

Congenital Adrenal Hyperplasia (CAH)

MEDICAL FACT SHEET POSITIVE NEWBORN SCREEN

Action required

Contact pediatric endocrinologist today.
See infant today.

Issues to discuss with Endocrinologist

- Laboratory evaluation of infant
 - § Should testing be performed by primary care or endocrinology clinic
 - § Serum 17-OHP
 - § Serum electrolytes

False Positives

- Common
- Screening result can be impacted by prematurity

False Negatives

- Screening result can be impacted by hormone therapy provided to the mother or infant

Review with family

Family has **not** been notified of result by MDH.

After discussion with pediatric endocrinologist, contact family to coordinate clinic visit, lab work, and referral to endocrinology clinic, if necessary. Child may already have signs of CAH, depending on CAH type.

Prompt follow-up and intervention are critical.

NICU issues

False positive newborn screens are seen commonly in the NICU. For infants weighing <1800g, all 3 screening results should be reviewed.

Newborn screens cannot be accurately interpreted if collected before 24 hours or after administration of hormone therapy.

Clinical summary

Congenital Adrenal Hyperplasia (CAH) is an autosomal recessive disorder involving impaired synthesis of cortisol by the adrenal cortex.

Children affected with CAH are at risk for life-threatening adrenal crises. Female newborns with classic virilizing CAH often have ambiguous genitalia. If an infant, male or female, is not screened and/or left untreated, symptoms begin to appear later in infancy and can include precocious puberty, advanced bone age, shock, and salt-wasting adrenal crises.

Affected children require life-long treatment with cortisol and glucocorticoids and monitoring by both primary care and specialty providers.

Incidence: ~ 1/18,000; affects all ethnic

Clinical expectations

Salt-wasting adrenal crises can occur even with proper treatment. Affected children will continue to need monitoring throughout childhood. During periods of stress, increased amounts of glucocorticoids will be required.

Ambiguous genitalia in females can be surgically addressed.

Resources

GeneTests: www.genetests.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

ACT Sheets: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

MN Newborn Screening Program:
www.health.state.mn.us/newbornscreening