

# Congenital Adrenal Hyperplasia (CAH)

## General Overview

---

### **Q. What is CAH?**

A. Congenital adrenal hyperplasia (CAH) is a treatable disorder that occurs when the adrenal glands do not function properly. The result is that normal amounts of essential hormones are not produced. If left untreated, CAH will cause problems with growth and development and can cause life-threatening illness.

### **Q. Is there only one form of CAH?**

A. No. There are several forms of CAH, but one specific enzyme problem accounts for over 90 percent of diagnosed cases. This form is called 21-hydroxylase enzyme deficiency. Several other enzyme deficiencies involved in the production of adrenal hormones can also cause CAH.

### **Q. How do the adrenal glands normally function?**

A. The adrenal glands produce many different hormones that help the body to function normally. The three main hormones involved in CAH are: cortisol (affects the body's metabolism and its response to injury and infections), aldosterone (affects how the kidneys work), and androgens (affect body growth and development).

### **Q. What happens to the adrenal glands in a child with CAH?**

A. In a person with CAH, the adrenal glands are unable to properly regulate the hormones they produce. The result is too little of both cortisol and aldosterone, and too much androgen hormones.

### **Q. What are the effects of having CAH if it is not treated?**

A. If untreated, children with CAH will experience abnormally rapid growth early in childhood (but stunted in the long run) and early appearance of body hair. Babies with the salt-wasting form of CAH (about 75 percent of cases) are at risk for rapid, uncontrolled loss of salt from the body that can result in death. The imbalance of hormones before birth may cause some girls to have ambiguous genitalia.

### **Q. What is the treatment for CAH?**

A. Congenital adrenal hyperplasia is treated with daily supplements of the hormone cortisol, and in many cases a salt-retaining hormone. To prevent problems, treatment must begin shortly after birth.

### **Q. Why would a child have CAH?**

A. Congenital adrenal hyperplasia is an inherited disorder. It results when a baby receives a double-dose of a non-working gene for cortisol production (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

### **Q. How common is CAH?**

About one in every 16,000 babies in Washington State is born with CAH.

For more information about CAH, please see the Disorders section of our website: [www.doh.wa.gov/nbs](http://www.doh.wa.gov/nbs).



Washington State Department of Health • Newborn Screening Program  
1610 NE 150th Street, K17-9 • Shoreline, WA 98155-0729 • [www.doh.wa.gov/nbs](http://www.doh.wa.gov/nbs)  
Phone: (206) 361-2902 • FAX: (206) 361-4996 • E-mail: [NBS.Prog@doh.wa.gov](mailto:NBS.Prog@doh.wa.gov)

Revised: 04/04