Congenital Adrenal Hyperplasia (CAH)
Fact Sheet

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-hydroxylace 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

Birth Weight (gm)	**17-OHP (ng/ml)**		
	Normal	**Concern**	**Urgent**
0-1500 gm	<150	100-134	≥135
1501-2000 gm	<75	75-99	≥100
2001-2500 gm	<60	60-69	≥70
>2251 gm	<50	50-59	≥60

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
Glucocorticosteroids: to replace cortisol and suppress excessive corticotropin productions and suppresses excessive 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.

Maine Newborn Screening Program
(207) 287-5357 or 1-800-698-3624

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