

Positive Result:

Blood Spot Screen Result Notification



Elevation of 17-Hydroxyprogesterone (17-OHP)

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of a hormone called 17-hydroxyprogesterone (17-OHP).

What does this mean?

High levels of 17-OHP is often seen in newborns born before their due date. However, it can also indicate that your child has congenital adrenal hyperplasia (CAH). The word congenital means the problem starts at birth. A positive result does not mean your baby has CAH, but more testing is needed to know for sure.

What happens next?

Your baby's doctor or a specialist familiar with CAH (a pediatric endocrinologist) will help arrange for more testing. Your baby will also likely be scheduled to be seen by the specialist.

What is congenital adrenal hyperplasia?

CAH is a condition that affects how the body makes hormones. Hormones help the body work and grow properly. A person with CAH doesn't make enough of a hormone called cortisol (also called the "stress hormone").

There are three types of CAH: non-classical, simple virilizing, and salt-wasting. Simple virilizing and salt-wasting are grouped together as classical CAH. Non-classical CAH is the least severe and salt-wasting is the most severe.

What health problems can it cause?

If untreated, children with non-classical CAH may show signs of early puberty or no symptoms at all.

If simple virilizing CAH is not treated, a child might develop:

- Rapid growth in childhood
- Early puberty

If salt-wasting CAH is not treated, a child might develop:

- Poor feeding and weight gain
- Sleepiness
- Dehydration (loss of fluid)
- Life-threatening loss of salt from the body

Girls with classical CAH (simple virilizing and salt-wasting types) may have male-looking genitalia.

Children with CAH can benefit from prompt and careful treatment.

What treatment options are available?

Although CAH cannot be cured, it can be treated. The most common treatment for CAH are medications that replace the missing hormones that aren't being made correctly. Surgery is an option for girls who have male-looking genitalia.

Children with CAH should see their regular doctor and a doctor who specializes in CAH (an endocrinologist).

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org