

## Congenital Adrenal Hyperplasia (CAH)

CAH is a family of diseases whose common feature is an enzymatic defect in the steroidogenic pathway leading to the biosynthesis of cortisol. The 21-hydroxylase deficiency accounts for 90-95 percent of CAH cases, resulting in ambiguous genitalia in females and salt-losing crisis in either males or females. Early detection and treatment is essential to prevent death in infants with salt-losing CAH.

**Prevalence:** 1:12,000

**Analyte Measured:** 17-hydroxyprogesterone (17-OHP)

**Reporting Ranges:** Reporting ranges are weight dependent

<b>Birth Weight (gm)</b>	<b>17-OHP (ng/ml)</b>
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**Normal**

0-1500 gm	< 150
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1501-2000 gm	< 80
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2001-2500 gm	< 60
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> 2500 gm	< 50
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Repeats requested or follow-up per consultant for values > normals.

**Feeding Effect:** None

**Timing Effect:** False positive 17-OHP results may occur if sample is collected before 24 hours of age.

**Other Effect:** EDTA can cause a false positive screening result.

**Confirmation:** Repeat screen (for results of 'concern') or confirmatory test (for results that are 'urgent') as directed by program staff or pediatric endocrinology consultant.

**Treatment:**

Referral will be made to Pediatric Endocrinologist.  
**Glucocorticoids:** to replace cortisol and suppress excessive corticotropin and androgen production.  
**Mineralocorticoids:** for salt-losing and elevated plasma renin.  
**NaCl supplement:** may be necessary.  
**Ambiguous genitalia:** Surgery

**Comment:**

Repeat of non-normal 17-OHP newborn screening tests by a “reference” laboratory other than the laboratory recommended by the pediatric endocrinologist may cause confusion in interpreting results due to different reporting methods.