Information on a Positive Newborn Screening Result for

Congenital Adrenal Hyperplasia (CAH)

MARITIME Newborn Screening / Dépistage néonatal DES MARITIMES

What is newborn screening (NBS)?

These are routine tests done soon after birth. A few drops of blood from a baby's heel are put onto a piece of absorbent paper (blotter). The blood is tested for serious but treatable conditions. These tests are done because a newborn can look healthy but have one of these conditions and need treatment. We want to find babies with these conditions so that we can start treatment to keep them as healthy as possible.

Summary:

- This result means a baby may have congenital adrenal hyperplasia (CAH). **More testing is needed**.
- **There is treatment for CAH.**
- Maritime Newborn Screening connects the baby with an **endocrinology** clinic for follow-up testing.

What are the possible results from NBS?

Most babies have a "screen negative" (normal) result. This means a baby has a decreased chance of having the conditions on newborn screening, and no follow-up is needed. Some babies (~10%) require a repeat of the newborn screen and another sample to be collected and screened. A "screen positive" result means a baby has an increased chance to have a condition from newborn screening, but it does not mean the baby has this condition. More testing is needed to give a final answer.

What does it mean if a baby has a positive screen result for Congenital Adrenal Hyperplasia (CAH)?

This result does not mean the baby has CAH. It means that more testing is needed because the baby **may** have CAH. Babies with CAH grow and develop better if treatment begins as soon after birth as possible. Follow-up testing is done quickly to find out if a baby has CAH. This is done for the baby's best possible outcome.

What happens when there is a positive newborn screen result for CAH?

A healthcare provider from Maritime Newborn Screening (MNBS) will contact the primary care provider and the family to discuss the result. MNBS will connect the baby with an **endocrinology** team. Endocrinology will arrange follow-up testing and appointments as soon as possible. Families may feel worried about the baby's newborn screen result. Many families in this situation feel this way. The healthcare teams are here to support families during the next steps.

What is CAH?

CAH is an **inherited** condition, meaning the baby is born with it (congenital). CAH is also an **endocrine** condition, meaning there is a problem with hormones. CAH affects the hormones **aldosterone**, **cortisol**, and **androgens**. This causes the adrenal glands to be bigger than normal (hyperplasia). Most cases (~95%) of CAH happen because an enzyme called **21-hydroxylase** (**21-OH**) does not work properly, which causes hormone changes.

CAH caused by 21-OH deficiency can be mild or severe. The newborn screening test is made to find babies with the severe type because they need immediate treatment. In the severe type of CAH, dehydration, salt deficiency, low blood pressure, and low blood sugar can lead to a life-threatening **adrenal crisis**. Some female babies with CAH have external genitalia that is more masculine than typical ("atypical genitalia"). In rare cases CAH is caused by other non-working enzymes. Newborn screening typically detects only the severe type of 21-OH deficient CAH, and does not detect other types of CAH.

How many babies have CAH?

CAH is a rare disease. 21-OH deficient CAH affects about 1/15,000 babies.

Why screen for CAH?

Some female babies and most male babies with CAH have no symptoms at birth. However, babies with CAH are at risk of a life-threatening **adrenal crisis** in the first few weeks of life. An adrenal crisis happens because the hormone imbalance in CAH causes dehydration and low blood sugar. Newborn screening allows us to find babies with CAH as soon as possible to start treatment and prevent an adrenal crisis.

Information on a Positive Newborn Screening Result for

Congenital Adrenal Hyperplasia (CAH)

MARITIME Newborn Screening Dépistage néonatal DES MARITIMES

How is a baby diagnosed with CAH?

Babies are seen by an endocrinologist/pediatrician for follow up testing and diagnosis. Bloodwork confirms a diagnosis of CAH. The diagnosis is made by an endocrinologist. If a baby is diagnosed with CAH, the family is also connected with a genetic counsellor from Maritime Medical Genetics Service. It can take a few days/weeks to confirm a potential diagnosis. This waiting period can be hard for families. The healthcare teams are here to support families.

How is CAH treated?

CAH treatment begins as soon as possible and is usually life-long. Children with CAH are treated and monitored by a specialty care team that includes an endocrinologist. Treatment replaces the hormones missing in CAH. With the recommended treatment and follow-up, children with CAH can grow and develop like other children. Treatment may need to be started before the confirmation testing is back to avoid an adrenal crisis. It can be stopped if the baby does not have the condition.

How does a baby get CAH?

CAH is an **inherited condition**. This means the baby is born with it. When a baby has CAH, we know that biological parents are likely carriers of the condition. Carriers do not have the condition but can have a baby with the condition if the baby inherits a non-working copy of the *CYP21A2* gene from each parent. This is autosomal recessive inheritance (see Figure 1).

Could a family have another baby with CAH?

Yes, if biological parents are carriers there is a 25% (1 in 4) chance in each pregnancy to have a child with CAH. We offer genetic counselling through **Maritime Medical Genetics Service at the IWK** to talk about the following:

- Inheritance of CAH
- Chance of CAH in future pregnancies
- Testing options for other family members

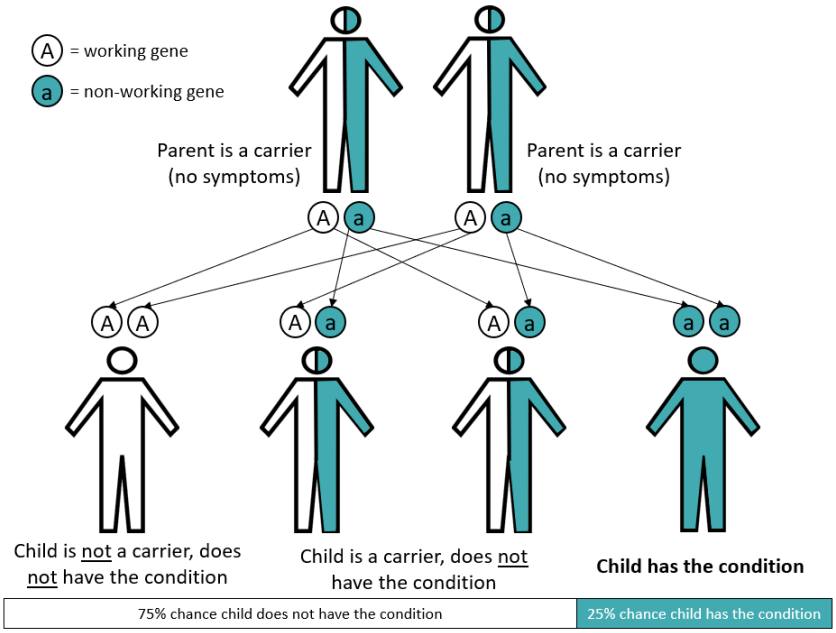


Figure 1. Autosomal recessive inheritance when both parents are carriers of congenital adrenal hyperplasia (CAH).

Where can I get more information?

- For more information on newborn screening, please visit our website at www.maritimenewbornscreening.ca or call the newborn screening genetic counsellor at 902-470-2783.
- Websites for families:
  - KidsHealth: https://kidshealth.org/en/parents/congenital-adrenal-hyperplasia.html
  - CARES Foundation: https://caresfoundation.org/
- Websites for healthcare providers:
  - MedlinePlus: https://medlineplus.gov/genetics/condition/21-hydroxylase-deficiency/
  - UpToDate: https://www.uptodate.com/contents/classic-congenital-adrenal-hyperplasia-due-to-21-hydroxylase-deficiency-in-infants-and-children-clinical-manifestations-and-diagnosis
  - GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK448098/ and https://www.ncbi.nlm.nih.gov/books/NBK1171/

NOTE TO PARENTS/GUARDIANS: *This material is provided for informational purposes and provides basic information only. This material is not intended to be and does not take the place of medical advice, diagnosis, or treatment. Please talk to your health care provider if you have any questions or concerns.*

IWK Health
Maritime Newborn Screening
IWK Health, Halifax, N.S.
Last Update: Nov 2026