INFORMATION FOR PARENTS/GUARDIANS:
My Baby Has a Positive Congenital Adrenal Hyperplasia 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, like congenital adrenal hyperplasia (CAH).

What does it mean if my baby has a positive CAH newborn screening result?

This result does **not** mean that your baby has CAH. It means that more testing is needed because your baby **might** have CAH. Babies with CAH can grow and develop normally if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has CAH.

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 CAH until follow up testing has been done.

What is congenital adrenal hyperplasia (CAH)?

CAH is an inherited (genetic) disease that is present from birth (congenital). CAH causes the adrenal gland to be bigger than normal (hyperplasia).

The adrenal glands are small glands that sit on top of the kidneys. They make many hormones that are important for normal growth, development and function of the body.

Babies with CAH can not make enough of certain adrenal hormones. The main hormones affected by CAH are cortisol (stress-fighting hormone), aldosterone (salt-retaining hormone) and androgens (male hormones).

CAH can range from mild to serious and affects about 1 in every 15 000 babies born in Ontario.

How do I find out if my baby has CAH?

Blood tests are done to determine if a baby who screened positive actually has CAH.

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 more testing as soon as possible.

Why screen for CAH?
Adrenal hormones are essential – we can not live without them. Babies who have CAH and do not receive treatment are at risk for salt-wasting (losing salts from the body that are important for normal body function). Salt-wasting is a life-threatening event that can happen in the first few weeks of life. Screening means that babies with CAH receive early treatment and avoid a salt-wasting event. Babies diagnosed with CAH need lifelong treatment and care, but can grow and develop normally.

How is CAH treated?
Babies with CAH are treated and monitored by a specialist called an endocrinologist. Babies with CAH are given the hormones their body is unable to make, usually in the form of pills. Treatment is life long. Babies with CAH who begin treatment early can grow up to be like any other child the same age.

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

Where can I get more information?
For more information on newborn screening, please talk to your local health care provider or visit the Parents section of our website at www.newbornscreening.on.ca.

For more information on CAH, please visit the Congenital Adrenal Hyperplasia Foundation website at www.caresfoundation.org.

NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for congenital adrenal hyperplasia (CAH). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

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