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Newborn Screening Test

What is the Newborn Screening Test?
Newborn Screening is actually a battery of tests done at the same time. Here are some of the tests done:

Congenital Hypothyroidism: 1 in 3000
A baby born with hypothyroidism cannot make enough thyroid hormone. Thyroid hormone keeps a baby's body growing strong and healthy. Without it, delayed growth and brain damage occur. With early detection and thyroid hormone treatment, these children have normal growth and intelligence.

Phenylketonuria (PKU): 1 in 12,000
A baby with PKU is missing an enzyme that is needed to process the essential amino acid phenylalanine, found in certain foods. Without treatment, phenylalanine builds up in the baby's blood and causes mental retardation. If PKU is found early, the baby is fed a special formula that is low in phenylalanine. Most children with PKU who are treated early have normal intelligence.

Galactosemia: 1 in 50,000
A baby with this condition cannot process galactose, a sugar found in milk. If Galactosemia is not treated, it can cause damage to the baby's eyes, liver, and brain. These problems are prevented with a special diet.

Medium Chain Acel-CoA (MCAD): 1 in 12,000
A baby born with MCAD may have problems using fats stored in their body as an energy source. The baby is healthy when eating well. If the baby has a cold or the flu, they may not be able to use the stored fatty acids for energy. There is a risk of sudden unexpected death which can be prevented by using a special diet and avoiding fasting.

Long Chain 3 Hydroxyacyl-CoA Dehydrogenase (LCHAD): 1 detected in BC/Yukon every 2 years
Very similar to MCAD, a baby born with this may have problems using fats stored in their body as an energy source. Treatment is also through special diet and avoidance of fasting.

Glutaric aciduria type I (GA-I): 1 detected in BC/Yukon every 5 years
A baby born with GA-I is missing the enzyme needed to breakdown several types of amino acids (lysine, tryptophan and others). This block in metabolism leads to a buildup of substances such as glutaric acid which causes health problems or can even be fatal. Treatment is with a special diet, avoidance of fasting and supplementation with carnitine and riboflavin.

How is the test done?
A few drops of blood are collected from the heel of your baby's foot. The same blood sample is used for all tests. We are happy to do the test when you are feeding in order to minimize the pain to the baby.

When is the test done?
Hypothyroidism and Galactosemia can be detected any time after the birth, but the other tests will not be accurate before the baby has had a chance to try to digest food, i.e. there is a high chance of a false negative if done before the first 24 hours. Therefore, the Newborn Screen is usually it is done between 24 and 72 hours.

What if the test is positive?
If the test comes back positive, more in-depth testing will be done. If further testing shows that your baby has one of the disorders, you will be referred to a specialist and treatment will start in a few days.

Why does NB Screening in other provinces include more than 6 tests?
It's true that some provinces and states have screening programs that involve more tests. The BC Government has announced that further screening tests will soon be added to those currently being done.