MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY: MCAD

A Guide for Parents

Sponsored by the
Pacific Northwest
Regional Genetics Group (PacNoRGG)

This booklet contains general information about medium chain acyl CoA dehydrogenase deficiency (MCAD).

As every child with this disorder is different, the information in this booklet may not apply to your child specifically. Feel free to ask your child's health provider any questions you may have about the enclosed information.

Please share this booklet with anyone who cares for your child, such as health providers, nutritionists, and other health care workers, school professionals, childcare providers, and members of your family.

For your reference, a glossary of medical terms is included in the back of the booklet.

Medium Chain ACYL-CoA Dehydrogenase Deficiency: MCAD

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD: pronounced me-dee-yum chain a-sil-ko-a dee-hi-dro-gen-aze) is an inherited disorder. It is estimated that 1 in 5,000-12,000 infants are born with this disorder.

Children with MCAD have an inactive enzyme called medium-chain acyl-Coenzyme A dehydrogenase. Enzymes are substances in the body which enable chemical reactions to occur. The enzyme involved in MCAD deficiency normally breaks down fats from food we eat, as well as fats already stored in the body. (See Diagram 1.) In children with MCAD, this enzyme does not work very well.

Normally, when a person eats fats they are broken down and used for energy. Since children with MCAD lack the enzyme to break down fats completely, the body must use glucose as an alternate energy source. Glucose is a type of sugar in the blood, and although it is a good source of energy, there is a limited supply available. Once the glucose in the body has been used up, hypoglycemia (low blood sugar) can occur.

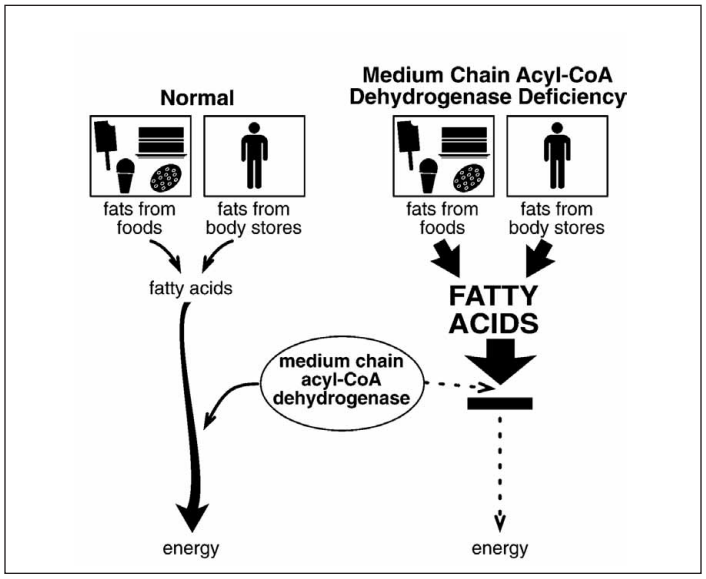
When a person fasts (doesn't eat for a while), the body normally uses stored fat for energy. Since children with MCAD are unable to change stored fat into energy the body can use, the body chooses glucose again as an alternative energy source.

Children with MCAD must avoid periods of fasting and seek immediate medical treatment if such episodes occur (such as during illness). Treatment for MCAD also includes dietary modification and supplementation.

Fats from foods or body stores are normally broken down to become sources of energy. Because the enzyme called medium chain acyl-CoA dehydrogenase does not work properly, this breakdown process does not occur.

Diagram 1.

Metabolic defect in medium chain acyl-CoA dehydrogenase deficiency (MCAD).



Symptoms of MCAD

Many children with MCAD are not diagnosed for weeks, months, or even years. The initial symptoms experienced are variable. However, initial symptoms usually occur during infancy or early childhood, and often the first symptomatic episode occurs after a period of fasting.

Symptoms may include extreme tiredness, skin clamminess, behavior changes, irritability, fever, diarrhea, and vomiting. The affected person may or may not have low blood sugar. If untreated, coma and even death could occur. People with MCAD are normal between episodes of crisis, which are often provoked by periods of fasting without carbohydrate/glucose supplementation. With proper treatment, children with MCAD can expect to live typical, healthy lives, with typical growth and development.

It is very important to inform everyone involved in your child's medical care that he/she has MCAD, and that he/she receives prompt medical attention during periods of fasting.

Treatment

There are four parts to successful treatment of MCAD:

1. Avoidance of fasting for more than 4-6 hours.

When an infant or young child does not eat for more than 4-6 hours, his or her body naturally turns to stored fats for energy. Your child with MCAD lacks the enzyme which converts stored fats into energy. Avoiding fasting has proven to be an effective means of treating MCAD.

If your child with MCAD deficiency evenly spaces meals to avoid long periods of fasting, the body will have plenty of energy from food, and will not have to rely on other sources of stored energy. Ask your health provider what is appropriate for your child.

2. A reduced fat, complex carbohydrate food pattern.

It should be noted that this is not a fat-free food pattern, but simply low in fats. The body needs small amounts of fat to function properly, and fats should never be completely eliminated from the food pattern.

Carbohydrates are the best possible source of energy. They contain many forms of sugar that the body can break down into fuel. If the body can use sugar from food for energy, it won't have to use its precious supply of stored glucose. This food pattern will be discussed more completely later in this booklet.

3. Supplemental Carnitine.

Carnitine is a safe, natural chemical which helps the body produce muscle energy, and also helps remove some breakdown products of fat that children with MCAD cannot process.

Children with MCAD may be lacking carnitine. Its use varies with the needs of each individual child. Ask your health provider if carnitine is appropriate for your child.

4. Immediate contact with your child's health provider when illness occurs.

Every child gets sick from time to time. It may be the flu, a cold, an infection, or something more severe. Regardless of the illness, the body uses extra energy in an effort to heal itself. Normally, the body will turn to stored fats for that energy. Again, children with MCAD are unable to use stored fats. Risks after illness and/or fasting include hospitalization, long-term disability and death.

Always call your child's health provider when your child is vomiting, has diarrhea, has an infection, or has a fever of more than 101 degrees Fahrenheit.

Nutrition and Dietary Guidelines

A food pattern with reduced fat intake is recommended for a child with MCAD. Foods that are high in complex carbohydrates are good sources of energy for people with MCAD. Following is a list of high carbohydrate foods that can be eaten freely. Many other foods may be added to the list. Ask your nutritionist for ideas.

A. Foods High in Carbohydrates:

Cereals and Grains:

cold cereals
hot cereals
bagels
breads
pasta noodles
croutons
English muffins
rice
rolls
buns
pita bread
fat-free products
tortillas
animal crackers
graham crackers
lentils
crackers
popcorn
corn
potatoes
yams
sweet potatoes

Vegetables:

carrots
broccoli
cauliflower
cabbage
vegetable juices
onions
mushrooms
lettuce
spinach
radishes
squash
greens
beans
peas
asparagus

Fruits:

apples
oranges
bananas
apricots
grapes
berries
cherries
peaches
pears
raisins
fruit juices
kiwi
pineapple
tomatoes

B. Foods High in Fat:

butter	high fat meats
margarine	hot dogs
oils	sausage
cheese	bacon
whole milk	luncheon meats
whole milk products	fried foods
cream	nuts, seeds and their products
ice cream	peanut butter
sour cream	

Your nutritionist can help you create a food pattern specifically geared toward your child's needs for growth and development. The nutritionist can also introduce you to a wider variety of low fat foods. Because your child will be limiting certain foods, the food pattern may not always have enough vitamins and minerals to meet your child's growing needs. **A general multivitamin which also includes calcium and iron is essential.**

You may have many questions regarding the amounts of fat in each food, and the following book may be a good reference to have. Your nutritionist should be able to tell you how it can be purchased.

Bowes & Church's
Food Values of Portions
Commonly Used, 17th ed.
by Jean AT Pennington, JB Lippincott

Length of Treatment and Medical Visits

Medium-chain acyl-CoA dehydrogenase deficiency does not go away. Treatment will be required throughout life, although slight changes may be needed as growth and development needs change.

Some children with MCAD remain well and free of illness for years. This does not mean the disorder has gone away. These children still have MCAD, and there is still a risk of a serious and potentially fatal illness if treatment is not followed.

Because your child with MCAD has a lifelong condition that could harm growth and development, your child should be closely followed by a health provider and a nutritionist.

Medical visits offer many advantages for your child, including developmental, physical, nutritional, and neurological assessments. The medical team will want to learn how your child is getting along with parents, siblings, and friends, and work with you to solve problems. The goal is to help your child develop skills needed to take responsibility for managing his or her own condition. The staff knows this is not an easy task, and wants to provide as much support as they can to you and your family. Most importantly, these visits offer you an opportunity to ask questions and get answers.

At a medical visit, you and your child can expect any of the following:

What to expect:	How it will help:
Discussion of medical history since last appointment	To determine if treatment is working, and to see if changes are needed; To talk about concerns at home, at school, with friends and/or with caregivers
Physical exam	To look at neurological status and other measures of physical well-being
Record of height, weight, and head size	To monitor child's growth and treatment
Food records	To look at food choices, assess the amounts of fats and proteins eaten, and adjust as needed
Developmental exam	To assess child's learning and development
Blood draw	To measure levels of amino acids and other compounds in the blood

Organizing Your Information

You may want to buy a 3-ring notebook binder with tab dividers to record information, questions, and food patterns. Here you can collect treatment plans, growth and medication records, questions, articles, food lists, recipes, and other information that may be useful to you. One section might hold food records, and another a graph of your child's growth and development. Make a list of questions as you think of them, so you'll remember them at your next medical visit.

Social Concerns

All family members play a very important role in your child's treatment. Other children in your family, as well as the child with MCAD, should be taught about the special high carbohydrate, low-fat food pattern. Encourage all family members, including brothers and sisters, to help choose and prepare foods low in fat and high in carbohydrates. Explain MCAD to everyone who will participate in the care of your child (relatives, teachers, day care providers, friends, baby-sitters, and others). They must understand the importance of the strict food pattern and become familiar with foods allowed and not allowed. Be sure to emphasize the importance of the special food pattern for normal growth and development. Also teach siblings and relatives not to feel sorry for your child with the disorder because he or she cannot eat certain foods.

Treat your child with MCAD as normally as possible. Despite efforts to make your affected child feel good about himself or herself, there may be a time when your child becomes aware of his or her uniqueness and simply wants to be like everyone else. Be sure to help your child celebrate his or her individuality and realize that every person is different in some way.

Genetics

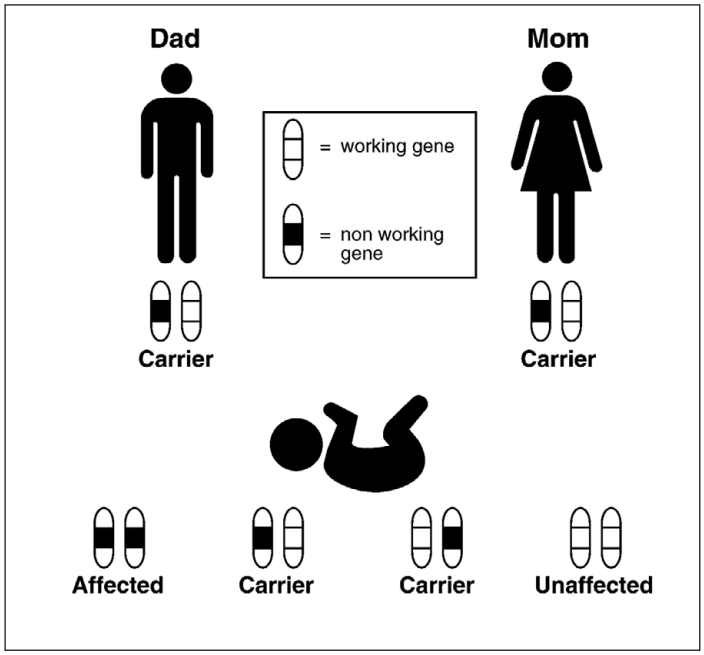
Within each child there are two copies of every gene; one copy from the mother and the other copy from the father. Most often, genes work normally. Sometimes however, a gene is changed from its original form. This is called a mutation. Mutations usually cause genes not to work correctly.

The gene change that causes MCAD is inherited in what is called an autosomal recessive pattern. This means that one copy of the changed, or non-working, gene must be inherited from each parent for a child to be affected with the disorder. The parents' health is not affected because their other copy of the gene is working correctly. Therefore, each parent is called a gene "carrier." It is important to remember that all people carry several of these "hidden" recessive genes. Also, it is no one's fault that your child was born with MCAD deficiency. Both boys and girls can have the disorder.

The chance that two parents who carry the same changed gene will have a child with the disorder is one in four, or 25% with each pregnancy. The chance these parents will have a child that is healthy but a carrier is one in two, or 50% with each pregnancy. There is also a one in four, or 25% chance that these parents will have a child who is neither affected nor a carrier. (See Diagram 2.)

For parents of affected children and affected individuals planning to have children, a consultation with a genetic counselor is recommended. The genetic counselor can explain the chances and risks of any future children being affected with the disorder, and can also explain options for testing.

Diagram 2.
Autosomal recessive inheritance



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Resources

Following is a list of agencies that may be helpful to you. Each agency specializes in different areas, such as health care, physical or mental development, support groups, or general information. Since each child is affected differently, not all agencies may be useful to you.

NATIONAL

Genetic Alliance, Inc.
4301 Connecticut Ave NW Ste 404
Washington DC 20008
202/966-5557; 800/336-GENE (4363)
info@geneticalliance.org
http://www.geneticalliance.org

Metabolic Information Network
PO Box 670847
Dallas TX 75367-0847
214/696-2188; 800/945-2188
Fax: 214/696-3258
mizesg@ix.netcom.com

MUMS: National Parent-to-Parent Network
150 Custer Court
Green Bay Wisconsin 54301-1243
920/336-5333
Fax: 920/339-0995
mums@netnet.net
http://www.netnet.net/mums

National Center for Learning Disabilities
381 Park Ave S Ste 1401
New York NY 10016
212/545-7510; 888/575-7373
Fax: 212/545-9665
http://www.ncld.org

National FOD (Fatty Oxidation Disorders) Family Support Group
805 Montrose Drive
Greensboro NC 27410
336-547-8682
goulddan@aol.com
http://www.fodsupport.org

National Parent Network on Disabilities (NPND)
1130 - 17th Street NW Ste 400
Washington DC 20036
202/463-2299
Fax: 202/463-9405
npnd@mindspring.com
http://www.npnd.org

National Society of Genetic Counselors
233 Canterbury Drive
Wallingford PA 19086-6617
610/872-7608
nsgc@aol.com
http://www.nsgc.org

NORD: National Organization for Rare Disorders
PO Box 8923
New Fairfield CT 06812
203/746-6518; 800/999-6673
Fax: 203/746-6481
orphan@rarediseases.org
http://www.rarediseases.org

Washington State Parent-to-Parent Program
4738 - 172nd Court SE
Bellevue WA 98006
425/641-7504
800/821-5927
statep2p@earthlink.net
http://www.arcwa.org

ALASKA

PARENTS: Parents as Resources Engaged in Networking and Training
4743 Northern Lights
Anchorage AK 99508
907/337-7678
Fax: 907/337-7671
parents@parentsinc.org
http://www.parentsinc.org/

IDAHO

Idaho Parents Unlimited
4696 Overland Road Ste 568
Boise ID 83705
208/342-5884; 800/242-4785 (ID only)
Fax: 208/342-1408
ipul@rmci.net
http://home.rmci.net/ipul

MONTANA

PLUK: Parents, Let's Unite for Kids
516 N 32nd Street
Billings MT 59101
406/255-0540; 800/222-7585
Fax: 406/255-0523
plukinfo@pluk.org
http://www.pluk.org

OREGON

Coalition in Oregon for Parent Education (COPE)
999 Locust Street NE
Salem OR 97303
503/581-8156; 888/505-COPE (2673)
Fax: 503/391-0429
orcope@open.org
http://www.open.org/~orcope/index.htm

WASHINGTON

Washington PAVE
6316 S 12th
Tacoma WA 98465
253/565-2266; 800/572-7368 (WA only)
Fax: 253/566-8052
wapave9@washingtonpave.com
http://www.washingtonpave.org

Treatment Plan

Prescribed food pattern:

Vitamin and/or mineral supplement: _____

Specific foods to be avoided:

Medication _____ Dose _____ Schedule _____

_____ _____ _____

Important names and phone numbers:

Health Care Provider:

Nutritionist:

Hospital:

Genetic Counselor:

To Schedule Clinic Appointments:

Public Health Nurse:

Remember

Raising a child with a rare metabolic disorder can be challenging and often confusing. Your health care providers are there to help you, and can answer the questions you will have along the way. Please do not hesitate to call upon them as you make the many changes necessary for successful treatment of your child's disorder.

Glossary

Carbohydrate - the primary fuel for the body

Carrier - a person who carries one non-working (mutated) gene in a pair of genes. Carriers do not have the disorder, they simply carry one gene for it.

Carnitine - a non-toxic, natural chemical which helps to decrease the harmful by-products of normal protein breakdown.

Enzyme - a substance in the body that enables chemical reactions to occur.

Food pattern - another term for diet. A food pattern consists of foods and beverages to be included or avoided on a daily basis.

Gene - the smallest unit of hereditary material.

Genetics - the study of heredity.

Glucose - a type of sugar found in the blood and used by the body for energy.

Hypoglycemia - low blood sugar.

Mutation - occurs when a gene is changed from its original form.

Scientific References:

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Revised 2002

Project Director: Kerry Silvey, MA, CGC

Editor: Shelly Queneau Bosworth, MS, CGC

This project was partially funded by project #6 H46 MC 00091-15 SI of the Maternal and Child Health Bureau, Department of Health and Human Services. An equal opportunity affirmative action institution.

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Design & Production: Northwest Media Inc.