

# Borderline positive (slightly abnormal) newborn screen for congenital adrenal hyperplasia (CAH)



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

A short time after your baby was born, some blood was collected from their heel to test for some rare disorders, including congenital adrenal hyperplasia (CAH). We screen newborns in this way to find conditions where early treatment can help babies to stay well.

## Why your baby needs a second test

Your baby's screening test for CAH was **borderline** (slightly abnormal). The laboratory needs to retest your baby using a fresh sample to check the result.

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

Common reasons for a borderline CAH result are:

- the testing card had too much blood on it
- the baby was born prematurely with adrenal enzymes that are not yet fully developed.

## About CAH

CAH is an inherited condition where the adrenal gland does not produce the right amount of hormones. It can affect these three hormones.

**Cortisol:** With CAH, this hormone can be lower than normal. When it is working normally, it helps regulate energy levels, blood pressure and glucose metabolism.



**Aldosterone:** With CAH, this hormone can be lower than normal. When it is working normally, it helps regulate salt and body fluid volume.

**Androgens** (male hormones): CAH can increase these hormones and cause someone to develop early signs of puberty.

CAH can be mild or severe. If a baby with a borderline result has CAH, they are likely to have a mild form of it.

Children with mild CAH are more likely to experience early puberty and be shorter as adults, but they remain healthy as babies. If they need treatment, it can prevent all of the problems that CAH can cause. Please discuss any of your concerns with your midwife.

## What happens next

Your midwife will collect another blood spot (heel prick) sample from your baby and send the testing card to the laboratory. 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 the level is still not completely normal, your baby will be referred to a paediatrician for a full assessment.

The treatment for CAH is to replace the hormones the baby can't make. The replacement hormones are given as a medicine.

## Where to go for more information

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

For more about what it is like to live with CAH, see Richard's story at <https://www.nsu.govt.nz/pregnancy-newborn-screening/newborn-metabolic-screening-programme-heel-prick-test/about-newborn/tell>