Children's of Alabama®

CONGENITAL ADRENAL HYPERPLASIA

Children's of Alabama Parent's Guide

What is Congenital Adrenal Hyperplasia?

Congenital Adrenal Hyperplasia, also called CAH, is an inherited disorder that affects the adrenal glands. Children with CAH receive one CAH gene from each of their parents, and often there is no known family history of the disorder.

Infants born with CAH cannot produce adrenal hormones normally. CAH can be severe (classic) or mild (non-classic). Newborn screening tests screen for severe CAH, so some infants will be diagnosed at birth. Mild forms of CAH may not be diagnosed until childhood or adulthood.

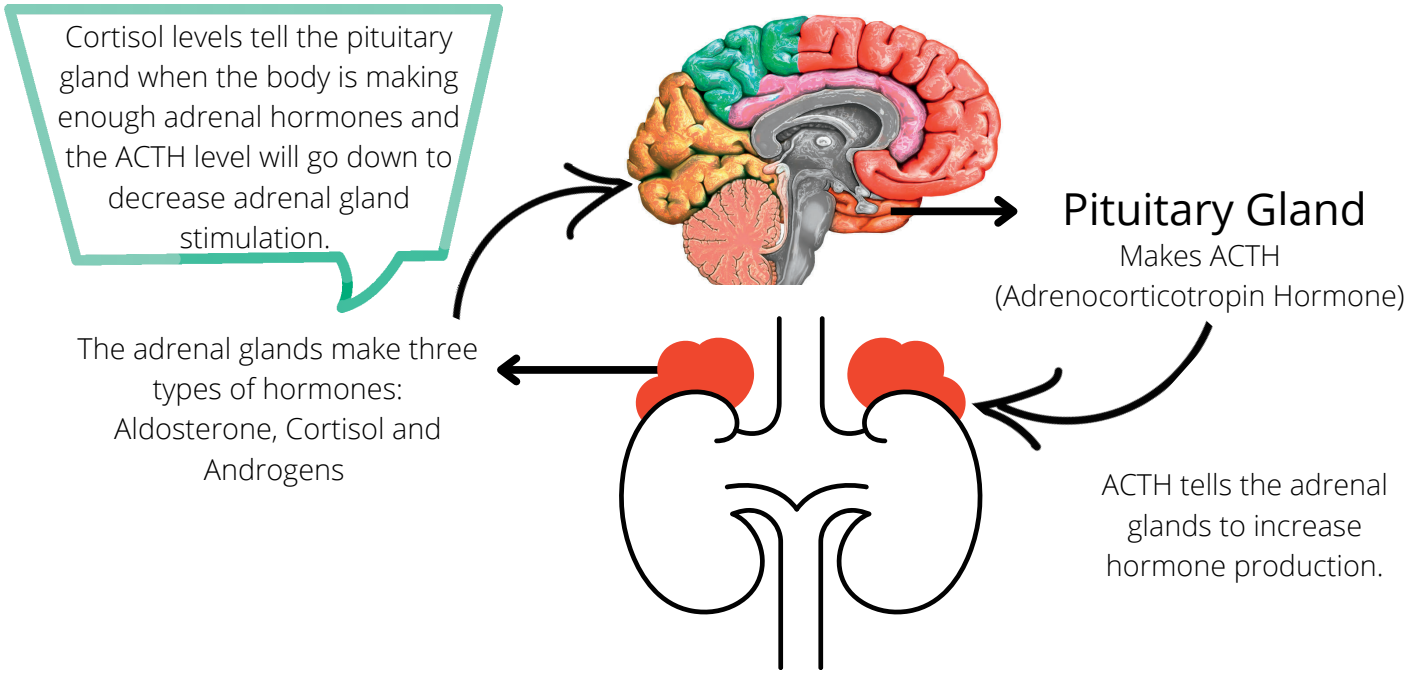
CAH is diagnosed in approximately one of every 15,000 infants.

How does CAH affect adrenal hormones?

Babies with CAH lack an important enzyme in the production of certain adrenal hormones. This could be thought of as a roadblock in the hormone production pathway. Most often, infants with CAH have little to no *21-hydroxylase* enzyme activity. This enzyme is needed to make Aldosterone and Cortisol.

Aldosterone is a salt and water-regulating hormone, and **cortisol** is an energy and blood sugar regulating hormone. In severe CAH (classic) the body cannot produce aldosterone or cortisol, which can be life threatening. In milder cases the body may make some of these hormones, but it may not be enough.

When the cortisol level is low, the body responds by increasing ACTH production from the pituitary gland. ACTH stimulate the adrenal glands to work harder to make hormones. When the adrenal glands cannot make aldosterone or cortisol, they make too much of the adrenal **androgens**. High androgen levels can cause increased body hair, body odor, acne, and early puberty if left unchecked.

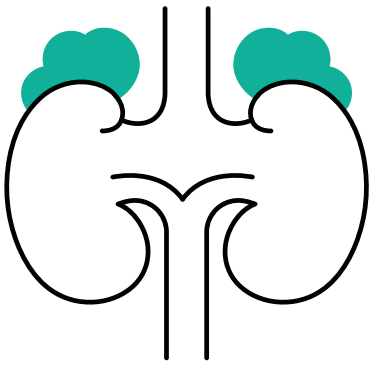


Signs and Symptoms

The signs and symptoms of CAH are different for each infant. Some babies may show no signs during infancy, and others can become severely ill within the first few weeks of life.

Symptoms of more severe illness may include:

- Genitalia changes
- Weight loss
- Feeding difficulties
- Dehydration
- Vomiting or Diarrhea
- Irregular Heartbeat



How is CAH diagnosed?

Newborn Screening is critical to detecting congenital adrenal hyperplasia early. Blood testing is then required to diagnose CAH.

Cortisol levels and electrolyte levels are helpful in diagnosing CAH. The 17-OHP (***17-hydroxyprogesterone***) level also helps to determine the severity of your child's CAH.

Sometimes, a stimulation test will be needed to diagnose CAH. This is a special test where levels are measured before and again after a medication (Cortrosyn) is given to stimulate the adrenal glands to make hormones.

Genetic testing is often obtained at a later time to help better understand your infant's diagnosis.

Treatment

The treatment of Congenital Adrenal Hyperplasia is hormone replacement.

Infants are prescribed medications to replace the hormones that their body cannot produce.

Fludrocortisone is used to replace Aldosterone and Hydrocortisone is used to replace Cortisol. Infants with classic CAH may also require extra sodium.

These medications come in tablet form, and will need to be crushed and mixed with a small amount of water or formula.

If you feel your infant is struggling to take the tablet form of the medication, please discuss this with your provider.

The beginning of treatment can be a stressful time for many parents, but know that you will develop a medication routine that will work for you and your infant.

It is important to know that Hydrocortisone dosing will need to be increased at times when your child is sick or injured. There are also special circumstances where the cortisol replacement must be given by injection. Your provider will discuss with you what to do when your child is not well. At each visit be sure to discuss your "Sick Day Plan."

Consistency is very important when giving these medications. It is best to not miss doses of medication. We recommend "making up" any missed doses as soon as possible. You should also discuss any changes to the medication routine with your provider, as these variables may change how the medication is absorbed.

Adrenal Crisis

Notify your provider if your child is showing any of the following symptoms:

- Excessive sleeping
- Tremors (shaking)
- Weight loss
- Irritability
- Abdominal Pain
- Vomiting or Diarrhea
- Poor feeding
- Fever
- Weak cry
- Confusion
- Pale
- Rapid breathing

- It is best to not give medication in a full bottle--this way you can ensure that your infant gets their full dose.
- Speak with your provider regarding any changes to your infant's formula or medications (even vitamins).
- As your infant grows, the medication may be administered differently. Review medication administration with each clinic visit.
- All children with CAH should wear a medical alert identification that states: "Adrenal Insufficiency"

The Future

Your child has a bright future. Thanks to newborn screening and readily available thyroid hormone replacement, infants who are born with congenital hypothyroidism grow and develop the same as other children.

The important thing to remember is that consistent medication administration and routine lab evaluation and follow-up are key to excellent outcomes in childhood and beyond.

Resources

Alabama Department of Public Health: Newborn Screening
www.adph.org/newbornscreening

Baby's First Test www.babysfirsttest.org

American Thyroid Association www.thyroid.org

The Magic Foundation www.magicfoundation.org