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

What is newborn screening?
These are routine tests done soon after birth on every baby born in Ontario. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, including phenylketonuria (PKU).

What does it mean if my baby has a positive PKU newborn screening result?
This result does **not** mean that your baby has PKU. It means that more testing is needed because your baby **might** have PKU. Babies with PKU can grow and develop normally if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has PKU.

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 PKU until follow up testing has been done.

What is phenylketonuria (PKU)?
PKU is a rare inherited (genetic) disease that does not allow a baby to break down an amino acid (a building block of protein) called phenylalanine (Phe). Phenylalanine is found in most of the foods we eat, including breast milk and infant formula. Phenylalanine levels build up in babies with PKU and are toxic to the developing brain. Children with untreated PKU have serious and permanent mental retardation as well as other health problems.

PKU affects about 1 in every 12 000 babies born in Ontario.

What causes PKU?
The most common cause of PKU happens when the enzyme phenylalanine hydroxylase is either missing or not working properly. This enzyme's job is to break phenylalanine down into another amino acid called tyrosine. If this enzyme can not do its job, phenylalanine levels become too high and cause health problems.

There are also other, rarer causes of PKU that can be picked up by newborn screening.

How do I find out if my baby has PKU?
Blood tests, and sometimes urine tests, are done to find out if a baby has PKU.

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 follow up testing as soon as possible.

Why screen for PKU?
Babies who have PKU look normal when they are born but will develop mental retardation if they are not treated. If PKU is diagnosed and treated early, mental retardation can be prevented and babies with PKU can grow and develop normally.

How is PKU treated?
Babies with PKU are treated and monitored by a team of health care specialists including a metabolic doctor and a dietician. Babies with PKU are given a special diet that is low in phenylalanine but has other important proteins needed for normal growth and development. Babies with PKU also have their phenylalanine levels, weight gain and development checked on a regular basis.

How does a baby get PKU?

PKU is an inherited (genetic) disease. A baby with PKU inherited two non-working copies of the phenylalanine hydroxylase (the enzyme that turns phenylalanine into tyrosine) gene, one copy from each parent. People who have one non-working copy of the PKU gene are called "carriers." Carriers of PKU are healthy, do not have, and will never develop, symptoms of PKU.

Where can I get more information?

For more information on newborn screening, please visit the Parents section of our website at www.newbornscreening.on.ca or talk to your local health care provider.

For more information on PKU, please visit the Children's PKU Network website at http://www.pkunetwork.org, the National Society for Phenylketonuria website at http://www.nspku.org or the Your Health website at www.ygyh.org.

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