

Borderline positive (slightly abnormal) newborn screen for metabolic diseases



You have just learned that your baby has had a **borderline (slightly abnormal) newborn screen**. This information will answer some of your questions.

About newborn screening

Your baby had a blood sample taken from their heel when they were about two days old. The sample was collected on a special card and sent to the laboratory to be tested for over 23 serious but treatable disorders, including some rare metabolic disorders.

About metabolic disorders

Metabolism is the way the body breaks down food into products it uses for energy, growth and repair. If a person has a metabolic disorder, their body has problems with breaking down food normally or with using broken-down food. Metabolic disorders are genetic and are usually treated with special diets, vitamins or medication.

Why your baby needs a second test

The test result from your baby's first sample was **borderline** (slightly abnormal) for metabolic disorders. The laboratory needs to retest your baby using a fresh sample to check the result.

Please don't worry – this is **not** a serious result. If your baby's result was very abnormal and showed a metabolic disorder was likely, the laboratory would already have notified

your midwife and referred your baby to a metabolic specialist.

Most babies with abnormal screening results do not have anything wrong.

Common reasons for a borderline metabolic result are:

- the testing card had too much blood on it
- the sample has taken a long time to get to the laboratory
- the sample has been exposed to too much heat
- baby's liver enzymes are still maturing, which all systems do after a baby is born.

What happens next

Your midwife will arrange to collect another blood sample from your baby. The laboratory will have the test result one to two days after it gets the sample. It will then call or text your midwife with the result.

In most cases the repeat sample is normal and babies do not need any more testing.

If your baby's repeat sample is not entirely normal, your baby will be referred to a metabolic specialist or paediatrician for a full assessment.

Where to go for more information

For more information about newborn metabolic screening, go to <https://www.nsu.govt.nz/pregnancy-newborn-screening/newborn-metabolic-screening-programme-heel-prick-test>