

# Tyrosinemia Type 1 (TYR1)

(Sounds like TIE-ROW-SIN-E-MIA TYPE 1)



## What is newborn blood spot screening?

This is a blood test, from a heel poke, that is done a day or 2 after babies are born to test for treatable conditions including tyrosinemia type 1 (TYR1) that you, your doctor, or midwife can't see by just looking at your baby. This screening test is different from those done during pregnancy.



## What does it mean if my baby needs more tests for TYR1?

A baby needs to have more testing for TYR1 when their screening test result is abnormal or positive. This doesn't mean your baby has TYR1. It means your baby has a higher chance of having the condition and will need more tests to find out if they have TYR1 or not.

It's normal to feel worried if your baby needs more testing. Your specialists and other healthcare providers are there to support you. They'll explain what will happen and answer your questions.

## What is TYR1?

TYR1 is a metabolic condition that affects how your body uses protein from the food you eat to make energy. This includes breastmilk and regular formula. With TYR1, your body can't break down a building block of protein called tyrosine. Tyrosine then builds up in your body and can cause serious health problems.

## What causes TYR1?

TYR1 is genetic. This means babies with TYR1 are born with a gene from both parents, which has a change in it. This change stops the gene from working properly. TYR1 isn't caused by anything that happened during pregnancy.

There may be no signs of TYR1 at birth. It's important to remember that this condition can't be seen by looking at your baby.



## What can I expect with more testing?

Everything that will happen with more testing will be explained to you. Your baby may need to have their blood and urine tested to check for TYR1.

This testing may be done at a clinic in Edmonton or Calgary that specializes in TYR1.

## What if more testing shows my baby has TYR1?

If testing shows that your baby has TYR1 a specialized healthcare team will work with you to make a care plan for your baby. You and your baby will get the best care and support possible.

## How is TYR1 treated?

Babies with TYR1 usually go on a low-protein diet and drink a special formula. They may also need special medicine. The specialized healthcare team will give you detailed instructions about how to manage your baby's health.

