Winnipeg Regional Health Authority / Office régional de la santé de Winnipeg
Caring for Health / À l'écoute de notre santé

Program in Genetics and Metabolism
Newborn Screen Metabolic Follow-up
Winnipeg, Manitoba
204-787-2494

My Baby Has a Positive Newborn Screening Result:
Medium Chain Acyl-Co-A Dehydrogenase Deficiency

What is newborn screening?
These are routine tests done shortly after birth on every baby born in Manitoba. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, including medium chain acyl-Co-A dehydrogenase deficiency (MCADD).

What does it mean if my baby has a positive newborn screening test for MCADD?
This result does **not** mean that your baby has MCADD. It means that more testing is needed because your baby **might** have MCADD. Babies with MCADD can grow and develop normally if MCADD is diagnosed early, so it is important to have follow-up testing done quickly to find out if your baby has MCADD.

You may feel worried about your baby's screen positive result. Many parents in this situation feel this way. Remember, we do not know for sure that your baby has MCADD until more testing has been done.

What is MCADD?
MCADD is an inherited (genetic) disease that causes a baby to have problems using fat as an energy source. The human body needs energy to perform all of its daily activities. The body's main source of energy is a type of sugar called glucose. If a baby has not eaten for a while – such as when they are sick or if they miss a meal – the body has used up all its glucose stores and switches to using fat as an energy source.

When a baby has MCADD, his/her body can not switch to using fat for energy. Babies with MCADD can get very sick if their bodies are not able to use to use fat for energy when it is needed. However, some babies with MCADD never develop any health problems associated with MCADD; however, there is no way to know who will develop problems caused by MCADD and who will not.

MCADD affects about 1 in every 10 000 babies born in Manitoba.

What causes MCADD?
MCADD happens when an enzyme called medium chain acyl-co-A dehydrogenase is either missing or not working properly. This enzyme's job is to break down the "medium" sized fats that we eat, or that are stored in the body, into smaller sized fats and eventually into energy.

How do I find out if my baby has MCADD?
Blood and urine tests are done to find out if a baby who screened positive actually has MCADD.

Children's Hospital Health Sciences Centre Winnipeg

This parent information brochure was developed using materials from and with the assistance of the Ontario Newborn Screening Program, located at the Children's Hospital of Eastern Ontario

University of Manitoba

When can my baby have these tests?
Your baby's doctor or a health care professional at the Winnipeg Children's Hospital will call you to talk about the results of your baby's positive newborn screen and arrange more testing as soon as possible.

Why screen for MCADD?
Babies with MCADD appear perfectly healthy at birth. But, they are at risk for a metabolic crisis during periods of not eating (for example, illness). A metabolic crisis is a serious health condition caused by low blood sugar and/or the build-up of harmful substances in the blood. Symptoms of a metabolic crisis are poor feeding, vomiting, lethargy, excessive sleepiness and irritability. If a metabolic crisis is not treated, breathing problems, seizures, coma, and sometimes death can occur. The goal of screening for MCADD is to identify an affected baby at birth before symptoms appear and with early treatment prevent a metabolic crisis from occurring.

How is MCADD treated?
Babies with MCADD are treated and monitored by a team of specialists including a metabolic doctor, a metabolic genetic counselor and a dietician. The treatment for MCADD is very simple: frequent feeding and avoiding fasting (in infancy this general means 6 to 8 hours). Sometimes, carnitine or cornstarch is given orally. With early diagnosis and careful management, children with MCADD can live healthy lives with normal growth and development.

How does a baby get MCADD?
MCADD is an inherited (genetic) disease. A baby with MCADD inherited two non-working copies of the MCADD gene, one from from each parent. People who have one non-working copy of the MCADD gene are called "carriers." Carriers of MCADD are healthy and do not have symptoms of MCADD.

Where can I get more information?
For more information on newborn screening, please talk to your local health care provider or visit the Winnipeg Metabolic Program website www.wrha.mb.ca/prog/genetics or the newborn screening section of the Cadham Provincial Laboratory website www.gov.mb.ca/health/publichealth/cpl/baby.html

For more information on MCADD, please visit the Fatty Acid Oxidation Family Support Group website at http://www.fodsupport.org

NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for medium chain acyl-co-A dehydrogenase deficiency (MCADD). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.