005  
[www.schoolnursebooks.com](http://www.schoolnursebooks.com)