NCC Pediatrics Continuity Clinic Curriculum:

Health Maintenance I:

Faculty Guide

verall Goal

To understand the recommended screening procedures utilized in the health maintenance of children.

Overall Objectives:

Over the next two modules, residents will become familiar with the AAP Recommendations for Preventive Pediatric Health Care, as outlined in the 2021 Periodicity Schedule*(See next page for the entire schedule)*.

The Periodicity Schedule includes the following screening procedures, some of which will be covered over the next two modules, and some of which will be covered in upcoming continuity modules:

1 Measurements:
   - ***a.*** **Length, Height, Weight, HC, BMI:** ***Health Maintenance I***
   - ***b.*** Blood Pressure

2 Sensory Screening:
   - *a.* Vision: *Health Maintenance III*
   - *b.* Hearing: *Health Maintenance III*

   Developmental/Behavioral Assessment:
   Development I-V & Behavior I-II

   Procedures:
   - ***a.*** **Newborn Metabolic/Hemoglobin:** ***Health Maintenance I***
   - ***b.*** Immunization: *Immunizations Module*
   - ***c.*** Hematocrit or Hemoglobin: *Health Maintenance II (next week)*
   - ***d.*** Lead: *Health Maintenance II (next week)*
   - ***e.*** Tuberculosis: *Health Maintenance II (next week)*
   - ***f.*** Dyslipidemia: *Health Maintenance II (next week)*
   - ***g.*** STIs: *Adolescence IV*

   Oral Health: *Dental Health Modules*

NCC Pediatrics Continuity Clinic Curriculum:
Health Maintenance I:
Faculty Guide

Pre-Meeting Preparation:

Please read the following enclosures, corresponding to the listed screening procedures:

1 Measurements: L/H, Wt, HC, BMI:
- **CDC Growth Chart Training Modules: Do the 1st & 2nd modules only!** *("Using the WHO Growth Charts" & "Overview of the CDC Growth Charts")*
- "Rules of Thumb for Growth" *(p. 4)*
- **Homework:** Print out an AHLTA growth chart + data table for a toddler ≤2yr you've seen in the past month. Bring in for group activity. *(If you haven't seen toddlers, print out an interesting AHLTA growth chart from one of your patients).*

2 Procedures: Newborn Metabolic/Hemoglobin:
- "Newborn Metabolic Screening: FAQs" *(pgs. 5)*
- State of Maryland: list of screened conditions *(pg. 6)*
- ACT sheets for hypothyroidism, PKU, & hemoglobinopathies *(pgs. 7-9—skim!)*
- **Homework:** Pick one of the other conditions on the Maryland NBS. Go to the ACT Website and print out the associated fact sheet for group activity.

Conference Agenda:

- *Review* "Health Maintenance I Quiz"
- Complete "Health Maintenance I Cases"
- **Group Activities:**
  - Growth Chart: **iscuss the benefits and pitfalls of using AHLTA charts. emonstrate ho to copy and paste charts, adjust for prematurity, and modify scale.**
    - *Those residents who brought non-toddler growth charts should share their observations with the group. Remind them that we will discuss VLBW and BMI-for-age charts in the NICU and Nutrition IV modules, respectively.*
  - ACT Sheets: Go around the table, and have residents answer the following questions: What is the **underlying cause** of your condition? What **immediate actions** do you need to take? What is the **consequence** of delayed treatment?

Extra-Credit:

- Growth Charts: WHO; CDC; VLBW; Special Needs
- Newborn Screening Fact Sheets *(AAP Technical Report, 2006)*

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN®

Recommendations for Preventive Pediatric Health Care

Bright Futures/American Academy of Pediatrics

Bright Futures
prevention and health promotion for infants, children, adolescents, and their families™

Each child and family is unique; therefore, these Recommendations for Preventive Pediatric Health Care are designed for the care of children who are receiving competent parenting, have no manifestations of any important health problems, and are growing and developing in a satisfactory fashion. Developmental, psychosocial, and chronic disease issues for children and adolescents may require frequent counseling and treatment visits separate from preventive care visits. Additional visits also may become necessary if circumstances suggest variations from normal.

These recommendations represent a consensus by the American Academy of Pediatrics (AAP) and Bright Futures. The AAP continues to emphasize the great importance of continuity of care in comprehensive health supervision and the need to avoid fragmentation of care.

Refer to the specific guidance by age as listed in the *Bright Futures Guidelines* (Hagan JF, Shaw JS, Duncan PM, eds. *Bright Futures: Guidelines for Health Supervision of Infants, Children, and Adolescents.* 4th ed. American Academy of Pediatrics; 2017).

The recommendations in this statement do not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

The Bright Futures/American Academy of Pediatrics Recommendations for Preventive Pediatric Health Care are updated annually.

	INFANCY								EARLY CHILDHOOD							MIDDLE CHILDHOOD						ADOLESCENCE										
AGE[1]	**Prenatal**[2]	**Newborn**[3]	**3-5 d**[4]	**By 1 mo**	**2 mo**	**4 mo**	**6 mo**	**9 mo**	**12 mo**	**15 mo**	**18 mo**	**24 mo**	**30 mo**	**3 y**	**4 y**	**5 y**	**6 y**	**7 y**	**8 y**	**9 y**	**10 y**	**11 y**	**12 y**	**13 y**	**14 y**	**15 y**	**16 y**	**17 y**	**18 y**	**19 y**	**20 y**	**21 y**
HISTORY Initial/Interval	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
MEASUREMENTS																															
Length/Height and Weight		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Head Circumference		●	●	●	●	●	●	●	●	●	●	●																			
Weight for Length		●	●	●	●	●	●	●	●	●	●																				
Body Mass Index[5]												●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Blood Pressure[6]		★	★	★	★	★	★	★	★	★	★	★	★	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
SENSORY SCREENING																															
Vision[7]		★	★	★	★	★	★	★	★	★	★	★	★	●	●	●	●	★	●	★	●	★	●	★	★	●	★	★	★	★	★
Hearing		●[8]	●[9] →			★	★	★	★	★	★	★	★	★	●	●	●	★	●	★	●	←		●[10]	→	←		●	→	←		● →
DEVELOPMENTAL/BEHAVIORAL HEALTH																															
Developmental Screening[11]								●			●		●																		
Autism Spectrum Disorder Screening[12]											●	●																			
Developmental Surveillance		●	●	●	●	●	●		●	●		●		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Psychosocial/Behavioral Assessment[13]		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Tobacco, Alcohol, or Drug Use Assessment[14]																						★	★	★	★	★	★	★	★	★	★
Depression Screening[15]																							●	●	●	●	●	●	●	●	●
Maternal Depression Screening[16]				●	●	●	●																								
PHYSICAL EXAMINATION[17]		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
PROCEDURES[18]																															
Newborn Blood		●[19]	●[20] →																												
Newborn Bilirubin[21]		●																													
Critical Congenital Heart Defect[22]		●																													
Immunization[23]		●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●
Anemia[24]						★			●	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★
Lead[25]							★	★	● or ★[26]		★	● or ★[26]		★	★	★	★															
Tuberculosis[27]				★			★		★			★		★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★
Dyslipidemia[28]												★			★		★		★	←	● →	★	★	★	★	★	←		●	→		
Sexually Transmitted Infections[29]																						★	★	★	★	★	★	★	★	★	★
HIV[30]																						★	★	★	★	←		● →	★	★	★
Hepatitis C Virus Infection[31]																													●	→	
Cervical Dysplasia[32]																															●
ORAL HEALTH[33]							●[34]	●[34]	★		★	★	★	★	★	★	★														
Fluoride Varnish[35]							←				●		→																		
Fluoride Supplementation[36]							★	★	★		★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★	★				
ANTICIPATORY GUIDANCE	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●

1. If a child comes under care for the first time at any point on the schedule, or if any items are not accomplished at the suggested age, the schedule should be brought up to date at the earliest possible time.
2. A prenatal visit is recommended for parents who are at high risk, for first-time parents, and for those who request a conference. The prenatal visit should include anticipatory guidance, pertinent medical history, and a discussion of benefits of breastfeeding and planned method of feeding, per "The Prenatal Visit" (https://pediatrics.aappublications.org/content/142/1/e20181218).
3. Newborns should have an evaluation after birth, and breastfeeding should be encouraged (and instruction and support should be offered).
4. Newborns should have an evaluation within 3 to 5 days of birth and within 48 to 72 hours after discharge from the hospital to include evaluation for feeding and jaundice. Breastfeeding newborns should receive formal breastfeeding evaluation, and their mothers should receive encouragement and instruction, as recommended in "Breastfeeding and the Use of Human Milk" (http://pediatrics.aappublications.org/content/129/3/e827.full). Newborns discharged less than 48 hours after delivery must be examined within 48 hours of discharge, per "Hospital Stay for Healthy Term Newborns" (http://pediatrics.aappublications.org/content/125/2/405.full).
5. Screen, per "Expert Committee Recommendations Regarding the Prevention, Assessment, and Treatment of Child and Adolescent Overweight and Obesity: Summary Report" (http://pediatrics.aappublications.org/content/120/Supplement_4/S164.full)
6. Screening should occur per "Clinical Practice Guideline for Screening and Management of High Blood Pressure in Children and Adolescents" (http://pediatrics.aappublications.org/content/140/3/e20171904). Blood pressure measurement in infants and children with specific risk conditions should be performed at visits before age 3 years.
7. A visual acuity screen is recommended at ages 4 and 5 years, as well as in cooperative 3-year-olds. Instrument-based screening may be used to assess risk at ages 12 and 24 months, in addition to the well visits at 3 through 5 years of age. See "Visual System Assessment in Infants, Children, and Young Adults by Pediatricians" (http://pediatrics.aappublications.org/content/137/1/e20153596) and "Procedures for the Evaluation of the Visual System by Pediatricians" (http://pediatrics.aappublications.org/content/137/1/e20153597).
8. Confirm initial screen was completed, verify results, and follow up, as appropriate. Newborns should be screened, per "Year 2007 Position Statement: Principles and Guidelines for Early Hearing Detection and Intervention Programs" (http://pediatrics.aappublications.org/content/120/4/898.full).
9. Verify results as soon as possible, and follow up, as appropriate.
10. Screen with audiometry including 6,000 and 8,000 Hz high frequencies once between 11 and 14 years, once between 15 and 17 years, and once between 18 and 21 years. See "The Sensitivity of Adolescent Hearing Screens Significantly Improves by Adding High Frequencies" (https://www.sciencedirect.com/science/article/abs/pii/S1054139X16000483).
11. Screening should occur per "Promoting Optimal Development: Identifying Infants and Young Children With Developmental Disorders Through Developmental Surveillance and Screening" (https://pediatrics.aappublications.org/content/145/1/e20193449).
12. Screening should occur per "Identification, Evaluation, and Management of Children With Autism Spectrum Disorder" (https://pediatrics.aappublications.org/content/145/1/e20193447).
13. This assessment should be family centered and may include an assessment of child social-emotional health, caregiver depression, and social determinants of health. See "Promoting Optimal Development: Screening for Behavioral and Emotional Problems" (http://pediatrics.aappublications.org/content/135/2/384) and "Poverty and Child Health in the United States" (http://pediatrics.aappublications.org/content/137/4/e20160339).
14. A recommended assessment tool is available at http://crafft.org.
15. Recommended screening using the Patient Health Questionnaire (PHQ)-2 or other tools available in the GLAD-PC toolkit and at https://downloads.aap.org/AAP/PDF/Mental_Health_Tools_for_Pediatrics.pdf.
16. Screening should occur per "Incorporating Recognition and Management of Perinatal Depression Into Pediatric Practice" (https://pediatrics.aappublications.org/content/143/1/e20183259).
17. At each visit, age-appropriate physical examination is essential, with infant totally unclothed and older children undressed and suitably draped. See "Use of Chaperones During the Physical Examination of the Pediatric Patient" (http://pediatrics.aappublications.org/content/127/5/991.full).
18. These may be modified, depending on entry point into schedule and individual need.
19. Confirm initial screen was accomplished, verify results, and follow up, as appropriate. The Recommended Uniform Screening Panel (https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html), as determined by The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, and state newborn screening laws/regulations (https://www.babysfirsttest.org/newborn-screening/states) establish the criteria for and coverage of newborn screening procedures and programs.

(continued)

KEY: ● = to be performed ★ = risk assessment to be performed with appropriate action to follow, if positive ← ● → = range during which a service may be provided

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(continued)

20. Verify results as soon as possible, and follow up, as appropriate.
21. Confirm initial screening was accomplished, verify results, and follow up, as appropriate. See "Hyperbilirubinemia in the Newborn Infant ≥35 Weeks' Gestation: An Update With Clarifications" (http://pediatrics.aappublications.org/content/124/4/1193).
22. Screening for critical congenital heart disease using pulse oximetry should be performed in newborns, after 24 hours of age, before discharge from the hospital, per "Endorsement of Health and Human Services Recommendation for Pulse Oximetry Screening for Critical Congenital Heart Disease" (http://pediatrics.aappublications.org/content/129/1/190.full).
23. Schedules, per the AAP Committee on Infectious Diseases, are available at https://redbook.solutions.aap.org/SS/immunization_Schedules.aspx. Every visit should be an opportunity to update and complete a child's immunizations.
24. Perform risk assessment or screening, as appropriate, per recommendations in the current edition of the AAP *Pediatric Nutrition: Policy of the American Academy of Pediatrics* (Iron chapter).
25. For children at risk of lead exposure, see "Prevention of Childhood Lead Toxicity" (http://pediatrics.aappublications.org/content/138/1/e20161493) and "Low Level Lead Exposure Harms Children: A Renewed Call for Primary Prevention" (http://www.cdc.gov/nceh/lead/ACCLPP/Final_Document_030712.pdf).
26. Perform risk assessments or screenings as appropriate, based on universal screening requirements for patients with Medicaid or in high prevalence areas.
27. Tuberculosis testing per recommendations of the AAP Committee on Infectious Diseases, published in the current edition of the AAP *Red Book: Report of the Committee on Infectious Diseases.* Testing should be performed on recognition of high-risk factors.
28. See "Integrated Guidelines for Cardiovascular Health and Risk Reduction in Children and Adolescents" (http://www.nhlbi.nih.gov/guidelines/cvd_ped/index.htm).
29. Adolescents should be screened for sexually transmitted infections (STIs) per recommendations in the current edition of the AAP *Red Book: Report of the Committee on Infectious Diseases.*
30. Adolescents should be screened for HIV according to the US Preventive Services Task Force (USPSTF) recommendations (https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/human-immunodeficiency-virus-hiv-infection-screening) once between the ages of 15 and 18, making every effort to preserve confidentiality of the adolescent. Those at increased risk of HIV infection, including those who are sexually active, participate in injection drug use, or are being tested for other STIs, should be tested for HIV and reassessed annually.
31. All individuals should be screened for hepatitis C virus (HCV) infection according to the USPSTF (https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/hepatitis-c-screening) and Centers for Disease Control and Prevention (CDC) recommendations (https://www.cdc.gov/mmwr/volumes/69/rr/rr6902a1.htm) at least once between the ages of 18 and 79. Those at increased risk of HCV infection, including those who are persons with past or current injection drug use, should be tested for HCV infection and reassessed annually.
32. See USPSTF recommendations (https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/cervical-cancer-screening). Indications for pelvic examinations prior to age 21 are noted in "Gynecologic Examination for Adolescents in the Pediatric Office Setting" (http://pediatrics.aappublications.org/content/126/3/583.full).
33. Assess whether the child has a dental home. If no dental home is identified, perform a risk assessment (https://www.aap.org/en-us/advocacy-and-policy/aap-health-initiatives/Oral-Health/Pages/Oral-Health-Practice-Tools.aspx) and refer to a dental home. Recommend brushing with fluoride toothpaste in the proper dosage for age. See "Maintaining and Improving the Oral Health of Young Children" (http://pediatrics.aappublications.org/content/134/6/1224).
34. Perform a risk assessment (https://www.aap.org/en-us/advocacy-and-policy/aap-health-initiatives/Oral-Health/Pages/Oral-Health-Practice-Tools.aspx). See "Maintaining and Improving the Oral Health of Young Children" (http://pediatrics.aappublications.org/content/134/6/1224).
35. See USPSTF recommendations (https://www.uspreventiveservicestaskforce.org/Page/Document/UpdateSummaryFinal/dental-caries-in-children-from-birth-through-age-5-years-screening). Once teeth are present, fluoride varnish may be applied to all children every 3 to 6 months in the primary care or dental office. Indications for fluoride use are noted in "Fluoride Use in Caries Prevention in the Primary Care Setting" (http://pediatrics.aappublications.org/content/134/3/626).
36. If primary water source is deficient in fluoride, consider oral fluoride supplementation. See "Fluoride Use in Caries Prevention in the Primary Care Setting" (http://pediatrics.aappublications.org/content/134/3/626).

Summary of Changes Made to the Bright Futures/AAP Recommendations for Preventive Pediatric Health Care (Periodicity Schedule)

This schedule reflects changes approved in November 2020 and published in March 2021. For updates and a list of previous changes made, visit www.aap.org/periodicityschedule.

CHANGES MADE IN NOVEMBER 2020

DEVELOPMENTAL

- Footnote 11 has been updated to read as follows: "Screening should occur per 'Promoting Optimal Development: Identifying Infant and Young Children With Developmental Disorders Through Developmental Surveillance and Screening' (https://pediatrics.aappublications.org/content/145/1/e20193449)."

AUTISM SPECTRUM DISORDER

- Footnote 12 has been updated to read as follows: "Screening should occur per 'Identification, Evaluation, and Management of Children With Autism Spectrum Disorder' (https://pediatrics.aappublications.org/content/145/1/e20193447)."

HEPATITIS C VIRUS INFECTION

- Screening for hepatitis C virus infection has been added to occur at least once between the ages of 18 and 79 years (to be consistent with recommendations of the USPSTF and CDC).
- Footnote 31 has been added to read as follows: "All individuals should be screened for hepatitis C virus (HCV) infection according to the USPSTF (https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/hepatitis-c-screening) and Centers for Disease Control and Prevention (CDC) recommendations (https://www.cdc.gov/mmwr/volumes/69/rr/rr6902a1.htm) at least once between the ages of 18 and 79. Those at increased risk of HCV infection, including those who are persons with past or current injection drug use, should be tested for HCV infection and reassessed annually."
- Footnotes 31 through 35 have been renumbered as footnotes 32 through 36.

CHANGES MADE IN OCTOBER 2019

MATERNAL DEPRESSION

- Footnote 16 has been updated to read as follows: "Screening should occur per 'Incorporating Recognition and Management of Perinatal Depression Into Pediatric Practice' (https://pediatrics.aappublications.org/content/143/1/e20183259)."

CHANGES MADE IN DECEMBER 2018

BLOOD PRESSURE

- Footnote 6 has been updated to read as follows: "Screening should occur per 'Clinical Practice Guideline for Screening and Management of High Blood Pressure in Children and Adolescents' (http://pediatrics.aappublications.org/content/140/3/e20171904). Blood pressure measurement in infants and children with specific risk conditions should be performed at visits before age 3 years."

ANEMIA

- Footnote 24 has been updated to read as follows: "Perform risk assessment or screening, as appropriate, per recommendations in the current edition of the AAP *Pediatric Nutrition: Policy of the American Academy of Pediatrics* (Iron chapter)."

LEAD

- Footnote 25 has been updated to read as follows: "For children at risk of lead exposure, see 'Prevention of Childhood Lead Toxicity' (http://pediatrics.aappublications.org/content /138/1/e20161493) and 'Low Level Lead Exposure Harms Children: A Renewed Call for Primary Prevention' (https://www.cdc.gov/nceh/lead/ACCLPP/Final_Document_030712.pdf)."

HRSA
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Rules of Thumb for Growth

Weight: Average infant birth weight is 7 lbs (~3.2kg)

1. Weight loss in 1st few days—5-10% of birthweight.
2. Return to birthweight @ 7-10 days
3. Double birthweight @ 4 months
4. Triple birthweight @ 1year
5. Quadruple birthweight @ 2 years

Expected Weight Increase:

1. 20-30g/day from birth-3 months
2. 15-20g/day from 3-6 months
3. 5-10g/day from 6-12 months
4. 250g/month (0.5-1 lbs/month) from 1-2 years
5. 2.3kg/year (4 lbs/year) from 2 years - adolescence

Height: Average length at birth is 20 inches (~51 cm)

1. Birth length increases by 50% at 12 months.
2. Doubles by age 4 years.
3. Triples by age 13 years.

Expected Height Increase:

1. 10 inches or 25 cm from birth- 12 months
2. 5 inches or 12.5 cm from 13- 24 months
3. 2.5 inches or 6.25 cm from 2 years - adolescence

Head Circumference (Occipital Frontal Circumference):

Average OFC at birth is 35 cm (25% of adult)

1. 0.5 cm/week from 0 – 2 months
2. 0.25 cm/week from 2 - 6 months
3. 1 cm/mo from 0-12 months (i.e. 12 cm increase since birth)

➔ **Rule of 3s & 9s:**
Mean OFC increases 5 cm from 0-3 mo, 3-9 mo, 9 mo-3 yrs, and 3-9 yrs

Growth Chart Trends

Plotting a child's growth at different ages and seeing if he follows a curve is more important than where he is at any one time. Even if a child is at the 5th %ile for his weight, if he has always been at the 5th %ile, then he is likely growing normally. It would be concerning if he had previously been at the 50th or 75th %ile and had now fallen down to the 5th %ile.

☀ Also remember that children between the ages of **6 and 24 months** can normally move up or down on their percentiles, often crossing one or two percentile ba ds—this is called **rechanneling**. Older children, however, should follow their growth curve fairly closely, and **flattening curves are always abnormal.**

Newborn Metabolic Screening

What are we testing for?

The State of Maryland uses the Maryland Health Department to process and analyze newborn screens. This panel tests for _ 1 disorders—the majority by tandem mass spectrometry and some by other technologies. *See the next page for a list of disorders by category.*

Is parental permission required?

Yes. Maryland requires that parents give permission before newborn screening tests are done. Permission forms are given to parents by the MICC nurses. Signed forms are in the paper record.

When should newborns be tested?

- Each baby should be tested **twice**.
- The **first test** is usually done shortly before the newborn baby leaves the hospital. For the most reliable results, it should be done after the baby has received at least 24 hours of breast or formula feedings. A screen collected <24hrs is not valid for several conditions on the panel (e.g. 10% of congenital hypothyroidism is identified on the 2nd screen).
- The **second test** should be done when the baby is >7 days old; ideally between 10 days and 2 weeks. At WRNMMC, we routinely obtain a second blood sample at the 2 week follow-up, even if the first test was normal. The second screen picks up several children each year with conditions that are not detectable on the initial screen. This is particularly important for cystic fibrosis: in 2006, Maryland implemented the IRT/IRT system, which means that 2 elevated IRT levels are needed to determine increased risk for CF.
- Dr. Greenwald is the POC for newborn screens performed at WR-B. (Results will be available to him a d sted a d e el s read s eet prior to making it into A S for review)

Where can I find more information about an abnormal test result?

The American College of Medical Genetics has a website that details many metabolic disorders for which screening is taking place. These **"ACT" sheets** contain information for providers on how to manage abnormal screening tests.

⇨ Click the link, and you will then see a table in which disorders are broken down by the following categories: endocrine, hemoglobin, genetic, galactosemias, fatty acid oxidation, organic and amino acidemias, immunodeficiency, and lysosomal storage.

Examples of ACT sheets for hypothyroidism, PKU, and hemoglobinopathies are included below. **Remember to print out the ACT sheet for one of the conditions for group discussion.**

- **2,4-dienoyl-CoA reductase**
- **2- methylbutyryl-CoA dehydrogenase def.**
- **2-methyl-3hydroxybutyryl-CoA dehydrogenase deficiency**
- **3-hydroxy-3methylglutaric aciduria**
- **3-methylcrotonyl- CoA carboxylase deficiency**
- **Argininemia (Arginase deficiency)**
- **Argininosuccinate aciduria**
- **Beta-ketothiolase (Mitochondrial acetyl-CoA thiolase deficiency)**
- **Biotinidase deficiency**
- **Carnitine acyl-carnitine translocase deficiency**
- **Carnitine palmitoyltransferase deficiency type 1**
- **Carnitine palmitoyltransferase deficiency type 2**
- **Carnitine uptake deficiency**
- **Citrullinemia**
- **Citrullinemia**
- **Cobalamin C deficiency**
- **Congenital adrenal hyperplasia**
- **Congenital hypothyroidism**
- **Critical congenital heart disease**
- **Cystic fibrosis**
- **Galactosemia**
- **Glutaric acidemia**
- **Glutaric acidemia type II**
- **Hearing loss**
- **Homocystinuria**
- **Hyperphenylalaninemia**
- **Isobutyryl-CoA dehydrogenase deficiency**
- **Isovaleric acidemia**
- **Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency**
- **Malonic acidemia**
- **Maple syrup urine disease (Branched-chain ketoacid dehydrogenase deficiency)**
- **Medium chain acyl-CoA dehydrogenase deficiency**
- **Methylmalonic acidemia**
- **Methylmalonic acidemia**
- **Mucopolysaccharidoses type I**
- **Multiple carboxylase deficiency**
- **Phenylketonuria**
- **Pompe disease (Glycogen storage disease type II)**
- **Propionic acidemia**
- **SC disease**
- **Severe combined immune deficiency**
- **Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency**
- **Short chain acyl-CoA dehydrogenase deficiency**
- **Sickle beta thalassemia**
- **Sickle cell anemia**
- **Spinal muscular atrophy**
- **Trifunctional protein deficiency**
- **Tyrosinemia type I**
- **Tyrosinemia type II**
- **Tyrosinemia type III**
- **Very long chain acyl-CoA dehydrogenase**

Newborn Screening ACT Sheet
[Elevated TSH (Primary TSH test)]
Congenital Hypothyroidism

Differential Diagnosis: Primary congenital hypothyroidism (CH); transient CH.

Condition Description: Lack of adequate thyroid hormone production..

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening test result.
- Consult pediatric endocrinologist; refer to endocrinologist, if considered appropriate.
- Evaluate infant (see clinical considerations below).
- Initiate timely confirmatory/diagnostic testing as recommended by the specialist.
- Initiate treatment as recommended by consultant as soon as possible.
- Educate parents/caregivers that hormone replacement prevents mental retardation.
- Report findings to state newborn screening program.

Diagnostic Evaluation: Diagnostic tests should include serum free T4 and thyroid stimulating hormone (TSH); consultant may also recommend total T4 and T3 resin uptake. Test results include reduced free T4 and elevated TSH in primary hypothyroidism; if done, reduced total T4 and low or normal T3 resin uptake

Clinical Considerations: Most neonates are asymptomatic, though a few can manifest some clinical features, such as prolonged jaundice, puffy facies, large fontanels, macroglossia and umbilical hernia. Untreated congenital hypothyroidism results in developmental delay or mental retardation and poor growth.

Additional Information:

American Academy of Pediatrics
Genetics Home Reference

Referral (local, state, regional and national):

Testing
Clinical Services
Lawson Wilkins Pediatric Endocrine Society "Find A Doc"
Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

American Academy of Pediatrics
http://pediatrics.aappublications.org/cgi/content/full/117/6/2290?maxtoshow=&HITS=10&hits=10&RESULTFORMAT=&fulltext=congenital+hypothyroidism&searchid=1&FIRSTINDEX=0&sortspec=relevance&resourcetype=HWCIT

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=congenitalhypothyroidism

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/20744?db=genetests&country=United%20Statezs

Clinical Services

Lawson Wilkins Pediatric Endocrine Society "Find a Doc"
http://lwpes.org

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

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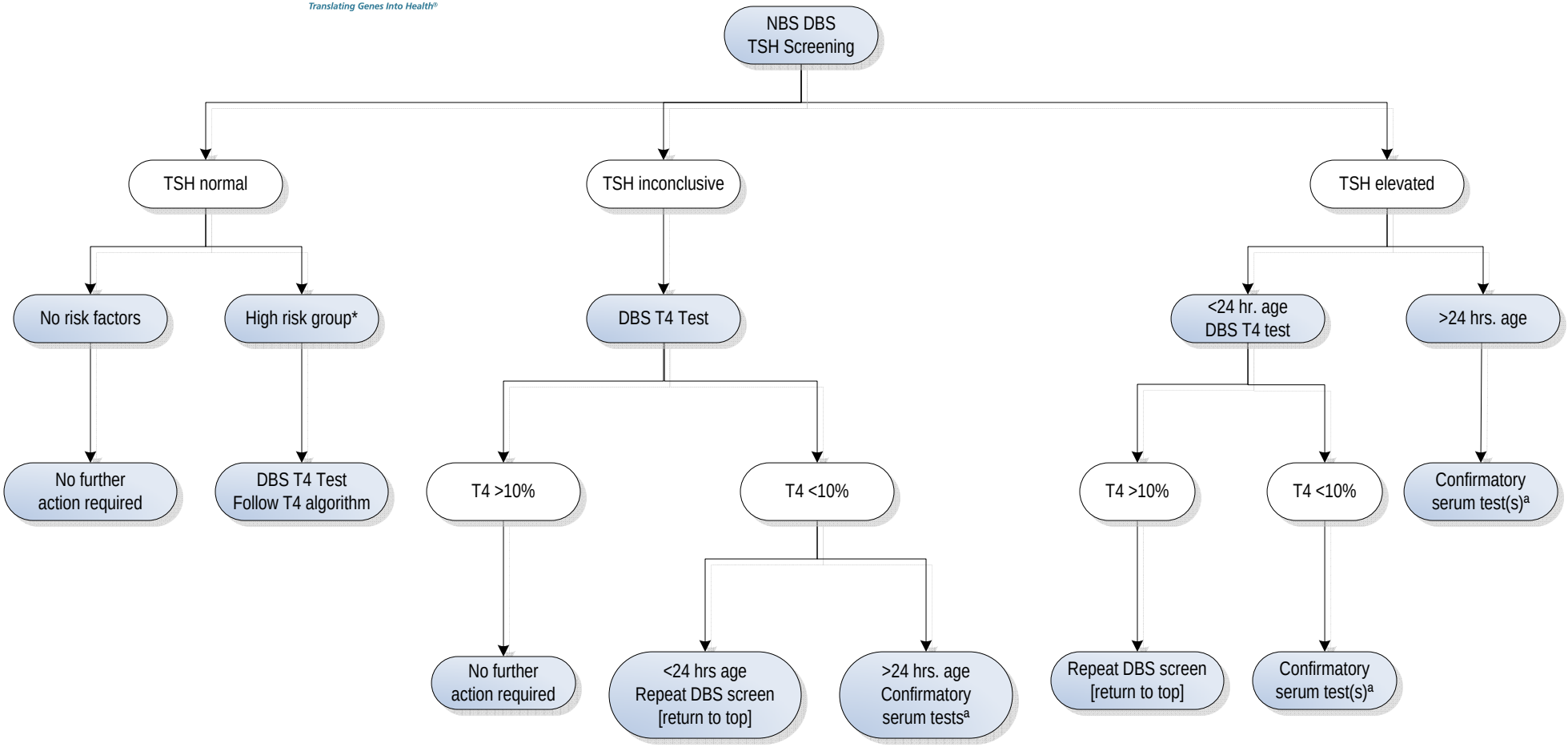
Congenital Hypothyroidism (TSH)



Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/Key
DBS = Dried blood spot
NBS = Newborn Screening
TSH = Thyroid stimulating hormone
T4 = Thyroxine or total thyroxine

* High risk group
<1500 gm
NICU admission
Same sex twin
Transfusion
CHD/other severe congenital anomaly
Drugs: dopamine, steroids, iodine

[a] Confirmatory Serum Tests
Free T4 [or]
4 and T3 resin uptake (T3RU)
TSH

(Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

Newborn Screening ACT Sheet
[Increased Phenylalanine]
Phenylketonuria (PKU)

Differential Diagnosis: Phenylketonuria (Classical PKU); non-PKU mild hyperphenylalaninemia; pterin defects; transient hyperphenylalaninemia.

Condition Description: In PKU the phenylalanine from ingested protein cannot be metabolized to tyrosine because of deficient liver phenylalanine hydroxylase (PAH). This causes elevated phenylalanine. Pterin defects result from deficiency of tetrahydrobiopterin (BH4), the cofactor for PAH and other hydroxylases. This produces not only increased phenylalanine but also neurotransmitter deficiencies.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Contact family immediately to inform them of the newborn screening result.
- Consult with pediatric metabolic specialist.
- Evaluate the newborn and refer as appropriate.
- Initiate confirmatory/diagnostic tests in consultation with metabolic specialist.
- Provide the family with basic information about PKU and dietary management.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma amino acid analysis which shows increased phenylalanine without increased tyrosine (increased phenylalanine:tyrosine ratio). Urine pterin analysis and red blood cell DHPR assay will identify pterin defects. Consider PAH mutation testing.

Clinical Considerations: Asymptomatic in the neonate. If untreated PKU will cause irreversible mental retardation, hyperactivity, autistic-like features, and seizures. Treatment will usually prevent these symptoms. Pterin defects cause early severe neurologic disease (developmental delay/seizures) and require specific therapy.

Additional Information:

Gene Reviews

Genetics Home Reference

PKU

Tetrahydrobiopterin Deficiency

Referral (local, state, regional and national):

Testing

Clinical Services

Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Reviews
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=pku

Genetics Home Reference

PKU
http://ghr.nlm.nih.gov/condition=phenylketonuria

Tetrahydrobiopterin Deficiency
http://ghr.nlm.nih.gov/condition=tetrahydrobiopterindeficiency

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2273?db=genetests&country=United%20States

Clinical Services
http://www.ncbi.nlm.nih.gov/sites/GeneTests/clinic?db=GeneTests

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

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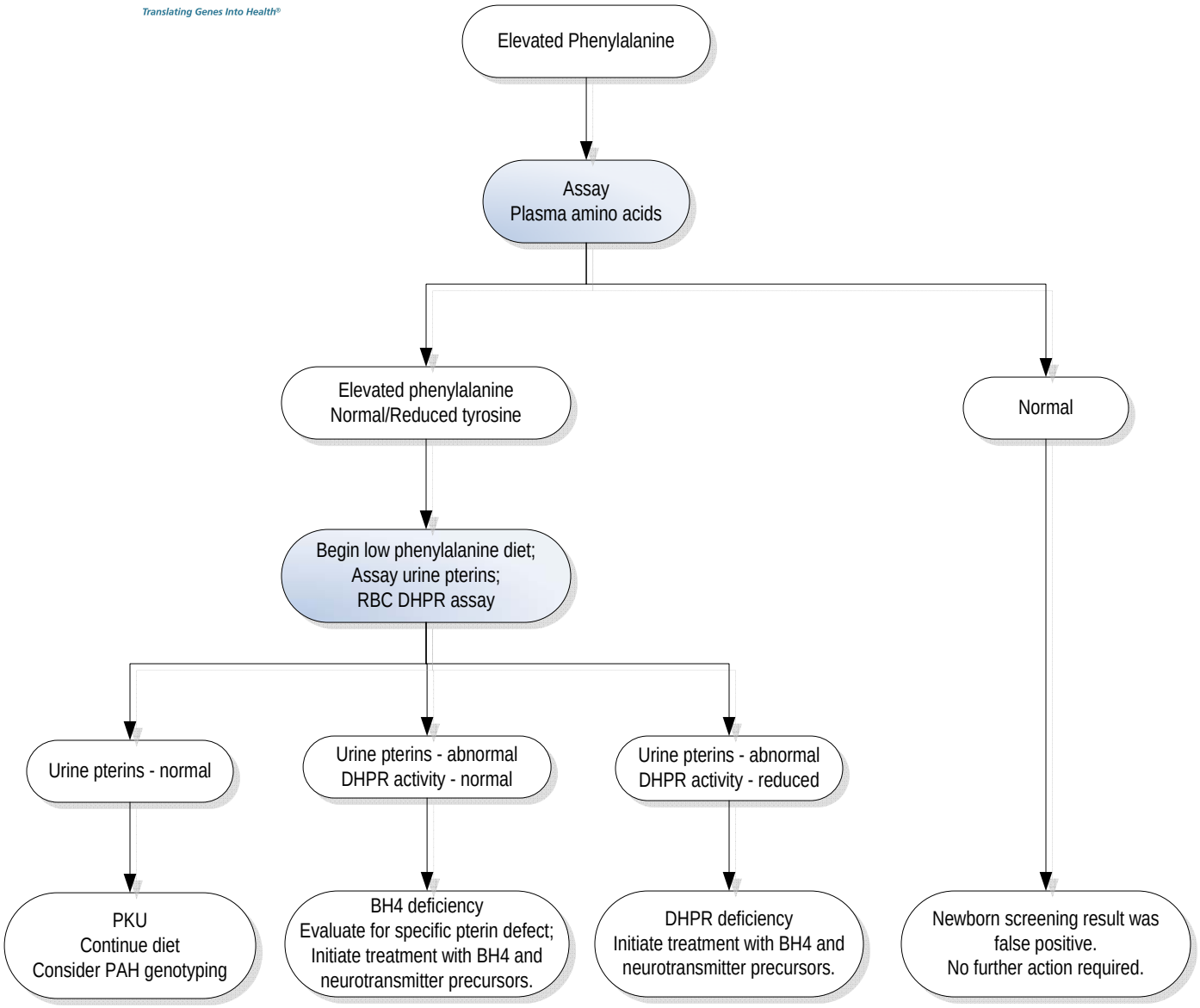
Phenylalanine Elevated



Action steps are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/Key
RBC = Red blood cell
PKU = Phenylketonuria
PAH = Phenylalanine hydroxylase
BH4 = Tetrahydrobiopterin
DHPR = Dihydropteridine reductase

(Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

Newborn Screening ACT Sheet
[FS]
Sickle Cell Anemia (HbSS Disease or HbS/Beta Zero Thalassemia)

Differential Diagnosis: Homozygous sickle cell disease (Hb SS), sickle beta-zero thalassemia, or sickle hereditary persistence of fetal hemoglobin (Hb S-HPFH).

Condition Description: A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin S in the absence of hemoglobin A. The hemoglobins are listed in order of the amount of hemoglobin present (F>S). This result is different from FAS which is consistent with sickle carrier.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result.
- Consult a specialist in hemoglobin disorders; refer if needed.
- Evaluate infant and assess for splenomegaly; do complete blood count (CBC) with mean corpuscular volume (MCV), and reticulocyte count.
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Initiate daily penicillin VK (125mg po bid) prophylaxis and other treatment as recommended by the consultant.
- Educate parents/caregivers regarding the risk of sepsis, the need for urgent evaluation if fever of ≥ 38.5° C (101° F) or signs and symptoms of splenic sequestration.

Diagnostic Evaluation: CBC, MCV, and reticulocyte count. Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FS pattern. DNA studies may be used to confirm genotype. Sickledex is not appropriate for confirmation of diagnosis in infants.

Clinical Considerations: Newborn infants are usually well. Hemolytic anemia and vaso-occlusive complications develop during infancy or early childhood. Complications include life-threatening infection, splenic sequestration, pneumonia, acute chest syndrome, pain episodes, aplastic crisis, dactylitis, priapism, and stroke. Comprehensive care including family education, immunizations, prophylactic penicillin, and prompt treatment of acute illness reduces morbidity and mortality. S-HPFH is typically benign.

Additional Information:

Grady Comprehensive Sickle Cell Center
Management and Therapy of Sickle Cell Disease
Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Protocols for Management of Acute and Chronic Complications
American Academy of Pediatrics
Sickle Cell Disease Association of America

Referral (local, state, regional and national):

Testing
Clinical Services
- Comprehensive Sickle Cell Center Directory
- Sickle Cell Information Center

Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

Grady Comprehensive Sickle Cell Center
http://www.scinfo.org/index.php?option=com_content&view=article&id=218:hemoglobins-what-the-results-mean&catid=11&Itemid=21

Management and Therapy of Sickle Cell Disease
http://www.nhlbi.nih.gov/health/prof/blood/sickle/index.htm

Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Protocols for Management of Acute and Chronic Complications
http://www.dshs.state.tx.us/newborn/pdf/sedona02.pdf

American Academy of Pediatrics
http://pediatrics.aappublications.org/cgi/content/full/109/3/526

Sickle Cell Disease Association of America
http://www.sicklecelldisease.org/

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2028?db=genetests&country=United%20States

Clinical Services

Comprehensive Sickle Cell Center Directory
http://www.scinfo.org/index.php?option=com_content&view=article&id=197&Itemid=34

Sickle Cell Information Center
http://www.scinfo.org/

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

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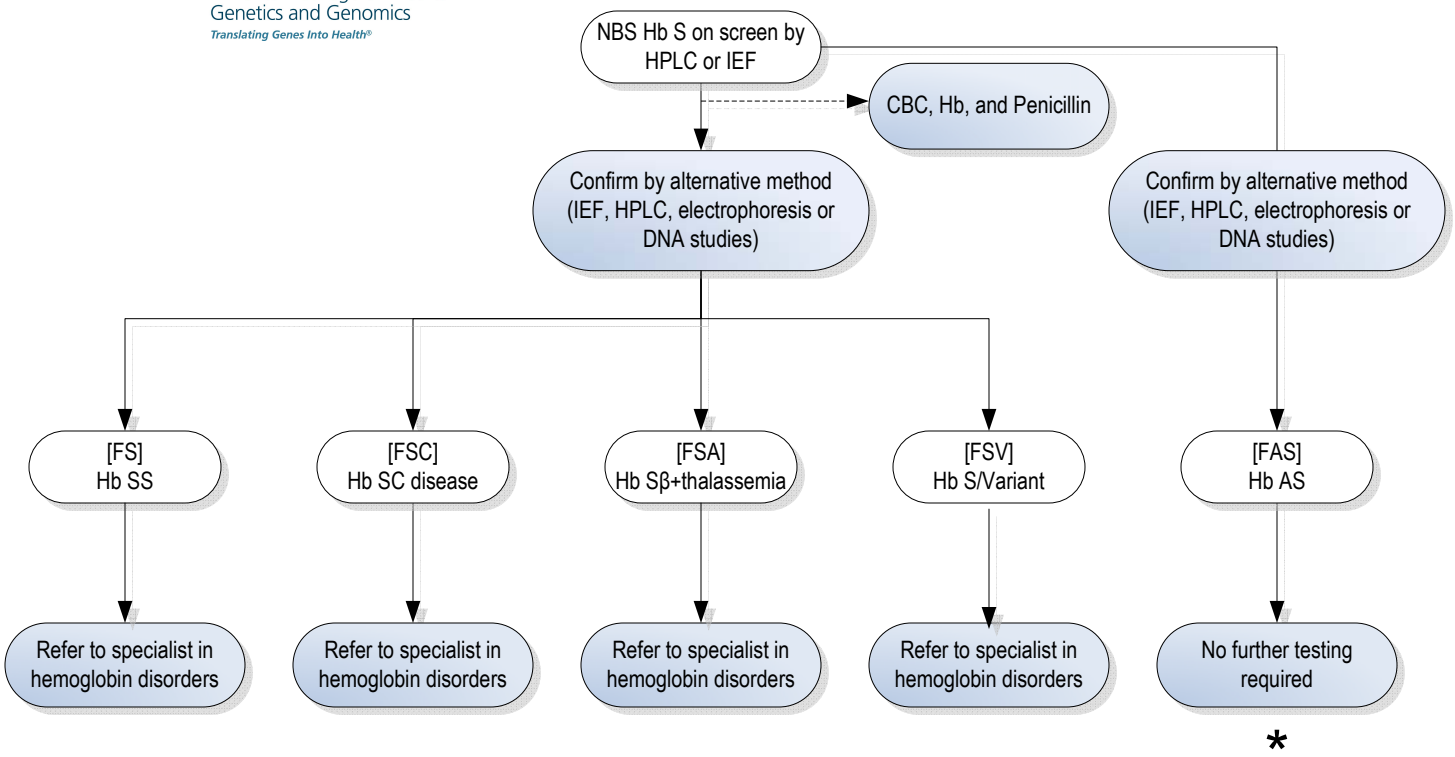
Hb S Screening



* Offer family members referral for hemoglobin disorders testing and genetic counseling.

Action steps are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/ Key

F, S, A, C, and V = The hemoglobins seen in neonatal screening.
HPLC: High performance liquid chromatography
IEF: Isoelectric focusing
‡ = Repeat testing at 6 months age is required if genotyping to confirm the newborn screening result is not done.

(Funded in part through MCHB/HRSA/HHS grant #U22MC03957)

Health Maintenance I Quiz:

1. Indicate the age(s) at which the following screening tests/referrals should be performed:
   A. Blood pressure: **to be performed yearly, starting at age 3 and earlier per risk factors**
   B. STI screening: **risk assessment yearly, starting at age 11; "all sexually active patients should be screened for STIs."** This will be discussed in more detail in Adolescent IV Health Maintenance Module.
   C. Developmental screening: **at 9mo, 18mo, & 30mo.** Also remind residents that Autism Screening should be done at 18mo & 24mo. This will be discussed in more detail in the upcoming Development Modules.

2. Match the following ages with the cut-off for "poor weight gain":
   A. 1 month-old: Less than __**20**__ grams/day **(goal is 20-30 g/day)**
   B. 4 month-old: Less than __**15**__ grams/day **(goal is 15-20 g/day)**
   C. 10 month-old: Less than __**5**___ grams/day **(goal is 5-10 g/day)**

3. The average infant will **triple** their birthweight and grow **10 in (25cm)** and increase head circumference by **12 cm (1cm/mo)**, by the end of the first year.

4. Fill out this table, comparing the 2000 CDC growth charts and the 2006 WHO charts:

	WHO	CDC
When to Use	0-2 yrs	2-20 yrs
Reference Population	**A "standard"/ideal** -Longitudinal sample (1997-2003) -6 countries including US -Breastfed (exclusive 4mo; through 12mo) -Solid foods introduced at 6mo -Excluded maternal smoking, low SES, etc.	**A "reference"** -Cross-sectional sample (1963-1994); multiple surveys in US. -Mostly formula-fed -VLBW and NHANES III (wt data) were only exclusions.
Cut-off Points	2nd & 98th %iles	5th & 95th %iles
Infant Growth	-Faster rate of weight gain in 1st 3mo; slower >3mo. Early recognition of obesity.	-Slower rate of weight gain in 1st 3mo; faster in >3mo olds.
Prevalence of: * Low weight-for-age & low W/L (FTT)	Lower	Higher
* Low length-for-age (short stature)	Similar	Similar
* High weight/length (overweight)	Lower	Higher

5. Maryland State Newborn Metabolic Screening is performed by **Maryland Health Department**, which tests for **51** disorders. On the ACMG website, these tests are broken down into **10** major categories. In Maryland, each baby is tested **two** times, first at **24 hrs** old and then at **10 days to 2 weeks** old.

Health Maintenance I Cases

Case 1: Dr. Greenwald hands you 2 cryptically folded pieces of paper, prior to your AM continuity clinic. When you unfold these papers, you see the Newborn Screen results for two babies scheduled today for 2-week well-baby-checks with you.

On the first, it shows that your 0900 appointment, Baby Hashimoto, has an abnormal thyroid screen (low T4 and high TSH). **What do you do next?**

Maryland uses a primary T4/back-up TSH method. If TSH is significantly elevated and T4 is low, a serum free T4 and TSH should be collected and urgent endocrinology referral should be made. **If T4 is low but TSH is normal or moderately elevated, this is a "borderline test," and a repeat NBS should suffice (in practice, though, many clinicians will still obtain the serum T4 & TSH).** If the follow-up specimen is abnormal, obtain serum T4 and TSH and request urgent endocrinology consult.

False positive and false negative results are possible with this screening. Specimen collection prior to 24 hours of age, prematurity, and illness can all affect the results. Nearly 90% of congenital hypothyroidism cases are detected by NBS; the remaining are detected clinically.

On the second, it shows that your 0930 appointment, Baby Folling has an abnormal PKU screen. **What do you do now?**

The differential diagnosis is phenylketonuria (Classical PKU), non-PKU mild hyper-phenylalaninemia, pterin defects, and transient hyperphenylalaninemia. **Follow-up specimen should be obtained within 24 hours. If the follow-up is abnormal, obtain serum amino acids and request endocrine/genetics consult.** An increased phenylalanine without increased tyrosine (increased P:T ratio) is diagnostic of hyperphenylalaninemia.

Trivia: Dr. Ivar Folling was the Norwegian physician and biochemist who first described the disease phenylketonuria in 1934, after following a family with two children who developed mental retardation and were noted to have a strong odor to their urine, ultimately found to be due to excess phenylpyruvic acid.

Case 2: Following your two 2-week well-baby-checks, you have two 12-month well-baby-checks. Because you were delayed by counseling your two previous patients, you are late for both your 1000 and 1030 well-baby-checks. When you enter each room successively, the parents are pacing impatiently. They each tell you that they are concerned by their babies' growth, and they would like a referral to a "specialist" immediately.

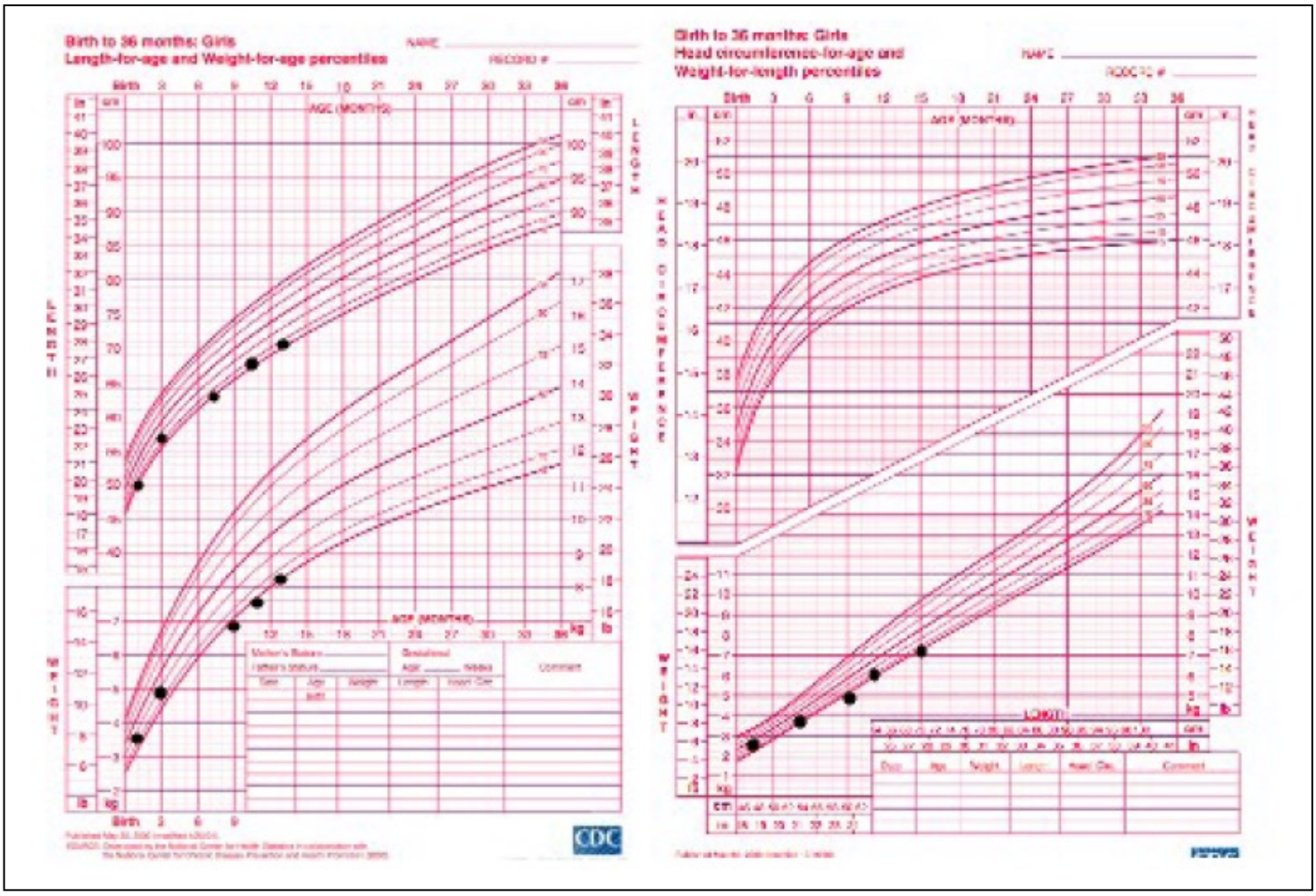
Review the growth charts (female baby & male baby) *on the next page*. **Which baby, if any, will you refer to a "specialist", and which specialty will you consult?**

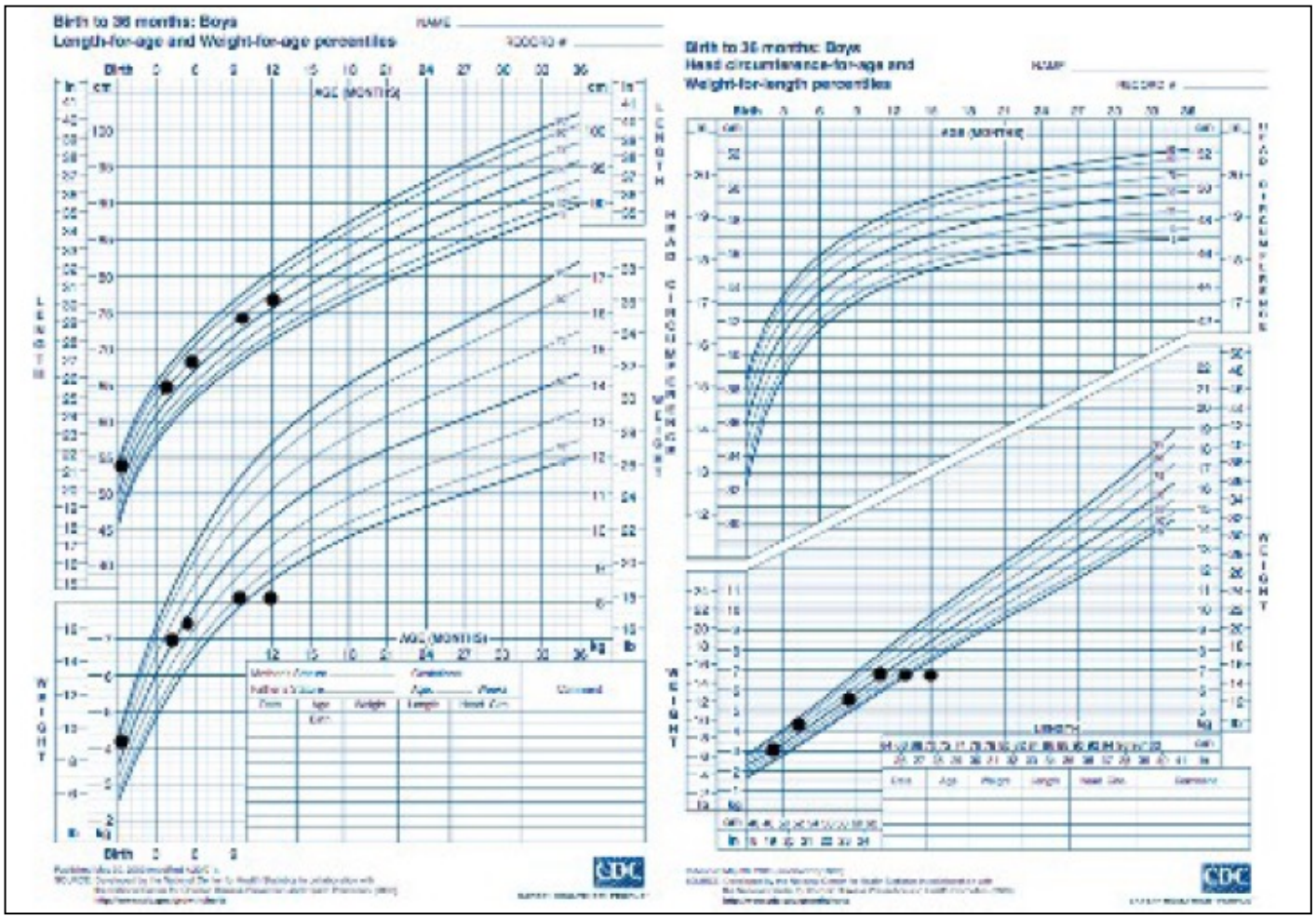
The male baby—children can often cross one or two growth percentiles between 6 and 24 months, but flattening curves are always abnormal. See more detailed explanation below:

- Female Baby: Likely **familial short stature** (i.e. the child's parents are both small and she is genetically pre-dispositioned to being short). Another factor that can affect growth potential is being born SGA.
- Male Baby: **Failure to thrive (FTT)**. The child maintained his growth in length, but his weight plateaued, falling below the 5th percentile. Of note, the child's slow growth might be missed if one only plotted weight and length, and not the weight-for-length chart. On this graph, chubby babies are higher and lean babies are lower.

The question of a **referral** may be left open to discussion. In general, the male baby's graph, showing poor weight gain but preserved length, reflects low energy substrate— e.g. due to poor intake, malabsorption, or increased metabolic demands. When FTT is due to endocrine or genetic causes, both weight and length are typically affected. *(See Board Review Q's)*

Therefore, if ANY specialty is to be consulted, **GI and/or Nutrition** would be most appropriate. That said, please emphasize to residents that most cases of FTT, such as Male Baby in this example, **can be managed by the general pediatrician**.





Case 3: The following growth chart *(see next page)* plots the head circumference of a 19mo female with poor weight gain and mild hypotonia. Please answer the following questions:

A. Is this normal or abnormal? Abnormal

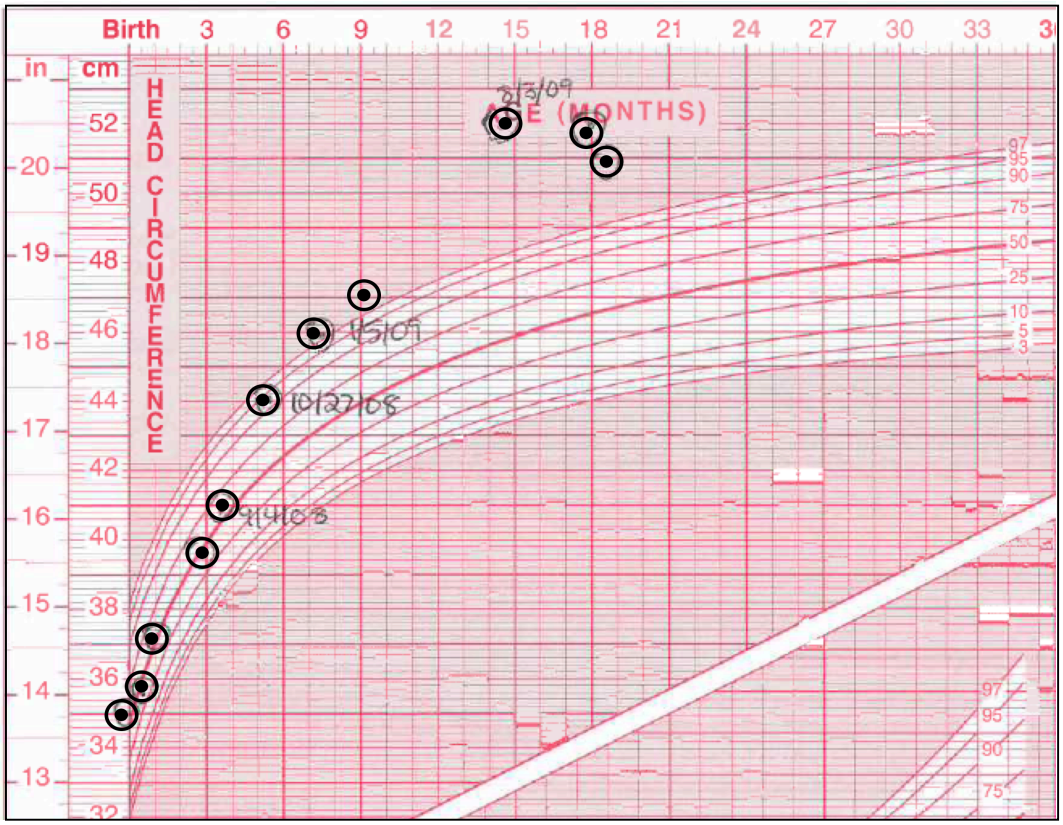
B. What is your differential diagnosis? Human error; hydrocephalus (obstructive or non-obstructive); tumor; head trauma (accidental or non-accidental); infection.

C. How would you initially evaluate this patient?

- CBC with differential, coagulation panel, non-contrast head CT.
- Consider primary head ultrasound or MRI depending on feasibility (e.g. open fontanel at earlier age) and availability.
- Residents who pick up on the concern for NAT may recommend an ophtho exam, skeletal survey, and/or LFTs and U/A as work-up for potential occult injuries.

D. If you could go back in time, when would you evaluate this patient? At 3.5 months? At 5 months? At 7 months? At 9 months? At 14 months? At 18 months?

This question may be left open to discussion. **An often used rule is that a head-circumference that crosses two major percentiles should be evaluated formally.** One could argue, then, that this child should have been evaluated as early as 5mo, when she rose from the 50-75th %ile to the 90-95th %ile.



Additional Background-Case III:

This growth chart came from an **actual M&M case** at WRAMC. The patient ended up receiving a CT scan at 18 months which showed **obstructive hydrocephalus** and subdural fluid collection with subacute hemorrhage. A subsequent MRI showed **large bilateral subdural hematomas**. Skeletal survey showed multiple old fractures; ophtho exam was negative for retinal hemorrhages. The patient had bilateral subdural drains placed (the last point on the HC graph).

The reasons for the missed evaluation of this rapidly increasing HC relate to various systems-based issues (e.g. multiple AHLTA entries for this patient, HC not measured or viewed at certain visits, difficulty coordinating follow-up and studies). The take-home point is that **residents need to look at ALL the growth charts at ALL well-baby/child visits** and consider obtaining measurements when appropriate at acute visits.

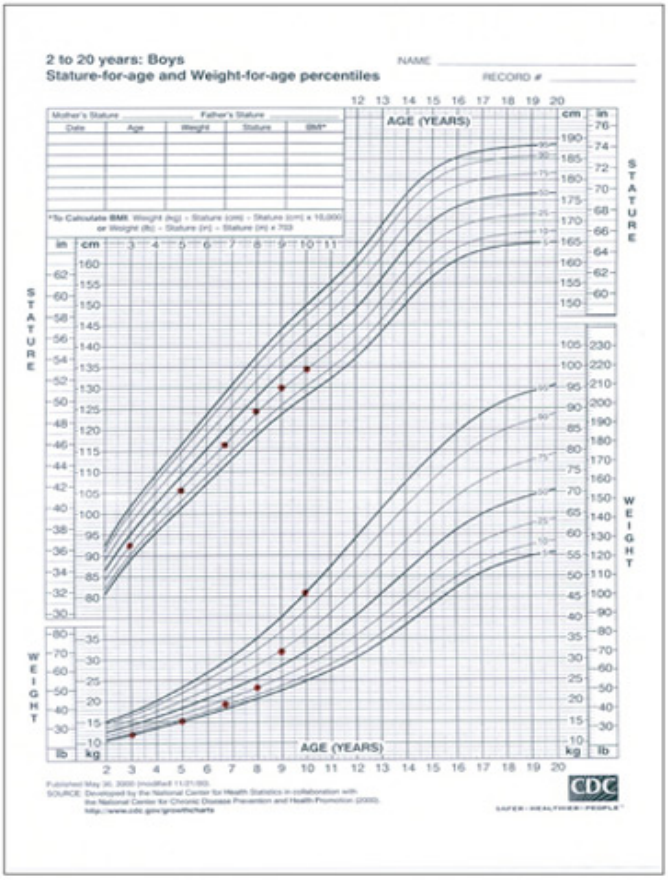
Health Maintenance I Board Review

1. The mother of a 10-year-old boy, whom you have been following since he was 3 years old, complains that he is always hungry and is gaining weight. The mother, who is overweight, reports that the boy refuses to exercise, and she cannot control his diet. She just read an article in a magazine about weight gain from Cushing syndrome and wonders if he could have this condition.

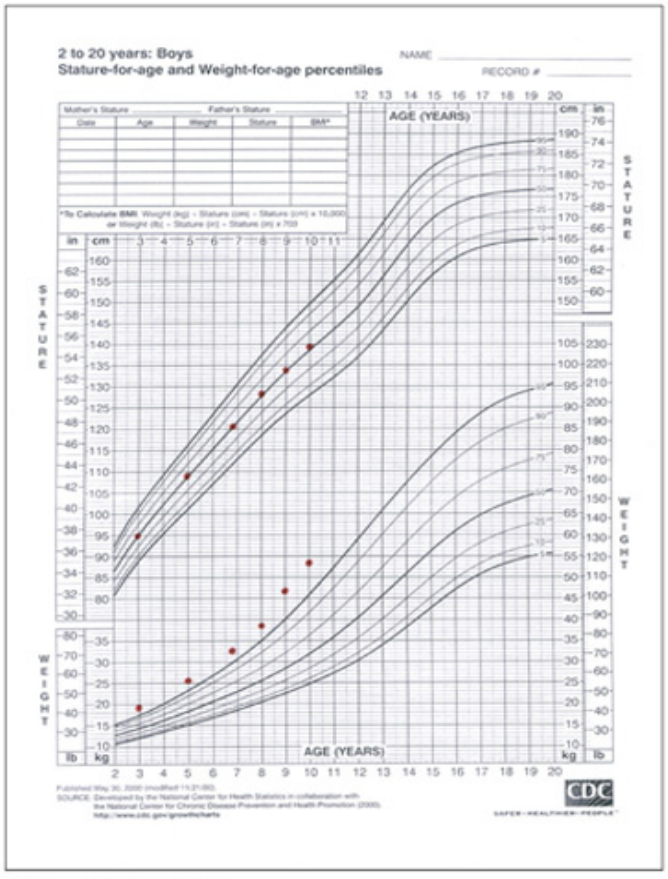
Of the following, the growth chart that suggests Cushing syndrome is
A. Growth chart A
B. Growth chart B
C. Growth chart C
D. Growth chart D
E. Growth chart E

Weight gain from exogenous obesity can be confused with **Cushing syndrome**, but glucocorticoid excess, as seen in Cushing syndrome, almost always is associated with attenuation of normal growth, as documented with Growth Chart C. The other growth charts are more typical for **exogenous obesity**, with height either enhanced or unchanged in the presence of weight gain.

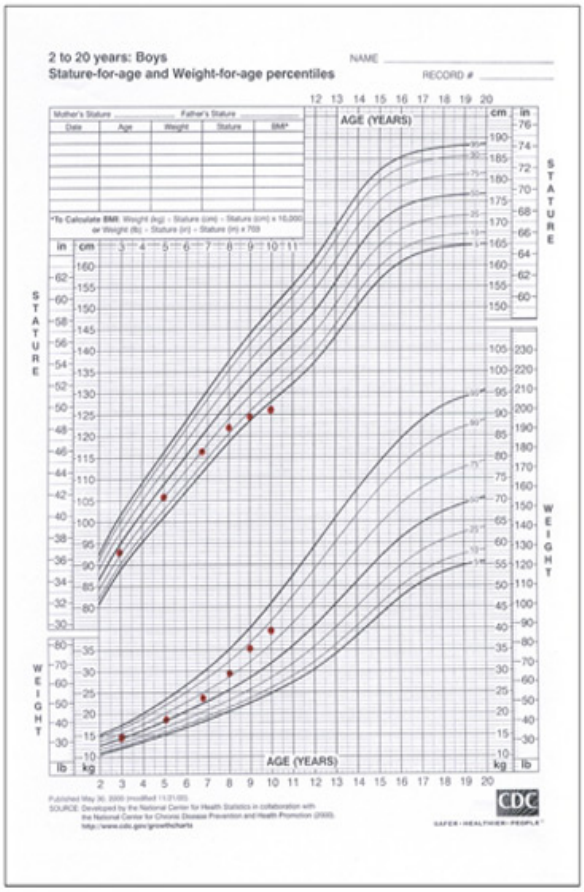
Other signs and symptoms of Cushing syndrome include hypertension, violaceous skin striae, "buffalo hump" and muscle weakness because of loss of muscle mass, centripetal obesity, cushingoid facies, easy bruisability, hirsutism, failure of pubertal progression or amenorrhea in women, loss of libido in men, headache, depression, and dysphoria.



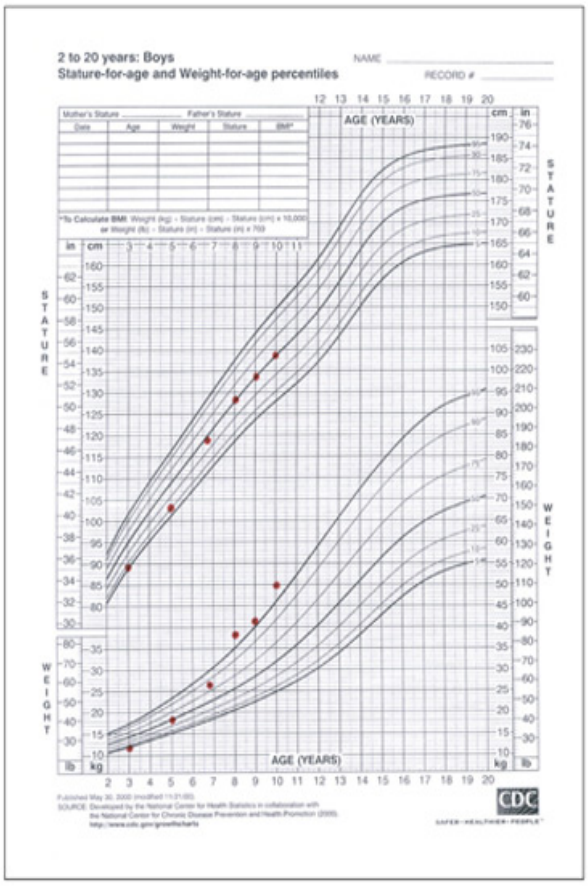
Growth Chart A



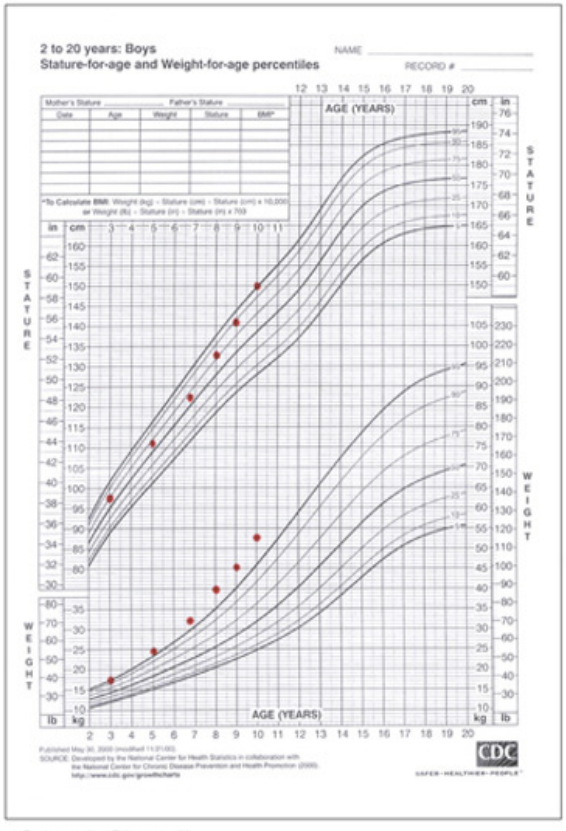
Growth Chart B



Growth Chart C



Growth Chart D



Growth Chart E

2. Of the following growth curves (see powerpoint), the one MOST likely to be associated with familial short stature in a boy who had a birthweight of 3.3 kg is

A. Item A

B. Item B

C. Item C

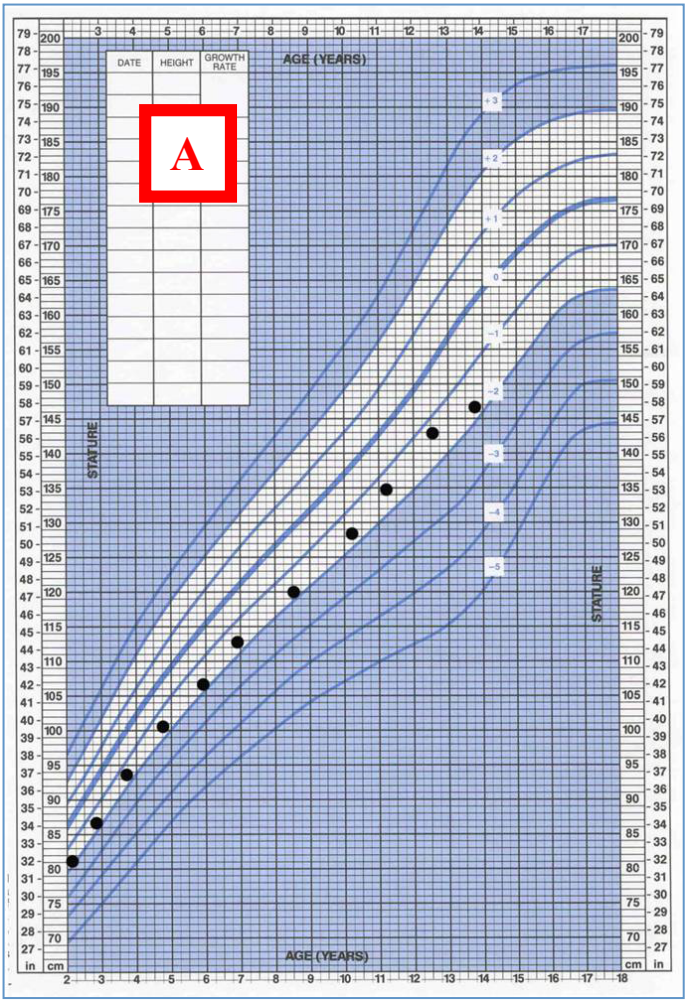
D. Item D

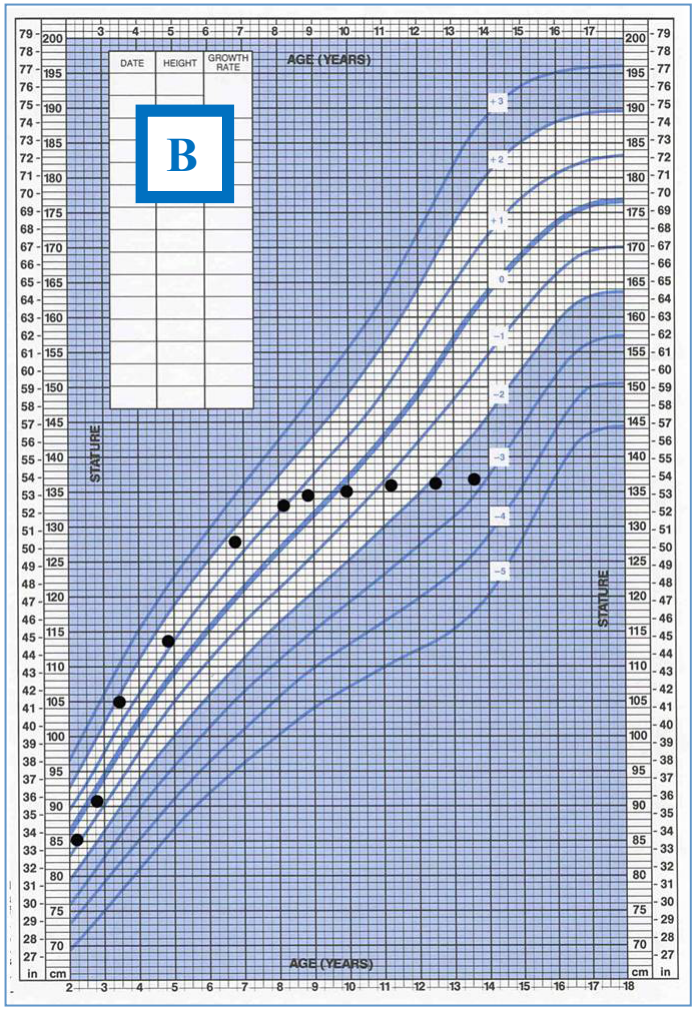
E. Item E

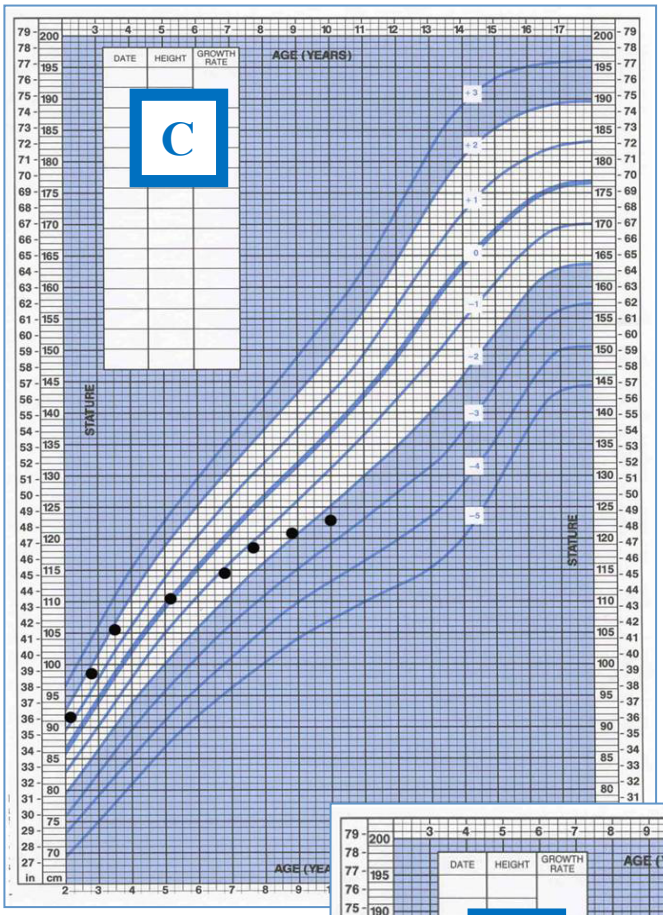
Children who are born relatively large but are destined to have short stature as adults because they come from short families (**familial short stature**) generally show a shift in growth percentiles so that by the time they are 2 years of age, they are growing at a steady rate and their height percentile is appropriate for their family. They mature at a normal time and achieve short normal adult stature after reaching full maturation, as in growth chart A.

Growth charts B, C, and D show the progress of children who have growth attenuation or arrest occurring or persisting past the second year. Such children likely have serious **underlying illnesses** interfering with linear growth. For example, a child who has celiac disease would be underweight and often experience weight loss before slowing in growth, while a child who has hypothyroidism would have a normal weight or be overweight for age, but have marked growth attenuation.

Growth chart E shows a continuation of growth with a growth spurt after other boys have reached adult height. A period of slowdown or attenuation in growth rate is documented just before the pubertal growth spurt, which may be relatively prolonged if puberty is late. This pattern is seen in **delayed puberty**, and it can be associated with relative short stature during childhood and a normal adult height.



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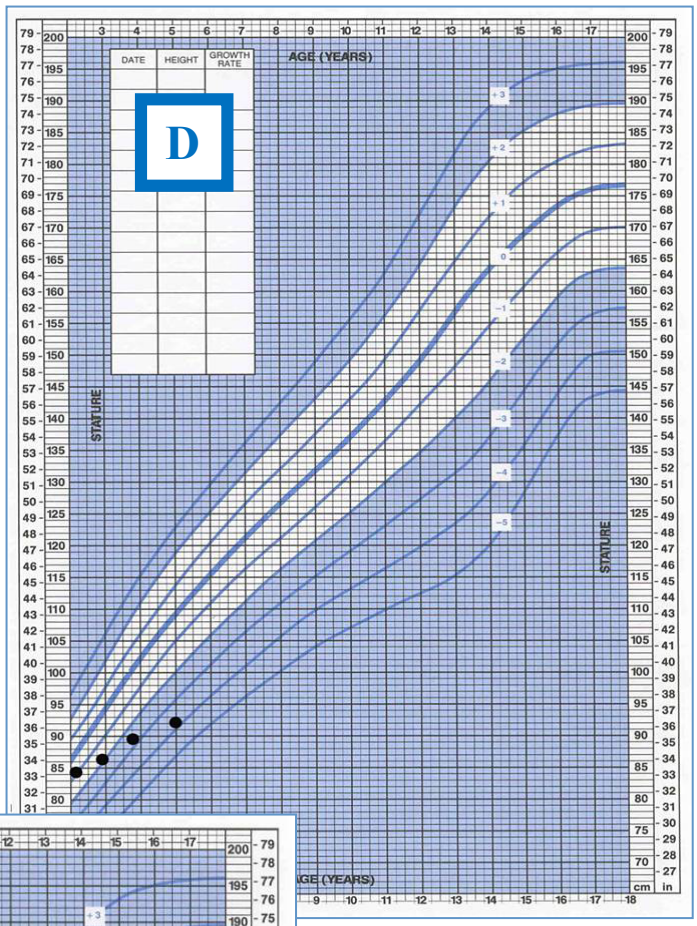
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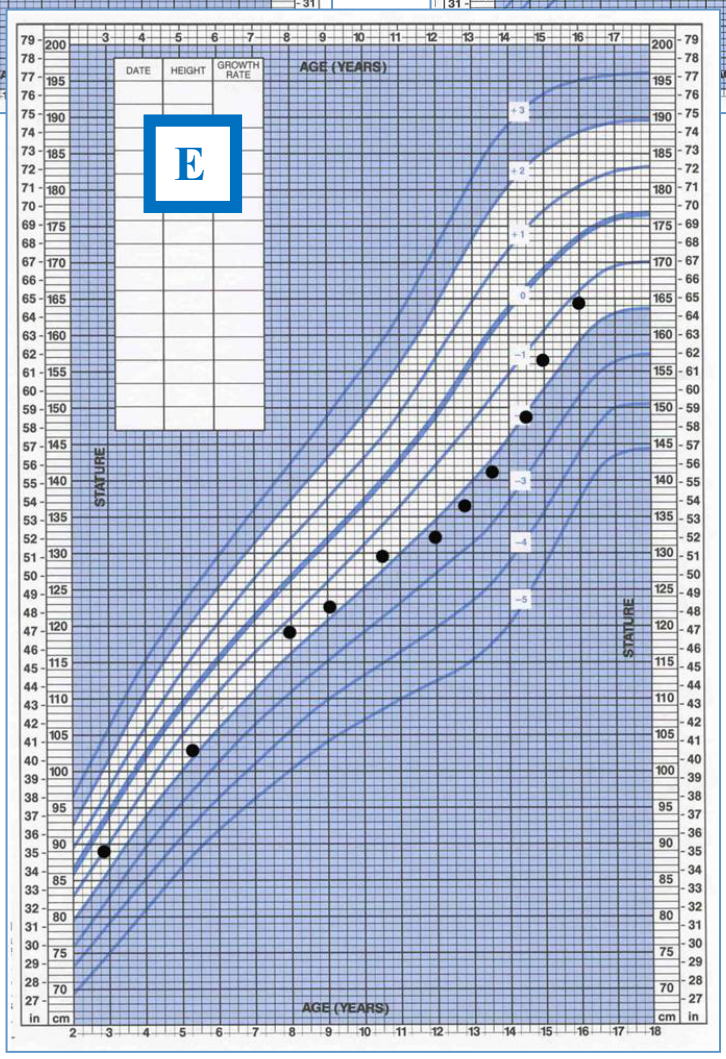
cm
in


E
DATE
HEIGHT
GROWTH RATE
AGE (YEARS)
STATURE
+3
+2
+1
0
−1
−3
−4
−5
in
cm