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-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

<table>
<thead>
<tr>
<th>Birth Weight (gm)</th>
<th>Normal</th>
<th>17-OHP (ng/ml)</th>
<th>Urgent</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-1500 gm</td>
<td>&lt;150</td>
<td>100-134</td>
<td>≥135</td>
</tr>
<tr>
<td>1501-2000 gm</td>
<td>&lt;75</td>
<td>75-99</td>
<td>≥100</td>
</tr>
<tr>
<td>2001-2500 gm</td>
<td>&lt;60</td>
<td>60-69</td>
<td>≥70</td>
</tr>
<tr>
<td>&gt;2251 gm</td>
<td>&lt;50</td>
<td>50-59</td>
<td>≥60</td>
</tr>
</tbody>
</table>

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
Sep. 2003 (207) 287-5357 or 1-800-698-3624