Newborn Screening Follow-up: Preventing Morbidity and Mortality in Children with Congenital Hypothyroidism and Congenital Adrenal Hyperplasia

Satellite Conference and Live Webcast
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Presentation Overview
- Discussion of the pathophysiology of congenital hypothyroidism and congenital adrenal hyperplasia, with primary emphasis on the diagnostic criteria and screening process
- Highlight case studies of various congenital hypothyroidism and CAH presentations

Presentation Overview
- Evaluate the value of long-term management following initial diagnosis
- Explore challenges encountered in the treatment process
- Review frequently asked questions regarding diagnosis confirmation and initiation of treatment

Presentation Overview
- Overview services provided by Children’s Hospital of Alabama

Exploring Pathophysiology: Congenital Hypothyroidism and Congenital Adrenal Hyperplasia
**Congenital Hypothyroidism**

- Inability of the thyroid gland to produce adequate thyroxine (T4) and triiodothyronine (T3)
- Presents in the newborn period
  - Even severe cases are often clinically silent in infants
  - Therefore newborn blood screening universally recommended

- Approximately 10% of cases are due to dyshormonogenesis
  - Defect in hormone synthesis, often inherited in autosomal recessive pattern
- Small percentage due to central hypothyroidism
  - Inadequate TSH stimulation

- Rarely, transient hypothyroidism secondary to transplacental passage of maternal medications or antithyroid autoantibodies

- Optimal thyroid hormone levels are critical for normal neurodevelopment
- Untreated congenital hypothyroidism can produce profound somatic and neurologic delay
  - Albert, et al., 2013, p. 36-64

**Congenital Hypothyroidism**

- Occurs in approximately 1:3,000 – 1:4,000 babies
  - More common in Hispanic population
- Approximately 85% of cases are sporadic and due to thyroid dysgenesis
  - Abnormal thyroid anatomy

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**Congenital Hypothyroidism**

- One of the most common preventable causes of mental retardation in the world
  - Huang, 2010, p. 115
- When treatment is initiated early (preferably within the first 14 days of life) and sustained, it is believed that children will have normal developmental outcomes
  - Balhara, Misra, and Levitsky, 2011, p. 536

**Congenital Adrenal Hyperplasia 21-Hydroxylation Deficiency**

- A family of inherited disorders affecting the adrenal gland’s ability to produce cortisol
- Most common form is 21-hydroxylase deficiency
  - 90-95% of cases
- Inherited in an autosomal recessive pattern

**Common Forms of CAH 21-Hydroxylase Deficiency**

- Salt-losing
  - Also Salt-wasting or Classical
- Simple Virulizing
- Attenuated
  - Non-classical
**Common Forms of CAH 21-Hydroxylase Deficiency**

- Adrenal Glands
- Kidneys

**Classical Salt-Wasting CAH 21-Hydroxylase Deficiency**

- Severe to complete deficiency of 21-Hydroxylase
- Inability of the adrenal cortex to produce Cortisol or Aldosterone
- Results in adrenal crisis and maximal secretion of adrenal androgens
- Near total masculinization of external female genitalia in females

**Simple Virulizing CAH 21-Hydroxylase Deficiency**

- Incomplete or partial 21-Hydroxylase deficiency
- Results in increased ACTH production in order to normalize Cortisol levels

**Simple Virulizing CAH 21-Hydroxylase Deficiency**

- Increased levels of Cortisol precursors
  - 17-Hydroxyprogesterone, etc.
- Increased Aldosterone production, results in normal sodium balance
**Simple Virulizing CAH 21-Hydroxylase Deficiency**

- Increased androgen production, due to increased ACTH stimulation and partial enzyme blockage

**Simple Virulizing CAH 21-Hydroxylase Deficiency**

- Variable degrees of female masculinization present at birth
- If undiagnosed at birth, may develop signs of puberty at a very early age, or advanced somatic growth and skeletal age

**Steroidogenesis**

![Steroidogenesis Diagram]

**Attenuated or Non-classical CAH**

- Minimal 21-Hydroxylase deficiency
- No female masculinization present at birth
- Only small changes are noted in steroidogenesis

**Attenuated or Non-classical CAH**

- Changes typically develop in girls during puberty due to excess androgen secretion
  - Development of hirsuitism
  - Severe acne
  - Menstrual irregularities
  - Small ovarian cysts possible

**Attenuated or Non-classical CAH**

![Attenuated or Non-classical CAH Graph]
Making the Diagnosis: Congenital Hypothyroidism and Congenital Adrenal Hyperplasia

Making the Diagnosis: Congenital Hypothyroidism and Congenital Adrenal Hyperplasia

- State Newborn Screening is optimally performed at 2 to 4 days of age
- Normal neonatal TSH surge occurs during the first hours of life
- Screens resulting in high TSH and low T4 are concerning for Congenital Hypothyroidism and warrant further attention

Confirming Congenital Hypothyroidism

Abnormal Newborn Screen Results
- TSH
- Total T4

- TSH ≥ 150 with collection at <24 hours
- Or
- TSH ≥ 40 with collection at > 24 hours

- If TSH ≥ 25 but <150 with collection at >24 hours
- Or
- If TSH ≥ 25 but <40 with collection at >24 hours

- Immediately have patient come in for serum labs: TSH and Free T4
- Repeat Newborn Screen Filter Paper Only

Benefits of Serum Lab Testing for TSH and Free T4

- Serum TSH and Free T4 confirm the diagnosis of congenital hypothyroidism
- Tests costs approximately $90 - $200
- No risk of unsatisfactory results
- Results are returned within 24 - 48 hours

Congenital Hypothyroidism Treatment Goals

- Levothyroxine 10-15 mcg / kg / day
- 37.5 - 50 mcg
- Tablet form only

- Recommend crushing tablet and mixing with a small amount of breast milk or formula each morning

- Rose and Brown, 2006, p. 2298
Congenital Adrenal Hyperplasia

- Newborn screening for CAH is designed to diagnose patients before adrenal crisis and avoid potential death

Limitations of Newborn Screening for Congenital Adrenal Hyperplasia

- It is not always possible to determine the subtype of CAH based on screening alone
  - Genotyping after diagnosis recommended

- Many cases of the mild non-classical form will be missed
- Preterm infants have higher 17-OHP levels due to immaturity of the adrenal cortex
  - Slaughter et al. 2010, p. 912-913

Confirming Classical CAH

Abnormal Newborn Screening Results

- CAH > 150
  - Female with ambiguous genitalia
  - Or
  - Presence of hyponatremia (low Na⁺), hyperkalemia (high K⁺), and hypochloremia (low Cl⁻) in male or female

Immediate Serum 17-Hydroxyprogesterone (17-OHP) level or Serum Adrenal Profile Panel (CAH-6)

Consult with Endocrinologist Recommended

Repeating Newborn Screening Versus Serum Testing

- Repeat Newborn Screening
  - Repeat required after abnormal results
  - Can delay diagnosis if collected incorrectly
  - Takes longer to have results
  - Premature infants results less reliable
Repeating Newborn Screening Versus Serum Testing

- Serum Testing
  - Serum 17-Hydroxyprogesterone
    - Cost $50
    - Provides more accurate information for gestational age and weight

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- ACTH Stimulation testing with 0-min CAH-6b panel and 60-min CAH-6b panel
  - Cost about $790
  - Most comprehensive diagnostic information

- Serum CAH-6b Panel
  - Cost about $395
  - Improves diagnostic capabilities

Repeating Newborn Screening Versus Serum Testing

- Can follow serial results and expect results to fall < 100 as baby gets older
- Serum CAH-6b Panel
  - Cost about $395
  - Improves diagnostic capabilities

Congenital Adrenal Hyperplasia Initial Treatment Goals

- Classical Salt-Wasting CAH
  - Hydrocortisone (Cortef) 2mg/mL solution
    - 15 - 20 mg/m2/day

- Fludrocortisone 0.1mg/mL solution
  - 0.05mg (0.5mL) PO BID
  - NaCl Solution
    - 3 - 5 mEq/kg/day divided every 3 hours

- Triple dose for stress
  - Fever >101
  - Injury
  - Illness
Congenital Adrenal Hyperplasia Initial Treatment Goals

- Solu-Cortef 100mg/2mL
  - Give 25 - 50mg IM x 1 in the event of adrenal crisis

- Classical Simple – Virulizing CAH
  - Hydrocortisone (Cortef) 2mg/mL solution
    - 15 - 20 mg/m²/day

- Triple dose for stress
  - Fever >101
  - Injury
  - Illness

- Repeated vomiting / diarrhea
- Unconsciousness

Case Studies: Presentation of Congenital Hypothyroidism

Children’s of Alabama Newborn Screening Database: Data 2007-2013
**Congenital Hypothyroidism: Case Study 1**

- Female patient
  - 38 weeks gestational age
  - Birth weight 3430 grams
  - Healthy other than prolonged hyperbilirubinemia
  - Required 1 day re-admission to hospital for jaundice

**Family History**

- 3rd biological child of mother and father
- No family history of thyroid abnormalities

**Initial Newborn Screening**

- 1 day 20 hours:
  - TSH >400 ulU/mL (<25)
  - T4 1.7 mcg/dL (5.1-30)

**Serum Repeat**

- 9 days:
  - TSH 57.79 mU/mL (0.4-8.6)
  - Free T4 0.3 ng/dL (0.8-1.8)
  - Treatment Started 9 days of life
  - Levothyroxine 37.5mcg

**Repeat Newborn Screening**

- 14 days
  - TSH 52.2 ulU/mL (<25)
  - T4 14.7 mcg/dL (5.1-30)

**Diagnosis based on initial NBS**

- Treatment started at 9 days of life
- Within goal of <14 days

**Family educated on disease process and therapy**

**Thyroid will be lifelong due to absence of thyroid gland**

**Congenital Hypothyroidism: Case Study 1**

- Prolonged hyperbilirubinemia and elevated repeat NBS TSH
  - Increased Levothyroxine to 50mcg daily

**Clinic visit**

- 3 weeks old
  - TSH 1.89 ulU/mL (0.72 - 13)

**Decreased Levothyroxine to 44mcg daily**

**Thyroid ultrasound**

- 6 months of age
  - No thyroid on ultrasound

**Free T4 3.2 ng/dL (0.75 - 1.54)**

**Decreased Levothyroxine to 44mcg daily**

**Thyroid ultrasound**

- 6 months of age
  - No thyroid on ultrasound
<table>
<thead>
<tr>
<th>Congenital Hypothyroidism: Case Study 2</th>
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<tbody>
<tr>
<td><strong>• Hispanic Female Patient</strong></td>
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<tr>
<td>- 34 weeks gestational age</td>
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<tr>
<td>- Birth weight 2070 grams</td>
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<tr>
<td>- Pregnancy complicated by untreated gestational diabetes</td>
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<tr>
<td>- Mother’s first pregnancy at 27 years of age</td>
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<td><strong>• Hospitalized in the NICU for 1 month due to prematurity and feeding difficulties</strong></td>
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<tr>
<td><strong>• No family history of thyroid problems</strong></td>
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<tbody>
<tr>
<td><strong>• Initial newborn screen collected:</strong></td>
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<tr>
<td>8 hours of life</td>
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<tr>
<td>- TSH &gt;400 uIU/mL (&lt;25)</td>
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<tr>
<td>- T4 1.8 mcg/dL (5.1-30)</td>
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<tr>
<td><strong>• Repeat newborn screen collected:</strong></td>
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<tr>
<td>5 days of life</td>
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<tr>
<td>- Unsatisfactory</td>
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<tr>
<td><strong>• Screen lab repeat collected:</strong></td>
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<tr>
<td>5 days of life *diagnosis confirmed</td>
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<tr>
<td>- TSH 640uIU/mL (0.46-13.0)</td>
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<tr>
<td>- Free T4 0.14 ng/dL (0.75-1.54)</td>
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<tbody>
<tr>
<td><strong>• Repeat newborn screen collected:</strong></td>
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<tr>
<td>17 days of life</td>
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<tr>
<td>- TSH 34.2 uIU/mL (&lt;25)</td>
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<tr>
<td>- T4 14.8 mcg/dL (5.1-30)</td>
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<tr>
<td><strong>• Repeat newborn screen collected:</strong></td>
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<tr>
<td>31 days of life</td>
</tr>
<tr>
<td>- TSH &lt;3.0 uIU/mL (&lt;25)</td>
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<tr>
<td>- T4 13.7 mcg/dL (5.1-30)</td>
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Congenital Hypothyroidism: Case Study 2

- Prior to hospital discharge an appointment was made with Pediatric Endocrinology
- Mom was unclear of instructions and did not come to the appointment
- Family’s address changed from that listed on the newborn screen

Appointment was rescheduled and family was notified by letter as phone number no longer worked

- A care coordinator referral was placed
- Family no-show for second appointment

Initial appointment with Pediatric Endocrinology

- 3 months of age
- TSH 133.2 (0.36 - 8)
- Free T4 0.48 ng/dL (0.75 - 1.54)
- Thyroid ultrasound
  - No identified thyroid

- Thyroglobulin < 0.2 ng/mL
- Large anterior fontanelle
- No jaundice
- Umbilical hernia
- Slight hypotonia
- Constipation per report

Restarted Levothyroxine 37.5 mcg PO daily

- Provided education via interpreter to mother and father
- Followed labs monthly until consistently normal
Congenital Hypothyroidism: Case Study 2
- Initially diagnosed and treated within 5 days of life (<14 days)
- TSH normalized within first weeks of treatment
- Lost to follow-up
  - Phone number changed
  - Address changed

Congenital Hypothyroidism: Case Study 2
- Transportation issues
- Language barrier
- Medication stopped by primary care physician
  - No contact with PMD because this was not listed on newborn screen or identified by OSH

Congenital Hypothyroidism: Case Study 2
- Untreated for several weeks prior to restart of therapy
- Some mild clinical manifestations of hypothyroidism noted on review of systems and exam

Congenital Hypothyroidism: Case Study 3
- Caucasian Female
  - 38 weeks gestational age
  - Birth weight 3374 grams
  - Mothers 4th pregnancy at 20 years of age
  - Well-baby
    - Discharged home 1 day after birth

Congenital Hypothyroidism: Case Study 3
- Family history is negative for thyroid problems in the mother, father, and half-siblings

Serum Labs
- 44 days of age
  - TSH 181 uIU/mL (0.48-13.0)
  - Total T4 2.2 mcg/dL (5.1-13)

Repeat Newborn Screen
- Collected at 36 days of age
  - TSH 57 mIU/mL (<25)
  - T4 3.3 mcg/dL (5.1-30)

Initial Newborn Screen
- Collected at 1 day of age
  - TSH 14.9 mIU/mL (<25)
  - T4 10.2 mcg/dL (5.1-30)

Repeat Newborn Screen
- Collected at 44 days of age
  - TSH 202.6 mIU/mL (<25)
  - T4 3.3 mcg/dL (5.1-30)
**Congenital Hypothyroidism: Case Study 3**

- Diagnosis based on abnormal second newborn screen with abnormal serum labs
- Treatment initiated at 37.5 mcg PO daily
  - Day 45 of life

- Provided education packet to PMD to give to mother
- Followed with pediatric endocrinology
  - 10 weeks old
  - 4 weeks after initiation of treatment

- TSH 215.41 mIU/mL (0.46 - 8.10)
- Free T4 0.31 ng/dL (0.75 - 1.54)

- Mom reports not giving medication
  - “Because I do not believe there is anything wrong with her”
- Review of systems
  - Constipation
- Physical exam
  - Normal tone

- No hernia
- No jaundice
- Anterior fontanelle soft / flat normal size
- Posterior fontanelle closed
- Education provided

- Labs followed monthly under close supervision
  - Multiple calls to mother unanswered
  - Social services consultation for possible medical neglect
Congenital Hypothyroidism: Case Study 3
• Diagnosis delayed due to late rise in TSH
• Treatment not given due to perceived health of baby
• Developmental delay more likely due to prolonged untreated hypothyroidism

Congenital Hypothyroidism: Diagnostic Pearls
• Repeat newborn screening accounts for ~12% of diagnosed cases of primary congenital hypothyroidism
  – Shapira, 2012
• Serum labs for TSH and Free T4 are diagnostic

Congenital Hypothyroidism: Diagnostic Pearls
• TSH >10 for over 2 - 3 weeks of age is diagnostic regardless of Free T4 levels
  – Balhara, Misra, and Levitsky, 2011, p. 533
  – It is important to treat elevated TSH levels early

Congenital Hypothyroidism: Diagnostic Pearls
– Decision to stop therapy can be made later when developmental delay is less of a risk
– Parent education is critical
  – Provide education as early as possible on the importance of continued therapy

Congenital Hypothyroidism: Diagnostic Pearls
– Provide education that congenital hypothyroidism is often a “silent” diagnosis

Case Studies: Presentation of Congenital Adrenal Hyperplasia
### CAH: Case Study 1

- **Male Patient**
  - Born at 36 weeks 2 days gestational age
  - Date of birth: 11/30
  - Birth weight: 2892 grams

### CAH: Case Study 1

- **NICU**
  - Hospitalized for 1-1/2 months following delivery
  - Initially had respiratory distress, poor perfusion, hypotension, cleft palate

### CAH: Case Study 1

- Developed hyponatremia, hyperkalemia, abnormal EEG, and prolonged hemodynamic instability
  - No family history of precocious puberty, short stature, adrenal problems, infertility, etc.
  - First biological child of mother and father

### CAH: Case Study 1

- Two half brothers biologically belonging to the father
  - Half brothers healthy with no early puberty or other concerns

### CAH: Case Study 1

- **Newborn Screening History**
  - Initial NBS sent on first day of life
    - CAH 33.2 ng/mL (<45 ng/mL)
  - Repeat NBS sent on 12/7
    - 7 days of age
    - Unsatisfactory screen

- Third NBS sent on 12/18 (18 days of age) due to unsat 2nd screen
  - CAH >150 ng/mL (<25 ng/mL)
  - Serum testing (CAH-6) also sent on 12/18 due to continued clinical concern
    - Hyponatremia, hyperkalemia, and hypotension
CAH: Case Study 1

- Serum CAH-6 Screening
  - 17-Hydroxyprogesterone 39,200 ng/dL (40 - 200)
  - Cortisol <1.0 mcg/dL (2 - 11)
  - Testosterone 255 ng/dL (75 - 400)
  - 17-Hydroxypregnenolone 3,960 ng/dL (<10 - 279)
  - Progesterone 1,040 ng/dL (<10-15)

- Serum Electrolytes
  - Na 123 mmol/L (134 - 143)
  - K 5.6 mmol/L (3.5 - 5.6)

- Endocrinology consulted by NICU physician as soon as CAH-6 screen showed significant 17-OHP elevation
  - Hydrocortisone started on day of life 20

- Prolonged hyponatremia after initiation of Hydrocortisone
  - Fludrocortisone started on day of life 27
  - Scheduled NaCl supplements started on day of life 27

- Karyotype – normal 46XY male

- Initial evaluation with endocrinology 1/17

- Final diagnosis:
  - Classical Salt - Wasting CAH

- Plan
  - Send genetic evaluation for CYP21A2 gene mutations and large gene deletions when > 1 year of age
CAH: Case Study 2

- Female Patient
  - Born at 36 weeks 4 days gestational age
  - Date of birth: 7/1
  - Birth weight: 6 pounds, 11 ounces
  - Birth length: 18.75 in.

CAH: Case Study 2

- Well baby
  - Hospitalized for 2 days following delivery
  - No family history of precocious puberty, short stature, adrenal problems, infertility, etc.

CAH: Case Study 2

- Newborn Screening History
  - Initial NBS sent at 2 days of age
    - CAH 13.7 ng/mL (<45 ng/mL)
  - Repeat NBS sent on 8/8
    - 39 days of age
    - CAH 66.1 ng/mL (<25 ng/mL)

CAH: Case Study 2

- Third NBS sent on 8/22 (52 days of age) due to abnormal 2nd screen
  - CAH >150 ng/mL (<25 ng/mL)
  - Endocrinology received newborn screen results on 8/28
    - 58 days of age
    - Mother and PMD notified of results

CAH: Case Study 2

- Patient evaluated in clinic on 8/29
  - 59 days of age

CAH: Case Study 2

- Physical Exam
  - Healthy appearing 2 month old bi-racial female
  - First child of biological mother and father
  - No genital ambiguity or clitoromegaly
  - No history of illness
CAH: Case Study 2

- Lab Evaluation
  - Cortrosyn (ACTH) Stimulation Test:
    - Baseline:
      - 17-Hydroxyprogesterone 1,988 ng/dL (11 - 170)
      - Cortisol 2.3 mcg/dL (3 - 22)

- 60 Minute Stimulated (125 mcg of ACTH IM)
  - 17-Hydroxyprogesterone 20,030 ng/dL (85 - 250)
  - Cortisol 11.2 mcg/dL (27 - 50)

- Electrolytes
  - Na 141 mmol/L (134 - 143)
  - K 5.5 mmol/L (3.5 - 5.6)

- CAH-11 Urine Studies
  - Increased steroid ratios suggestive of 21-Hydroxylase Deficiency

- Presumptive Diagnosis
  - Non - Classical CAH
  - No ambiguous genitalia
  - Knew she had the ability to produce Cortisol when “stressed” although response was borderline

- Plan
  - Continue to follow closely for growth and puberty
  - Repeat Cortrosyn stimulation testing in the future due to borderline Cortisol response
  - Send genetic screening for common CYP21A2 mutations when she is a little older
CAH: Case Study 2

- Follow-up testing
  - Genetic screening
    - One gene with a large mutation called P30L, and
    - One gene with a large gene conversion referred to as the 30kb deletion

- Resulting in a non-functional gene product
  - No normal copy of CYP21A2 gene

CAH: Case Study 2

- Final diagnosis
  - Classical Simple - Virilizing CAH
  - Started Cortef at 18mg/m2/day maintenance
  - Educated on stress dosing and adrenal crisis Solu-Cortef injection

Diagnostic Pearls

- Repeat newborn screening is CRITICAL in making the diagnosis of CAH

- Serum 17-Hydroxyprogesterone levels are crucial for making the diagnosis of CAH
  - If concern over an abnormal screen in an otherwise stable premature baby, start with serum 17-OHP

Diagnostic Pearls

- If concern of hypotension, hyponatremia, and hyperkalemia or ambiguous genitalia, send the CAH-6b panel
- If unsure about diagnosis following an abnormal NBS in a stable full-term child with no other clinic concerns, consult endocrinology for Cortrosyn stimulation testing
Diagnostic Pearls

- CAH levels (17-Hydroxyprogesterone) should decrease with time
  - If increasing this could indicate an abnormality
    - New, Ghizzoni, and Lin-Su, 2009, p.235

- There will be patients with non-classical CAH who are missed by screening and identified later in life due to early growth spurt or precocious puberty

- A CAH-6b panel can be a helpful diagnostic screening tool for these patients

Long-Term Follow-Up:
The Benefits and Challenges of Continuation of Care for Congenital Hypothyroidism and Congenital Adrenal Hyperplasia

- Frequently, up to 85% of patients remain on treatment for life

- In our experience, as many as 20% of patients are lost to follow-up within first 3 years of life
  - Why?

Congenital Hypothyroidism Long-Term Care

- Education
  - Initial diagnosis education
  - Continued review of importance of daily therapy
  - Continued review of importance of lab monitoring

- Reduced stressing that some patients have that come off therapy

- Frequent labs
  - Encouraged compliance with labs every 1-2 months for first year of life
**Congenital Hypothyroidism Long-Term Care**
- Review labs every 4 months for second year of life
- Review labs every 6 months for the remainder of life
  - Appointment compliance

**Congenital Hypothyroidism Long-Term Care**
- Need to be seen by endocrinologist every 4-6 months for remainder of therapy
  - Frequent phone contact
    - Maintaining accurate phone contact information

**Congenital Hypothyroidism Long-Term Care**
- Maintaining contact with families to provide education and answer any therapy related questions

**Congenital Adrenal Hyperplasia Long-Term Care**
- Treatment is lifelong and often multidisciplinary
- Education
  - Information regarding disease process
- Information regarding therapy, often complex with medication administration every 3 hours for the first years of life
- Stress dose teaching for Hydrocortisone

**Congenital Adrenal Hyperplasia Long-Term Care**
- Information regarding adrenal crisis and demonstration of Solu-Cortef injections
- Review importance of consistent dosing and frequent lab monitoring
  - Lab monitoring
**Congenital Adrenal Hyperplasia Long-Term Care**

- Monitor 17-OHP, Na, K, Renin frequently for the duration of therapy

  - Appointment compliance

    - Need to be followed by pediatric endocrinologist every 3-6 months

- Will need to transition to adult care at 18 years of age

  - Frequent phone contact

    - Contact maintained with families to answer any questions, review stress dosing as needed, etc.

**Support services**
- Many families seek out support services due to rarity of condition and complex nature of disease process

**Frequently Asked Questions: Endocrinology Newborn Screening**

**When should I do serum labs?**

- If newborn screen is abnormal, a serum TSH and Free T4 will be diagnostic for congenital hypothyroidism

**Frequently Asked Questions Congenital Hypothyroidism**

- What if the TSH is elevated but the Free T4 is normal?

  - You can recheck the TSH and Free T4 in 1-2 weeks, if this trend continues with TSH >10 for more than 2 weeks, we would recommend treatment with Levothyroxine
Frequently Asked Questions

**Congenital Hypothyroidism**

- If I send serum labs and they have normal results, do I need to recheck them?
  - No, if you have a normal TSH and Free T4 it is unlikely that it will become abnormal

- However, you should send repeat newborn screening as recommended by the ADPH

**Congenital Hypothyroidism**

- I have a patient you see for congenital hypothyroidism. Should I alter the Levothyroxine dosing based on labs done at my clinic?
  - No, please just ensure that all serum labs are faxed to our offices and we will change the Levothyroxine dose as indicated
  - You can always call our offices if you have any clinical concerns regarding a mutual patient

**Congenital Adrenal Hyperplasia**

- What should I do with abnormal CAH levels in a premature infant?
  - In an otherwise stable premature baby send serum 17-OHP levels
  - If elevated, may contact our offices to discuss

- Likely, we will ask that you follow these levels every 1-2 weeks to follow trends
Frequently Asked Questions
Congenital Adrenal Hyperplasia

- Should education be provided on stress dosing and Solu-Cortef before hospital discharge?
  - Yes, please contact our offices for education materials

- When is a Cortrosyn Stimulation Test indicated?
  - When the CAH levels are elevated in an otherwise stable, full-term infant with normal sodium and potassium levels
  - Borderline results in premature infants

- When should I send the CAH-6b panel?
  - If you are concerned about an infant with hyponatremia, hyperkalemia, hypotension, or ambiguous genitalia send the CAH6-b panel as soon as possible regardless of CAH newborn screening

- If you can provide general information for parents we will discuss this in more detail at the initial clinic visit

- Also, in a young child with premature adrenarche / puberty, when there is a question of non-classical CAH
Children’s of Alabama: Endocrinology Newborn Screening Resources

Newborn Screening Resources

- Patient Education available online
  - https://www.childrensal.org/NewbornScreening
  - Congenital Hypothyroidism
    “Parents Guide”
  - CAH-CARES Foundations

Newborn Screening Resources

- Patient Education Packets can be mailed or faxed to PMD office or patient directly
- Consultation available anytime for clinical questions or concerns:
  - 205 - 996 - 9166 or 205 - 638 - 9107
  - newbornscreening@peds.uab.edu

Newborn Screening Resources

- If parent has questions prior to appointment or if social services are needed for appointment, the parent may contact our offices at 205 - 996 - 9166
- We encourage PMDs to ask families if social services are needed

Newborn Screening Resources

- Working to develop video education material for congenital hypothyroidism and congenital adrenal hyperplasia, including stress dosing and Adrenal Crisis / Solu-Cortef teaching

Thank You So Much For Your Time