

Newborn Screening



Hear this on tape:

Vancouver: 660-2628

Other: 1-877-660-2628

ID# 1847

Rare disorders can cause mental retardation

In their first week of life, all babies in B.C. are offered a simple blood test. Just before the baby is discharged from hospital, a small blood sample is taken by a simple heel-prick. This sample is sent to the newborn screening lab for analysis.

This is an important test to find those few babies who may have a rare disorder that can cause permanent mental retardation.

With early detection and treatment, mental retardation from these rare disorders can be avoided.

Babies with these disorders *look* normal at birth (even after careful examination by a doctor or midwife). That is why the blood test is so important.

Three disorders that newborns are tested for:

- **Phenylketonuria (PKU)**
- **Congenital hypothyroidism (CH)**
- **Galactosaemia (GS).**

About 50,000 babies are tested for these disorders every year in B.C.

Phenylketonuria (PKU)

PKU is a rare condition. It is found in about 1 out of every 18,000 new babies. A baby with PKU doesn't have enough of a special enzyme that breaks down *phenylalanine* in the body.

Phenylalanine is an amino acid - one of the building blocks of proteins. It is found in foods such as meat, eggs, fish and milk, including breast milk.

A shortage of this enzyme leads to high levels of phenylalanine in the blood. High levels of phenylalanine cause damage to the baby's brain. This usually leads to severe and irreversible mental retardation.

Those very few babies found to have PKU are put on a special diet low in phenylalanine. This prevents brain damage. Children with PKU, when treated early, function within the broad normal range of ability.

Congenital Hypothyroidism (CH)

This is more common than PKU — about 1 in 3,000 new babies are affected. Like PKU, CH is easily detected by the blood screening test.

Congenital means the baby was born with the condition. Hypothyroidism means that the thyroid gland (found behind the "Adams apple" in the neck) is not working properly. The gland does not produce enough thyroxine. This is a hormone which is needed for normal growth and development. If CH is not detected and treated early in life, severe mental retardation will occur.

The treatment for CH is simple and effective. Babies are given thyroxine to replace the missing thyroid hormone. As with PKU, these babies go on to grow and develop normally.

Galactosaemia (GS)

GS is a rare condition, found in only 1 out of every 50,000 babies born in B.C. It is caused by the baby not having the special enzyme that breaks down galactose. Galactose is found in milk, including breast milk, and in most infant formulas. If the baby doesn't have this enzyme to break down galactose, it starts to build up in the body.

Symptoms of this disorder include a general failure to thrive, liver problems (jaundice), cataracts, mental retardation and possibly even infection that can cause death. Early detection is critical. Treatment is by giving the baby a galactose-free diet.

Will the needle prick hurt my baby?

It will only cause a moment of discomfort. You can be assured that the people who take the blood go out of their way to be kind and gentle to the babies.

When will the results be ready?

The results are usually ready in a few days. You will not be contacted if the results are normal (negative). If one of the screening test results is positive, you will be contacted by your family doctor or midwife and additional testing will be arranged. The results of follow-up tests will either be normal (ruling out the possibility of one of these disorders) or will confirm the diagnosis.

What if the result is positive for one of these rare disorders?

Almost all results are negative.

However, if your baby is one of the very few found to have one of these conditions, the early discovery of the disorder will let your baby get effective treatment as soon as possible.

You will be referred to a doctor who is experienced in treating these disorders.

When is the best time for testing?

The newborn screening test should be done during the first week of life. Day 2 or 3 is best.

The screening tests for Congenital Hypothyroidism (CH) and Galactosemia (GS) are reliable any time after birth. However, with Phenylketonuria (PKU), the phenylalanine takes time to increase in the blood. For this reason, the PKU test is not reliable when the test is done less than 13 hours after birth. If the test is done this early, a second sample will need to be taken within the first 2 weeks of life. To avoid repeat testing, it may be possible to postpone the blood test until after discharge from hospital. You should discuss this option with your physician or midwife.

- Babies born in hospital and staying longer than one day will be offered the test before discharge. In some cases, the test may not be given before discharge, but your physician, midwife or hospital nursing staff will arrange for the test to be given within the first week of life.
- Babies born in hospital but discharged within a few hours of delivery, will either return to hospital for testing or have the test done during a home visit within the first week of life. You should discuss these options with your physician, midwife or public health nurse.
- Babies born outside a hospital setting (e.g. at home) under the care of a midwife, will be offered the test during a home visit.

If you have questions about newborn screening, please ask your midwife, public health nurse, hospital nursing staff or your family doctor.