

Follow-up of Abnormal Screens for Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency
Revised 9/2/2011

| | | |
|---------------------------------|---|----------------------|
| Birth Weight \geq 2500 grams: | | |
| 17 OHP ng/ml | | Steroid Profile |
| \geq 55 | & | Abnormal |
| \geq 80 | & | Pending ¹ |

¹Completion of confirmatory testing required regardless of LC MS/MS results.



Day 1 Emergency Protocol:

1. Locate infant for confirmatory testing and clinical evaluation in the office or ER within 24 hours (if unable to locate infant within 8 hours or unable to meet 24 hour deadline for testing, contact Newborn Screening Endocrine Nurse).
2. Testing requirements:
 - 17-OHP to DLO (state contracted lab)
 - BMP
 - Repeat filter paper (optional)
3. Clinically assess infant for ambiguous genitalia and signs and symptoms (S&S) of adrenal insufficiency (see Table 1).
4. After assessment and review of BMP results, consult with a pediatric endocrinologist (PE) for medical management recommendations (key information includes gestational age, steroid therapy, BMP values, and clinical assessment findings).

Confirmatory Test Results Pending - Monitoring Protocol:

1. Immediately notify Pediatric Endocrinologist if infant becomes clinically unstable.
2. Upon receipt of 17-OHP results, contact the PE for management recommendations and final diagnosis.

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|---------------------------------|---|-----------------|
| Birth Weight \geq 2500 grams: | | |
| 17 OHP ng/ml | | Steroid Profile |
| \geq 55 to 80 | & | Pending |
| 30 to 54 | & | Abnormal |

| | | |
|----------------------------|---|-----------------|
| Birth Weight < 2500 grams: | | |
| 17 OHP ng/ml | | Steroid Profile |
| \geq 75 | & | Abnormal |



Borderline Protocol

1. Monitor Basic Metabolic Panel (BMP) daily (if BMP is abnormal-Na low and K high, promptly consult with a pediatric endocrinologist).
2. Repeat filter paper within 24-48 hours.
3. Monitor for S&S of adrenal insufficiency.
4. Consult with a Pediatric Endocrinologist (within 24 hours) for any of the following:
 - Abnormal BMP
 - Ambiguous genitalia for girls
 - Hypospadias or hyperpigmentation for boys
 - S&S of adrenal insufficiency

Table 1. S & S of Adrenal Insufficiency

- Change in feeding patterns
- Poor weight gain
- Vomiting
- Diaphoresis
- Tachypnea
- Pale mucous membranes
- Dehydration
- Decreased Na levels
- Increased K levels
- Hypoglycemia

| | | |
|----------------------------|---|-----------------|
| Birth Weight < 2500 grams: | | |
| 17 OHP ng/ml | | Steroid Profile |
| \geq 95 | & | Pending |



Low Birth Weight Protocol for Significant 17-OHP Elevations:

1. Notify physician of record
 - Preliminary report: significantly elevated 17-OHP with pending steroid profile
 - Follow-up is at the provider's discretion until steroid profile is back.
2. NBS follow-up will be in contact with the physician **ONLY** if the steroid profile is abnormal.
3. The provider is responsible for confirming the final newborn screening results.
4. Repeat filter paper is recommended for all sick or premature infants at 14 days of age.

Screen Results –
Not Consistent with CAH

| | | |
|---------------------------------|---|-----------------|
| Birth Weight \geq 2500 grams: | | |
| 17 OHP ng/ml | | Steroid Profile |
| < 30 | & | Not performed |
| < 50 | & | Normal |

| | | |
|----------------------------|---|-----------------|
| Birth Weight < 2500 grams: | | |
| 17 OHP ng/ml | | Steroid Profile |
| < 50 | & | Not performed |
| \geq 50 | & | Normal |