

**FACT SHEET**  
Healthcare Provider

**Congenital Adrenal Hyperplasia (CAH)**

**Description:**

Congenital adrenal hyperplasia (CAH) is an autosomal recessive condition. CAH consists of a group of disorders arising from specific defects in the enzymes of the adrenal cortex required for the biosynthesis of adrenal corticosteroids. The most common form results from 21-hydroxylase deficiency, which accounts for more than 90% of all recognized cases. A child affected with CAH can go into adrenal crisis if they experience any stressors, such as an infection, an injury, or surgery. An adrenal crisis, or the complete failure to maintain normal balance, can result in death. In the most severely affected female newborn, the fetal adrenal androgens may masculinize the external genitalia to the extent that an incorrect male sex assignment is made. In approximately 75% of affected newborn boys, a life threatening “salt-wasting syndrome,” may be the only clinical finding leading to the correct diagnosis. Treatment prevents acute adrenal insufficiency by replacing the deficient steroid hormones and prevents the long-term consequences of excess virilization, advanced bone maturation leading to precocious puberty, and adult short stature by suppressing the excess adrenal androgens.

**Incidence in General Population:**

1:25,000 live births

**Symptoms:**

- Ambiguous genitalia in females
- Enlarged penis and scrotum with increased pigmentation in males
- Frequent urination
- Poor feeding
- Vomiting
- Dehydration
- Electrolyte changes
- Cardiac arrhythmia
- Precocious puberty
- Premature skeletal maturation

**Newborn Screening Technology:**

Detection by an immunofluorescent assay (IFA) for 17-hydroxy progesterone (17OHP). IFA first measures the level of 17 OHP in the blood. For infants whose 17-OHP is in either the highest 3% of results or above 40 ng/ml, the test result is confirmed by repeat testing. The interpretation of the generated 17-OHP result is then based on the infant’s birth weight.

**Diagnosis:**

Measurement of serum 17-OH progesterone level and serum electrolytes is recommended along with referral to a pediatric endocrinologist. Early detection and treatment prevents adrenal insufficiency with dehydration, shock, and even death.

**Monitoring:**

Lifelong monitoring, management, and compliance with treatment are essential to the child’s well being.

**Treatment:**

Lifetime daily medication of hydrocortisone in children and prednisone or dexamethasone for older individuals to replace missing cortisol. In cases of salt-wasting CAH, in addition to hydrocortisone, fludrocortisone is prescribed to correct aldosterone deficiency. Infants and small children with salt-wasting CAH also may require salt tablets as dietary supplement. Regulation of medication dosage is vital because improper dosage can result in either growth delay or premature bone epiphyseal closure. Female infants with ambiguous genitalia may require re-constructive surgery.

**Immunizations:**

Immunizations must be kept current

**Growth and development:**

It is crucial to closely monitor all growth parameters on a regular basis.



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