



INFORMATION FOR PARENTS/GUARDIANS: My Baby Has a Positive Beta-Ketothiolase Deficiency 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 beta-ketothiolase deficiency (BKT).

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

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

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 BKT until follow up testing has been done.

What is beta-ketothiolase deficiency (BKT)?

BKT is a rare inherited (genetic) disease that does not allow a baby to break down an amino acid (a building block of protein) called isoleucine. Isoleucine is found in foods that contain protein, including breast milk and infant formula. If a baby cannot break isoleucine down, harmful substances build up in the blood and can make a baby very sick.

Some people with BKT never develop any health problems associated with BKT and some do not develop problems until childhood or adulthood. However, there is no way to know who will develop problems caused by BKT and who will not.

What causes BKT?

BKT happens when an enzyme called beta-ketothiolase (or mitochondrial acetoacetyl-CoA thiolase) is either missing or not working properly. This enzyme's job is to break down isoleucine (an amino acid) into different amino acids. Amino acids are the building blocks of protein.

How do I find out if my baby has BKT?

Blood and urine tests are done to find out if a baby who is screen positive actually has BKT.

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 BKT?

Babies who have BKT are at risk for a metabolic crisis. A metabolic crisis is a serious health condition caused by the build-up of harmful substances in the blood. Symptoms of a metabolic crisis are poor feeding, vomiting, lethargy, excessive sleepiness and irritability. If a metabolic crisis is not treated, seizures, coma, and sometimes death can occur. The goal of screening for BKT is to prevent a metabolic crisis.





How is BKT treated?

Babies with BKT are treated and monitored by a team of specialists including a metabolic genetics doctor and a dietician. Babies with BKT should not go a long time without eating. Sometimes, carnitine and a special diet low in protein are recommended. With early detection and careful treatment, children with BKT usually live healthy lives with typical growth and development.

How does a baby get BKT?

BKT is an inherited (genetic) disease. A baby with BKT inherits two non-working genes, one copy from each parent. People who have one non-working copy of the BKT gene are called “carriers.” Carriers of BKT are healthy and do not have symptoms of BKT.

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.

For more information on BKT, please visit the Organic Acidemia Association website at www.oaanews.org.

NOTE TO PARENTS/GUARDIANS: This information is only applicable if your baby has had a positive newborn screening result for beta-ketothiolase deficiency (BKT). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

