

## INFORMATION FOR PARENTS/GUARDIANS:

### My Baby Has a Positive Argininosuccinic Acidemia/Citrullinemia Newborn Screening Result

#### What is newborn screening?

These are routine tests done soon after birth on every baby born in Ontario. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, including two diseases called argininosuccinic acidemia (ASA) and citrullinemia.

#### What does it mean if my baby has a positive ASA/citrullinemia newborn screening result?

This result does **not** mean that your baby has ASA or citrullinemia. It means that more testing is needed because your baby **might** have ASA or citrullinemia. Babies with ASA or citrullinemia can be healthier if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has ASA or citrullinemia.

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 ASA or citrullinemia until follow up testing has been done.

#### What are argininosuccinic acidemia (ASA) and citrullinemia?

ASA and citrullinemia are two different inherited (genetic) conditions that both cause too much ammonia to build up in the body. Ammonia is a waste product normally made by the body when protein is broken down. If the body is unable to remove ammonia, it builds up and can cause serious health problems.

Some people with ASA or citrullinemia do not develop problems until childhood or adulthood. However, there is no way to know who will develop problems caused by ASA or citrullinemia early in life and who will not.

ASA affects about 1 in every 70 000 babies born in Ontario. Citrullinemia affects about 1 in every 60 000 babies in Ontario.

#### What causes ASA and citrullinemia?

ASA happens when the enzyme argininosuccinate lyase is either missing or not working properly. Citrullinemia happens when the enzyme argininosuccinic acid synthetase is either missing or not working properly. The job of these enzymes is to help the body get rid of ammonia by changing it into urea (a harmless waste product the body gets rid of in urine). If these enzymes cannot do their jobs, ammonia levels become too high and can cause serious health problems.

#### How do I find out if my baby has ASA or citrullinemia?

Blood tests and urine tests are done to find out if a baby who screened positive actually has ASA or citrullinemia.

#### When can my baby have these tests?

Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will call you to talk about the results of your baby's positive newborn screen and arrange follow up testing as soon as possible.

#### Why screen for ASA and citrullinemia?

Babies who have ASA or citrullinemia look normal when they are born but they are at risk of high levels of ammonia building up in their body. High levels of ammonia can cause excessive sleepiness, poor appetite, vomiting, low muscle tone (floppiness), and brain damage. Sometimes, high levels of ammonia can result in coma and death. The goal of screening for ASA and citrullinemia is to stop high levels of ammonia from building up in the body and help people with all types of ASA and citrullinemia live healthier lives.

#### How are ASA and citrullinemia treated?



Babies with ASA or citrullinemia are treated and monitored by a team of health care specialists including a metabolic doctor and a dietician. The treatment can include a special diet that is low in protein, a medical formula and medications. Babies with ASA or citrullinemia also have their ammonia levels, weight gain and development checked on a regular basis.

**How does a baby get ASA or citrullinemia?**

ASA and citrullinemia are inherited (genetic) diseases. A baby with ASA inherited two non-working copies of the argininosuccinate lyase enzyme gene, one copy from each parent. A baby with citrullinemia inherited two non-working copies of the argininosuccinic acid synthetase enzyme gene, one copy from each parent. People who have one non-working copy of the ASA gene or one non-working copy of the citrullinemia gene are called "carriers." Carriers of ASA and citrullinemia are healthy, do not have, and will never develop, symptoms of ASA or citrullinemia.

**Where can I get more information?**

For more information on newborn screening, please visit the Parents section of our website at [www.newbornscreening.on.ca](http://www.newbornscreening.on.ca) or talk to your local health care provider.

For more information on ASA or citrullinemia, please visit the National Urea Cycle Foundation website at <http://www.nucdf.org> or call 1 800 38 NUCDF (68233)

**NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for argininosuccinic acidemia (ASA) or citrullinemia. Please remember that this fact sheet was written for information purposes only. The fact sheet does not replace professional medical advice, diagnosis or treatment.**

