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

Satellite Conference and Live Webcast
Thursday, November 14, 2013
2:00 – 3:30 p.m. Central Time

Produced by the Alabama Department of Public Health
Video Communications and Distance Learning Division

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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

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

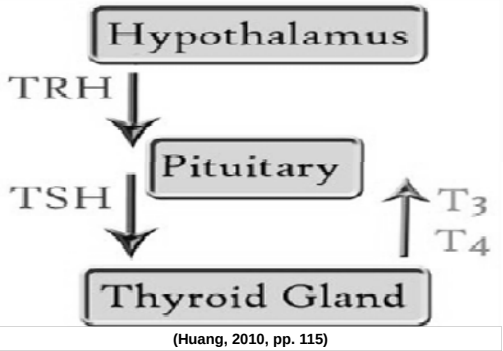
Congenital Hypothyroidism

- 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

Congenital Hypothyroidism

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

Congenital Hypothyroidism



(Huang, 2010, pp. 115)

Congenital Hypothyroidism

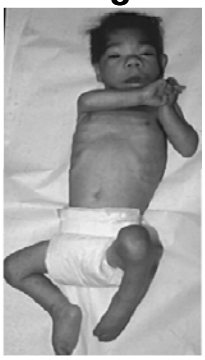
- Optimal thyroid hormone levels are critical for normal neurodevelopment
- Untreated congenital hypothyroidism can produce profound somatic and neurologic delay

– Albert, et al., 2013, p. 36-64

Congenital Hypothyroidism

- 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 Hypothyroidism



- 17 year old female with untreated congenital hypothyroidis
- Average height of a 3 year old and estimated bone age of 9 months
  - From Brent, G., Davies, T., Larsen, P., 2007

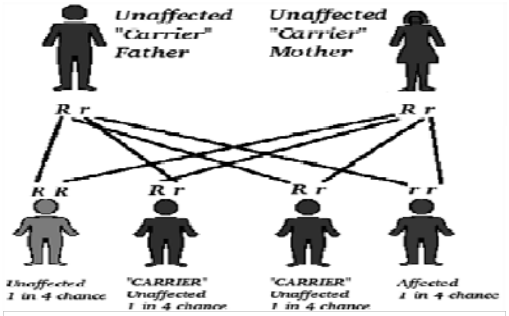
Congenital Adrenal Hyperplasia 21-Hydroxylase Deficiency

- 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

Congenital Adrenal Hyperplasia 21-Hydroxylase Deficiency

- Reported incidence ranges from 1:5,000 – 1:15,000
- Clinical presentation varies from mild to severe

Congenital Adrenal Hyperplasia 21-Hydroxylase Deficiency

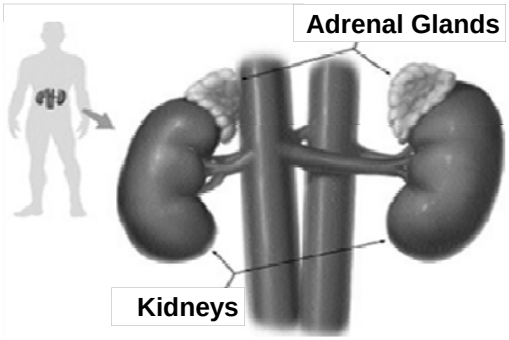


Donohoue, 2010, p. 153-160

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



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

Classical Salt-Wasting CAH 21- Hydroxylase Deficiency

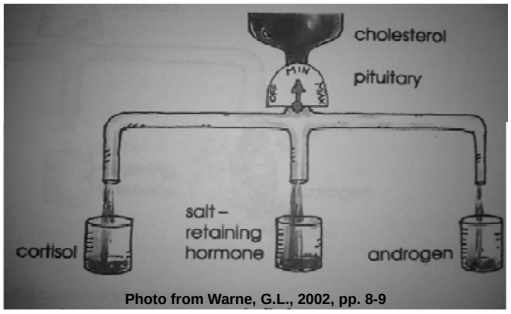


Photo from Warne, G.L., 2002, pp. 8-9

Classical Salt-Wasting CAH 21- Hydroxylase Deficiency

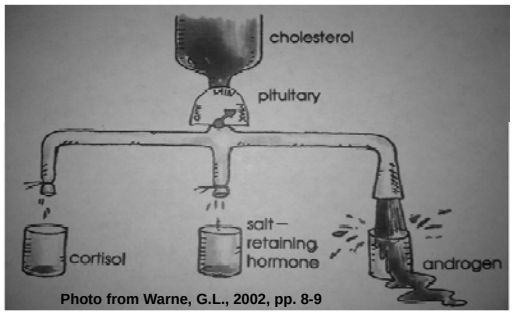


Photo from Warne, G.L., 2002, pp. 8-9

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

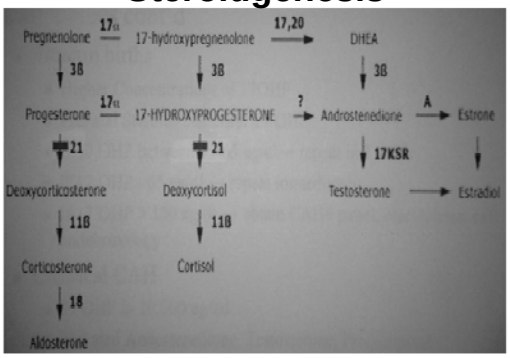
Simple Virulizing CAH 21-Hydroxylase Deficiency

- 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

Steroidgenesis



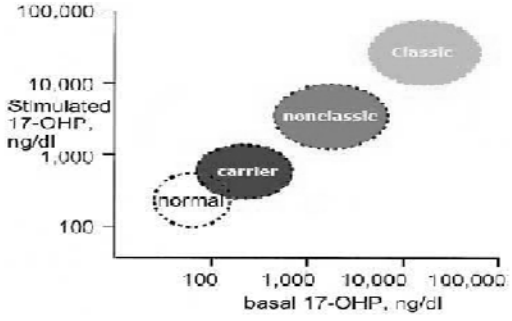
Attenuated or Non-classical CAH

- 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

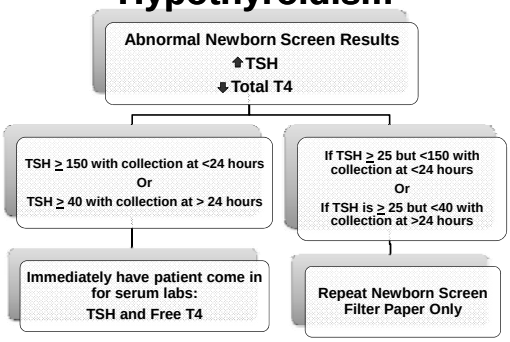


Making the Diagnosis: Congenital Hypothyroidism and Congenital Adrenal Hyperplasia

Congenital Hypothyroidism

- 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



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

Congenital Hypothyroidism Treatment Goals

- 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

Congenital Adrenal Hyperplasia

- Major benefit is identification of males
- Moderately high rate of false positives in premature infants
  - New, Ghizzoni, and Lin-Su, 2009, p.235
- Occasional false negatives with mild variants

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**

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

Limitations of Newborn Screening for Congenital Adrenal Hyperplasia

- 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

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

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

Repeating Newborn Screening Versus Serum Testing

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

Congenital Adrenal Hyperplasia Initial Treatment Goals

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

Congenital Adrenal Hyperplasia Initial Treatment Goals

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

Congenital Adrenal Hyperplasia Initial Treatment Goals

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

Congenital Adrenal Hyperplasia Initial Treatment Goals

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

Congenital Adrenal Hyperplasia Initial Treatment Goals

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

Congenital Adrenal Hyperplasia Initial Treatment Goals

- 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
  - Surgery

Congenital Adrenal Hyperplasia Initial Treatment Goals

- Repeated vomiting / diarrhea
- Unconsciousness

Case Studies: Presentation of Congenital Hypothyroidism

Children's of Alabama Newborn Screening Database: Data 2007-2013

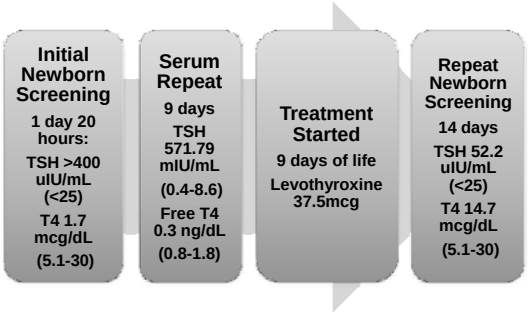
Congenital Hypothyroidism: Case Study 1

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

Congenital Hypothyroidism: Case Study 1

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

Congenital Hypothyroidism: Case Study 1



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 uIU/mL (0.72 - 13)

Congenital Hypothyroidism: Case Study 1

  - 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

Congenital Hypothyroidism: Case Study 1

- Diagnosis based on initial NBS
- Treatment started at 9 days of life
  - Within goal of <14 days
- Family educated on disease process and therapy
- Therapy will be lifelong due to absence of thyroid gland

Congenital Hypothyroidism: Case Study 2

- Hispanic Female Patient
  - 34 weeks gestational age
  - Birth weight 2070 grams
  - Pregnancy complicated by untreated gestational diabetes
  - Mother's first pregnancy at 27 years of age

Congenital Hypothyroidism: Case Study 2

  - Hospitalized in the NICU for 1 month due to prematurity and feeding difficulties
- No family history of thyroid problems

Congenital Hypothyroidism: Case Study 2

- Initial newborn screen collected: 8 hours of life
  - TSH >400 uIU/mL (<25)
  - T4 1.8 mcg/dL (5.1-30)
- Repeat newborn screen collected: 5 days of life
  - Unsatisfactory

Congenital Hypothyroidism: Case Study 2

- Screen lab repeat collected: 5 days of life *diagnosis confirmed
  - TSH 640uIU/mL (0.46- 13.0)
  - Free T4 0.14 ng/dL (0.75-1.54)

Congenital Hypothyroidism: Case Study 2

- Repeat newborn screen collected: 17 days of life
  - TSH 34.2 uIU/mL (<25)
  - T4 14.8 mcg/dL (5.1-30)

Congenital Hypothyroidism: Case Study 2

- Repeat newborn screen collected: 31 days of life
  - TSH <3.0 uIU/mL (<25)
  - T4 13.7 mcg/dL (5.1-30)

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

Congenital Hypothyroidism: Case Study 2

- 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

Congenital Hypothyroidism: Case Study 2

- Primary care physician saw patient in the interim and discontinued Levothyroxine due to suppressed TSH
- Transportation needs were arranged and patient came for follow-up at 3 months of age

Congenital Hypothyroidism: Case Study 2

- 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

Congenital Hypothyroidism: Case Study 2

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

Congenital Hypothyroidism: Case Study 2

- 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

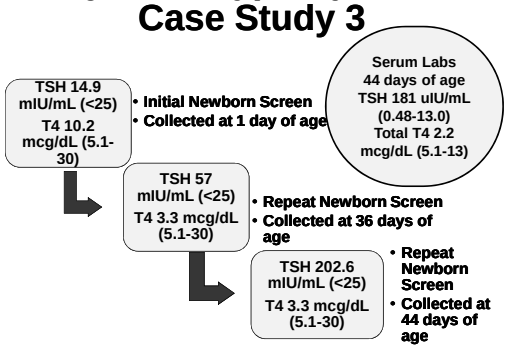
Congenital Hypothyroidism: Case Study 3

- 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

Congenital Hypothyroidism: Case Study 3



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

Congenital Hypothyroidism: Case Study 3

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

Congenital Hypothyroidism: Case Study 3

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

Congenital Hypothyroidism: Case Study 3

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

Congenital Hypothyroidism: Case Study 3

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

Congenital Hypothyroidism: Case Study 3

- 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

CAH: Case Study 1

  - 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

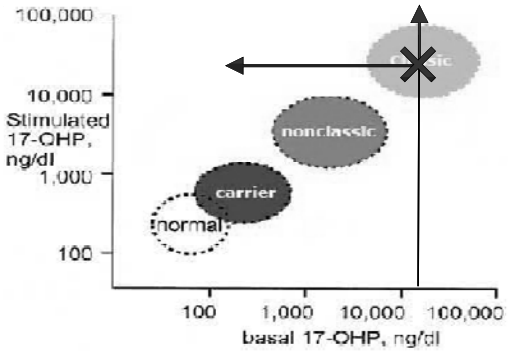
CAH: Case Study 1

- 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)

CAH: Case Study 1

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

CAH: Case Study 1



CAH: Case Study 1

- 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

CAH: Case Study 1

- 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

CAH: Case Study 1

- 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)

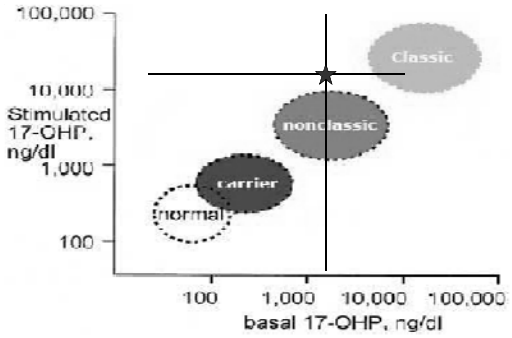
CAH: Case Study 2

- 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)

CAH: Case Study 2

- K 5.5 mmol/L (3.5 - 5.6)
- CAH-11 Urine Studies
  - Increased steroid ratios suggestive of 21-Hydroxylase Deficiency

CAH: Case Study 2



CAH: Case Study 2

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

CAH: Case Study 2

- 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

CAH: Case Study 2

- 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

Diagnostic Pearls

- 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

Diagnostic Pearls

- 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

Congenital Hypothyroidism Long-Term Care

- 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

Congenital Hypothyroidism Long-Term Care

  - 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

Congenital Adrenal Hyperplasia Long-Term Care

- 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

Congenital Adrenal Hyperplasia Long-Term Care

- 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.

Congenital Adrenal Hyperplasia Long-Term Care

- 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

Frequently Asked Questions Congenital Hypothyroidism

- 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***

Frequently Asked Questions Congenital Hypothyroidism

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

Frequently Asked Questions Congenital Hypothyroidism

- **I have a patient you see for congenital hypothyroidism. Should I alter the Levothyroxine dosing based on labs done at my clinic?**

Frequently Asked Questions Congenital Hypothyroidism

  - ***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***

Frequently Asked Questions 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***

Frequently Asked Questions Congenital Adrenal Hyperplasia

  - ***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***

Frequently Asked Questions Congenital Adrenal Hyperplasia

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

Frequently Asked Questions Congenital Adrenal Hyperplasia

- **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***

Frequently Asked Questions Congenital Adrenal Hyperplasia

- ***Also, in a young child with premature adrenarche / puberty, when there is a question of non-classical CAH***

Frequently Asked Questions Congenital Adrenal Hyperplasia

- **When should I send the CAH-6b panel?**

Frequently Asked Questions Congenital Adrenal Hyperplasia

- ***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***

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