VDH VIRGINIA DEPARTMENT OF HEALTH

Division of Child and Family Health- Newborn Screening Follow-Up Program

Congenital Adrenal Hyperpasia (CAH)

Health Care Professional Fact Sheet

A newborn screening test is a screen and not diagnostic testing. An "abnormal" or "critical" result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose the baby with the condition. Follow-up testing is **vital** to determine if the baby has the disorder indicated. In the event the condition is diagnosed, timely follow-up testing will result in earlier treatment and better outcomes.

Disorder Indicated: Congenital adrenal hyperplasia (CAH) is a collection of inherited conditions that affect the body's adrenal glands. In a person with CAH, the adrenal glands are unable to produce certain chemicals, including cortisol, a chemical that helps protect the body during stress or illness and helps regulate the amount of sugar in the blood. If left untreated, CAH could cause brain damage, coma, or death. However, if the condition is detected early and treatment is begun, individuals with CAH can have healthy growth and development.

Incidence	1 in every 15,000 newborns.
Analyte Measured	17-hydroxyprogesterone (17-OHP)

Screening Test Results (*by birth weight*)

Birth Weight	Normal	Abnormal	Critical
<1250 g	<55 ng/mL	≥ 55 ng/mL	≥ 65 ng/mL
1250 – 1749 g	<40 ng/mL	≥ 40 ng/mL	≥ 60 ng/mL
1750 – 2249 g	<35 ng/mL	≥ 35 ng/mL	≥ 45 ng/mL
≥2250 g	<25 ng/mL	≥25 ng/mL	≥ 45 ng/mL

Signs and Symptoms

Please note: these findings may not be present in young infants or in milder forms of the disease

The signs may vary from person to person and by the form of CAH. There are three main forms of CAH: the "salt-wasting form," the "simple virilizing form," and non-classic CAH (least severe form).

Salt-wasting form

- Most severe form- 75% of classic CAH cases
- Unrecognized this condition leads to severe dehydration

When a child has this form of CAH, you may see symptoms including:

- Poor feeding/ Weight loss
- Sleeping longer/Tiredness/ Irritability
- Vomiting/Diarrhea
- Rapid heart rate
- Male-like genitals in females

Simple virilizing form

- 25% of classic CAH cases
- Signs can begin before birth.

When a child has this form of CAH, you may see symptoms including:

- Enlarged clitoris (it may look like a small penis)
- Labia that are fused together (they may resemble a scrotum)
- Undescended or small testicles

Next Steps *may* include:

Consult with local pediatric endocrinologist
Provide parental education (see accompanying sheet)
Clinical assessment
Labs: Serum 17-OHP, electrolytes, glucose

Treatment (if indicated): Hydrocortisone pills, fludrocortisone, other supplements

Additional Resources

VDH Newborn Screening *http://vdhlivewell.com/newbornscreening*
Baby's First Test *www.babysfirsttest.org*
American College of Medical Genetics (ACMG) ACT Sheets *www.ACMG.net*
Genetics Home Reference *https://ghr.nlm.nih.gov/*
Cares Foundation *https://www.caresfoundation.org/*

Educational content adapted from www.babysfirsttest.org

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