



## **HCS or MCD**

### **Holocarboxylase Synthetase Deficiency**

### **Multiple Carboxylase Deficiency – Neonatal (MCD)**

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#### **What is it?**

HCS or MCD stands for holocarboxylase synthetase deficiency. It is one type of organic acid disorder. People with HCS or MCD have problems changing protein and carbohydrates from food into energy for the body.

#### **What causes it?**

HCS or MCD occurs when an enzyme, called holocarboxylase synthetase (HCS), is either missing or not working properly. This enzyme's job is to add a vitamin called biotin to other enzymes called carboxylases so that they can change the food we eat into energy for the body. When the HCS enzyme is not working, certain harmful substances build up in the blood and urine. This can cause serious health problems.

#### **If HCS or MCD is not treated, what problems occur?**

Each child with HCS or MCD is likely to have slightly different effects. Many babies with this condition start to have symptoms within hours of birth or during the first few days or weeks of life. Other babies have their first symptoms sometime in infancy, usually before age 2.

A small number of people with HCS or MCD never show symptoms and are found to be affected only after a brother or sister is diagnosed.

HCS or MCD causes episodes of illness called metabolic crises. Some of the first symptoms of a metabolic crisis are:

- 1) Poor appetite.
- 2) Extreme sleepiness or lack of energy.
- 3) Irritability.
- 4) Vomiting.
- 5) Low muscle tone (floppy muscles and joints).
- 6) Severe peeling skin rash.

Common lab findings are:

- 1) Low blood sugar, called hypoglycemia.
- 2) High levels of acidic substances in the blood, called metabolic acidosis.
- 3) Slightly high levels of ammonia in the blood.
- 4) Low platelets.
- 5) Ketones in the urine.
- 6) High levels of substances called organic acids in the urine.

If a metabolic crisis is not treated, a child with HCS or MCD can develop:

- 1) Breathing problems.
- 2) Seizures.
- 3) Swelling of the brain.
- 4) Coma, sometimes leading to death.

Untreated children with HCS or MCD often have other symptoms, whether or not they have metabolic crises. These can include:

- 1) Skin rashes or skin infections.
- 2) Hair loss.
- 3) Learning disabilities or mental retardation.
- 4) Delays in walking and motor skills.
- 5) Problems coordinating movements, called ataxia.
- 6) Rigid muscle tone, called spasticity.

- 7) Problems coordinating movements, called ataxia.
- 8) Rigid muscle tone, called spasticity.
- 9) Poor growth.
- 10) Seizures.
- 11) Hearing loss.
- 12) Vision loss.

Without treatment, brain damage can occur. This can result in mental retardation. If left untreated, many babies with HCSD die.

### **What is the treatment for HCSD?**

Your baby's primary doctor will work with a metabolic doctor to provide care for your child.

The main treatment for HCSD is a type of B vitamin called biotin. In babies found to have HCSD through newborn screening, biotin treatment can prevent symptoms from occurring. It can also reverse some of the health problems in children who already have shown symptoms. You will need a prescription from your doctor in order to purchase the amount of biotin your child will need.

Prompt treatment with biotin is needed to prevent mental retardation and serious medical problems. You should start the treatment as soon as you know your child has HCSD. Your child will need to take biotin by mouth on a daily basis throughout life.

Biotin is usually the only medication needed to treat HCSD. Your child will not need to restrict any activities or change his or her diet.

### **What happens when HCSD is treated?**

Babies who receive prompt and ongoing treatment with biotin before they have a metabolic crisis are expected to have normal growth and development.

Even with treatment, a few children have developed lifelong learning problems or mental retardation. In children who have already shown delays in learning or loss of hearing or

eyesight treatment can prevent additional effects. But, it may not be able to correct the effects that are already present.

### **What causes the HCS enzyme to be absent or not working correctly?**

Genes tell the body to make various enzymes. People with HCSD have a pair of genes that do not work correctly. Because of these gene changes, the HCS enzyme does not work properly or is not made at all.

### **Is HCSD inherited?**

HCSD is inherited and affects both boys and girls equally.

Everyone has a pair of genes that make the HCS enzyme. In children with HCSD, neither of these genes works correctly. These children inherit one nonworking gene for the condition from each parent.

Parents of children with HCSD rarely have the disorder. Instead, each parent has a single non-working gene for HCSD. They are called carriers. Carriers do not have HCSD because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25 percent chance in each pregnancy for the child to have HCSD. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

### **Can other members of the family have HCSD or be carriers?**

#### ***Having HCSD***

The brothers and sisters of a baby with HCSD have a small chance of being affected, even if they haven't had symptoms. Finding out whether other children in the family have this condition is important because early treatment may prevent serious health problems. Talk with your metabolic doctor or genetic counselor about testing your other children.

### ***HCS D Carriers***

Brothers and sisters who do not have HCS D still have a chance to be carriers like their parents. Except in special cases, carrier testing should be done only in people older than 18.

Each of the parents' brothers and sisters has a 50 percent chance to be an HCS D carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with HCS D.

When both parents are HCS D carriers, newborn screening results are not sufficient to rule out the condition in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

### **Can other family members be tested?**

#### ***Diagnostic Testing***

Diagnostic testing on blood or skin samples can be done for brothers or sisters of a child with HCS D. Talk to your doctor or genetic counselor if you have questions about testing for HCS D.

#### ***Carrier Testing***

Carrier testing for HCS D may be available. If you have questions about carrier testing, ask your genetic counselor or metabolic doctor.

### **How many people have HCS D?**

About one in 87,000 babies in the United States is born with HCS D.

### **Does HCS D happen more frequently in a certain ethnic group?**

No, HCS D does not happen more often in any specific race, ethnic group, geographical area or country.

### **Does HCS D go by any other names?**

HCS D is sometimes also called:

- 1) Holocarboxylase deficiency.
- 2) HLCS deficiency.
- 3) Multiple carboxylase deficiency, early-onset.

- 4) Infant multiple carboxylase deficiency.
- 5) MCD, neonatal form.

### **Where can I find more information?**

Organic Acidemia Association  
[www.oaanews.org](http://www.oaanews.org)

Children Living with Inherited Metabolic Diseases (CLIMB)  
[www.climb.org.uk](http://www.climb.org.uk)

Save Babies Through Screening Foundation  
[www.savebabies.org](http://www.savebabies.org)

Genetic Alliance  
[www.geneticalliance.org](http://www.geneticalliance.org)

### **Children's Special Health Services (CSHS)**

State Capitol Judicial Wing  
600 E. Boulevard Ave., Department 301  
Bismarck, ND 58505-0269  
Toll Free: 800.755.2714  
701.328.2436  
Relay TDD: 701.328.3975  
CSHS website: [www.ndhealth.gov/CSHS](http://www.ndhealth.gov/CSHS)  
North Dakota Department of Health website:  
[www.ndhealth.gov](http://www.ndhealth.gov)

### **Family support resources available from CSHS:**

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

### ***Family Resources***

Family to Family Network  
Center for Rural Health  
University of North Dakota  
School of Medicine and Health Sciences  
P.O. Box 9037  
Grand Forks, ND 58202-9037  
Toll Free: 888.434.7436  
701.777.2359  
Fax: 701.777.2353  
E-mail: [NDF2F@medicine.nodak.edu](mailto:NDF2F@medicine.nodak.edu)  
[www.medicine.nodak.edu/crh](http://www.medicine.nodak.edu/crh)

Pathfinder Services of ND  
Pathfinder Family Center  
1600 2<sup>nd</sup> Ave. SW, Ste. 19  
Minot, ND 58701  
Toll Free: 800.245.5840  
701.837.7500  
Relay TDD: 701.837.7501  
E-mail: [ndpath01@ndak.net](mailto:ndpath01@ndak.net)  
[www.pathfinder.minot.com](http://www.pathfinder.minot.com)



North Dakota Department of Health  
Newborn Screening Program  
600 E. Boulevard Ave., Dept. 301  
Bismarck, ND 58505-0200  
800.472.2286 or 701.328.2493  
[www.ndhealth.gov/familyhealth](http://www.ndhealth.gov/familyhealth)

Family Voices of North Dakota, Inc.  
P.O. Box 163  
Edgeley, ND 58433  
Toll Free: 888.522.9654  
701.493.2634  
Fax: 701.493.2635  
[www.geocities.com/ndfv](http://www.geocities.com/ndfv)

\*\*This fact sheet has general information. Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be recommended for some children but not others. All children should be followed by a metabolic doctor in addition to their primary-care provider.

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### **Disclaimer**

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