Information on a Positive Newborn Screen Result for

Tyrosinemia Type 1 (TYR1)

MARITIME Newborn Screening
Dépistage néonatal DES MARITIMES

Summary:
- This result means a baby may have Tyrosinemia Type 1 (TYR1). **More testing is needed** to give a final answer.
- There is treatment for TYR1.
- Maritime Newborn Screening connects the baby with the Maritime Medical Genetics Service metabolic team at the IWK for follow-up testing.
- If the baby has fever, diarrhea, swollen belly, bleeding, jaundice, or vomiting, please seek medical attention.

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.

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. More follow-up testing is needed to give a final answer.

What does it mean if a baby has a positive screen result for Tyrosinemia Type 1 (TYR1)?

This result does not mean that the baby has Tyrosinemia Type 1 (TYR1). It means that more testing is needed because the baby **may** have TYR1. Babies with TYR1 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 TYR1. This is done for the baby's best possible outcome.

What happens when there is a positive newborn screen result for Tyrosinemia Type 1 (TYR1)?

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 the **Maritime Medical Genetics Service metabolic team at the IWK.** The clinical team will arrange follow-up appointments and testing to happen as soon as possible (typically within 1-2 days). 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 Tyrosinemia Type 1 (TYR1)?

Tyrosinemia Type 1 (TYR1) is a **genetic** condition, meaning this is something the baby is born with. TYR1 is also a **metabolic** condition where the body is unable to process protein properly. The body gets energy from three main parts of food: carbohydrates, fats, and proteins. In TYR1 the body cannot process tyrosine, which is one of the building blocks of proteins. When tyrosine cannot be broken down, it builds up in the body's blood and organs. The buildup of tyrosine can cause different health problems, and some can be serious. Babies with TYR1 may have jaundice, vomiting, bleeding (e.g., nose bleeds), and trouble feeding. If not treated TYR1 can cause poor growth, as well as problems with the liver, kidneys, and brain.

How many babies have TYR1?

Tyrosinemia Type 1 (TYR1) is a rare disease that affects approximately 1 in 100,000 to 1 in 120,000 births. There are certain regions in the world where TYR1 is more common, but still rare. TYR1 affects approximately 1 in 74,000 births in Scandinavia, and 1 in 60,000 births in Finland. In Quebec TYR1 affects 1 in 16,000 births and the incidence is highest in the Saguenay-Lac Saint-Jean, Charlevoix, and Haute-Côte-Nord regions of Quebec at approximately 1 in 1,500 births.

Why screen for TYR1?

Babies with Tyrosinemia Type 1 (TYR1) 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. Treatment for TYR1 is most effective when started before or soon after symptoms begin. Newborn screening allows us to find babies that need treatment as soon as possible.

How is a baby diagnosed with TYR1?

To find out if a baby has Tyrosinemia Type 1 (TYR1), they are seen by a metabolic team at the IWK for follow up testing. This team typically includes a doctor, genetic counsellor, nurse, and dietician. Follow-up testing for the baby is done on blood and urine

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samples. Genetic testing (bloodwork) also helps confirm a diagnosis of TYR1. It can take a few weeks to confirm a diagnosis of TYR1. This waiting period can be hard for families. The healthcare teams are here to support families during this time.

How is TYR1 treated?

Treatment for TYR1 starts as soon as possible and continues over the lifetime. TYR1 is treated with a medication called "nitisinone," a low protein diet, and special medical food. Sometimes other treatments such as a liver transplant are considered. A metabolic team with a doctor, nurse, and dietician will help support a newborn with TYR1 and follow them closely.

How does a baby get TYR1?

TYR1 is an **inherited (genetic) condition**. This means the baby is born with the condition. When a baby has TYR1, we know that the 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 gene that causes TYR1 from each parent. This is called autosomal recessive inheritance (see Figure 1).

Could a family have another baby with TYR1?

Yes, if the biological parents are carriers there is a 25% (1 in 4) chance in each future pregnancy to have a child with TYR1. We offer genetic counselling through Maritime Medical Genetics Service at the IWK to talk about the following:

- Inheritance of Tyrosinemia Type 1
- Chance of recurrence in future pregnancies
- Testing options for other family members and in future pregnancies

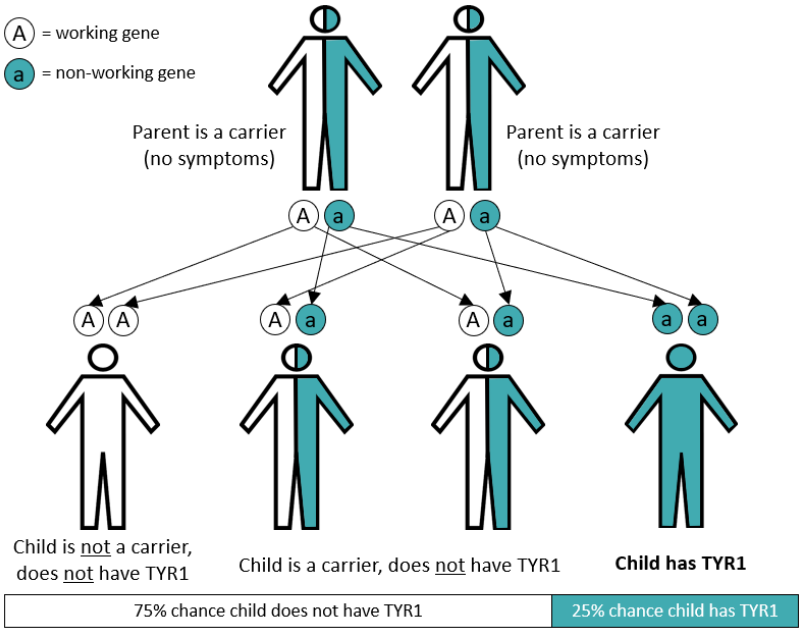

Figure 1. Autosomal recessive inheritance when both parents are carriers of Tyrosinemia Type 1 (TYR1).

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:
  - Tyrosinemia Society: https://www.tyrosinemia.org/
  - New Parent's Guide to Tyrosinemia: http://depts.washington.edu/tyros/abouttyr.htm

- Websites for healthcare providers:
  - MedlinePlus Genetics: https://medlineplus.gov/genetics/condition/tyrosinemia/
  - UpToDate: https://www.uptodate.com/contents/disorders-of-tyrosine-metabolism
  - GeneReviews: https://www.ncbi.nlm.nih.gov/books/NBK1515/

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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