When Baby Needs a Second Test for Congenital Adrenal Hyperplasia

A small sample of your baby's blood was collected soon after birth and sent to the DHEC laboratory for testing. This testing is called Newborn Screening. In SC, newborns are tested for several genetic and chemical disorders. Some times, a second test is needed to help your doctor decide if your baby has one of the disorders. In many cases, the second test will be normal. However, if your baby does have one of the newborn screening disorders, early treatment will give him or her the best chance to grow up healthy.

Because a compound called 17 hydroxyprogesterone (17-OHP) was high in your baby's first test, he or she could possibly have a disorder called Congenital Adrenal Hyperplasia (CAH).

Please be aware that a higher than expected 17-OHP can also be found in many babies who do not have CAH. Your baby needs to be retested quickly, though, as a precaution.

What is Congenital Adrenal Hyperplasia (CAH)?

Congenital adrenal hyperplasia (CAH) is a genetic disorder found in around one out of every 16,000 babies born each year. When a baby has CAH, he or she cannot produce one or more of the chemicals (called hormones) in the adrenal glands that are needed to keep the body's systems working like they should.

What are the adrenal glands?

The adrenal glands are some of the organs in the body that make hormones. Hormones are like chemical messengers that tell other organs or systems in the body what to do to work properly. The outer parts of the adrenal glands make three important hormones.

They are:
- Cortisol—helps the body keep a steady energy supply and blood sugar level
- Aldosterone—helps the body keep normal levels of "salt" minerals like sodium and potassium
- Androgens—help the body grow normally for both males and females

What happens in a baby with CAH?

Babies with CAH do not make enough cortisol and, in some cases, aldosterone. They also make more androgens than they need. Babies who are not treated will not grow and mature as they should. In severe cases, the baby can go into a coma.

How will I know if my baby really has CAH?

If your baby's newborn screening result showed very high 17-OHP, he or she probably has CAH. The newborn screening test will be repeated and additional tests may also be done to help the doctors figure out if your baby has CAH. The results of these other tests may take a while to come back. You will also be referred to a doctor who specializes in these kinds of disorders.

What do I need to do until I know the final results?

CAH can cause your baby to get very sick. Even minor illness can be a medical emergency in a baby with CAH. Follow your doctor's instructions very carefully. If your baby is hard to wake up or seems to be getting sick, your doctor may have you call 911 for emergency help.

How is CAH treated?

Treatment involves giving the baby the cortisol and aldosterone his or her body cannot make in the form of medicines. This will also stop the baby's body from making too many androgens. The baby will need to take this medicine for the rest of his or her life.

What else should I do to keep my baby as healthy as possible?

Don't forget to keep all of your well baby check-ups! Seeing the doctors regularly and giving your baby medicine every day are the best things you can do to help your baby grow and develop.