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## INFORMATION FOR PARENTS/GUARDIANS: My Baby Has a Positive Tyrosinemia Newborn Screening Result

### What is newborn screening?

These are routine tests done shortly after birth on every baby born in Ontario. A small sample of blood is taken from the heel of your baby and is tested for rare, treatable diseases, including tyrosinemia.

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

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

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 tyrosinemia until follow up testing has been done.

### What is tyrosinemia?

Tyrosinemia is a rare inherited (genetic) disease that does not allow a baby to break down tyrosine, an amino acid (building block of protein) found in many of the foods we eat, including breast milk and infant formula. Levels of tyrosine and other harmful substances build up in babies with tyrosinemia. If untreated, this build up can lead to serious and permanent health problems like poor growth, liver damage, kidney problems and neurological problems.

Tyrosinemia affects about 1 in every 100 000 babies born in Ontario.

### What causes tyrosinemia?

The most common cause of tyrosinemia happens when the enzyme fumarylacetoacetase (FAH) is either missing or not working properly. This enzyme's job is to break down tyrosine. If the FAH enzyme can not do its job, levels of a substance called succinylacetone become too high and cause health problems.

There are other, more rare forms of tyrosinemia which may also be picked up by newborn screening.

### How do I find out if my baby has tyrosinemia?

Blood and urine tests are done to find out if a baby who screened positive actually has tyrosinemia.

### 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 tyrosinemia?

Babies who have tyrosinemia look normal when they are born but will develop serious health problems and often die if they are not treated. Early signs of tyrosinemia can include feeding problems, extreme sleepiness and poor weight gain. When tyrosinemia is diagnosed and treated early, severe liver, kidney and neurological problems can be prevented and growth and intelligence can be normal.

### How is tyrosinemia treated?

Babies with tyrosinemia are treated and monitored by a team of health care specialists including a metabolic doctor and a dietician. Babies with tyrosinemia are given a special diet that is low in protein (low in tyrosine) and medications.



Babies with tyrosinemia are checked often for their tyrosine and succinylacetone levels, development and other health issues associated with tyrosinemia.

### **How does a baby get tyrosinemia?**

Tyrosinemia is an inherited (genetic) disease. A baby with tyrosinemia inherited two non-working copies of the fumarylacetoacetase (FAH) gene, one copy from each parent. People who have one non-working copy of the FAH gene are called "carriers." Carriers of tyrosinemia are healthy, do not have, and will never develop, symptoms of tyrosinemia.

### **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](http://www.newbornscreening.on.ca).

For more information on tyrosinemia, please see About Tyrosinemia: New Parent's Guide at <http://depts.washington.edu/tyros/abouttyr.htm>.

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

