Information on a Positive Newborn Screening Result for

Congenital Hypothyroidism (CH)

MARITIME Newborn Screening / Dépistage néonatal DES MARITIMES

What is newborn screening?

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 rare, 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 hypothyroidism (CH). **More testing is needed** for a final answer.
- There is treatment for CH.
- Maritime Newborn Screening connects the baby with an endocrinology/pediatric team for follow-up testing.
- If the baby has jaundice, poor feeding, low muscle tone, a puffy face, or constipation, please seek medical attention.

What are the possible results from newborn screening?

Most babies have a "screen negative" (normal) result. This means a baby has a decreased chance to have 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 tested. 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. Follow-up testing is needed to give a final answer.

What does it mean if a baby has a positive screen result for congenital hypothyroidism (CH)?

This result does not mean that the baby has congenital hypothyroidism (CH). It means that more testing is needed because the baby **may** have CH. Babies with CH grow and develop better if treatment begins before 14 days of life. Follow-up testing is done quickly to find out if a baby has CH. This is done for the baby's best possible outcome.

What happens when there is a positive newborn screen result for congenital hypothyroidism (CH)?

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 the clinical team providing the follow-up. The clinical team is either **IWK endocrinology** (Nova Scotia), or the **pediatrician/neonatologist on call** at the birth hospital (New Brunswick, Prince Edward Island). The clinical team will arrange follow-up appointments and testing to happen 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 congenital hypothyroidism (CH)?

Congenital hypothyroidism (CH) is when the thyroid is not working/underactive (hypothyroidism) and affects the baby from birth (congenital). The thyroid is a butterfly shaped organ in the neck. The thyroid normally produces hormones including thyroxine (T4) that the body and brain need to grow and develop. CH happens if the thyroid is not developed properly (missing, in the wrong location, or smaller than normal), or if the thyroid looks normal but is not working properly. In the short term, if CH is not treated it can cause jaundice, sleepiness, poor feeding, low muscle tone, a puffy face, and constipation. If CH is not treated in the long term, it can cause serious and permanent health problems like intellectual impairment, poor growth, heart problems, and hearing loss. *With treatment, children with CH are just as healthy and intelligent as their peers.*

How many babies have CH?

CH affects about 1 in 3,000 newborns. CH affects twice as many females as males, and we do not know why this happens.

Why screen for CH?

Babies with CH usually look normal at birth and may not show any health problems right away. If they are not screened by newborn screening they might not be diagnosed right away and have irreversible health problems. Our goal is to start treatment for CH before 14 days of life. Newborn screening allows us to find babies that need treatment as soon as possible.

How is a baby diagnosed with CH?

To find out if a baby has CH, they are seen by an endocrinology or pediatric/neonatology team for follow up testing. Blood tests are done to look at thyroid function. Scans are done to look at the location and shape of the thyroid. It can take a few days to confirm a diagnosis of CH. This waiting period can be hard for families. The healthcare teams are here to support families.

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How is CH treated?

CH is treated with a medication called **levothyroxine** (L-T4; Synthroid®). Levothyroxine replaces what the thyroid cannot make. Levothyroxine is a daily medication taken as a pill (adults, older children) or a pill crushed and mixed with a small amount of water or breastmilk (infants). Treatment for CH is typically life-long, but some children with CH may only take levothyroxine for the first few years of life. Children may be tested when they are 3 years old to see if treatment with levothyroxine is continued or not. This trial is monitored by an endocrinologist/pediatrician.

How does a baby get CH?

There are two main types of CH. Most cases (80-85%) of CH are caused when the **thyroid does not develop properly** during pregnancy. The thyroid may be missing, in the wrong place in the body, or smaller than normal. The reason why this happens is often **unknown**, and about 5% of the time it is due to a **genetic** cause. In about 15% of cases of CH the **thyroid looks normal but does not work properly** (does not make enough hormones). Approximately 20% of the time this type of CH has a **genetic** cause, otherwise the reason why it happened is **unknown**.

Could a family have another baby with CH?

Possibly. Most cases of CH are not inherited (genetic). If a baby is thought to have a genetic form of CH, other family members will be checked closely. If the CH has a genetic cause and parents are both identified to be carriers of a non-working gene that causes CH, there is a 25% (1 in 4) chance in each future pregnancy to have a child with CH. There are also other ways CH can be passed on (inherited) in a family. If a genetic cause of CH is identified, the family will be offered genetic counselling through Maritime Medical Genetics Service at the IWK.

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-hypothyroidism.html
  - American Thyroid Association: https://www.thyroid.org/congenital-hypothyroidism/
  - CH Parent Guidebook: https://www.health.state.mn.us/docs/diseases/cy/chparentguide.pdf

- Websites for healthcare providers:
  - StatPearls: https://www.ncbi.nlm.nih.gov/books/NBK558913/
  - UpToDate: https://www.uptodate.com/contents/treatment-and-prognosis-of-congenital-hypothyroidism
  - MedlinePlus: https://medlineplus.gov/genetics/condition/congenital-hypothyroidism/

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.*

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