



# NCC Pediatrics Continuity Clinic Curriculum: **Health Maintenance I:**

## **Overall 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 2023 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*
- 3) Developmental/Behavioral Assessment:  
*Development I-V & Behavior I-II*
- 4) 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*
- 5) Oral Health: *Dental Health Modules*



# NCC Pediatrics Continuity Clinic

## Curriculum: **Health Maintenance I:**

### **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:**

*(“Using the WHO Growth Charts” & “Overview of the CDC Growth Charts”)*

- “Rules of Thumb for Growth” (p. 4)
- **Homework:** Print out an MHS Genesis growth chart + data table for an infant or toddler  $\leq 2$ yr you’ve seen in the past month. Bring in for group activity. *(If you haven’t seen toddlers, print out an interesting Genesis growth chart from any 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:** Discuss the benefits and pitfalls of using MHS Genesis charts. Demonstrate how to navigate to the charts, adjust for prematurity, toggle between charts and tables, and edit incorrect data points.
    - ❖ *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?

### **Resources:**

- Growth Charts: [WHO](#); [CDC](#); [Extended-BMI-For-Age](#); [VLBW\(Fenton\)](#) ; [Special Needs](#)
- [Newborn Screening Fact Sheets \(AAP Technical Report, 2006\)](#)
- [Maryland Newborn Screening Program Contact Information](#)
- [Virginia Newborn Screening Program Contact Information](#)



(continued)

14. Screen for behavioral and social-emotional problems per “Promoting Optimal Development: Screening for Behavioral and Emotional Problems” (<https://doi.org/10.1542/peds.2014-3716>), “Mental Health Competencies for Pediatric Practice” (<https://doi.org/10.1542/peds.2019-2757>), “Clinical Practice Guideline for the Assessment and Treatment of Children and Adolescents With Anxiety Disorders” (<https://pubmed.ncbi.nlm.nih.gov/32439401>), “Screening for Anxiety in Adolescent and Adult Women: A Recommendation From the Women’s Preventive Services Initiative” (<https://pubmed.ncbi.nlm.nih.gov/32510990>), and “Anxiety in Children and Adolescents: Screening” (<https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/screening-anxiety-children-adolescents>). The screening should be family centered and may include asking about caregiver emotional and mental health concerns and social determinants of health, racism, poverty, and relational health. See “Poverty and Child Health in the United States” (<https://doi.org/10.1542/peds.2016-0339>), “The Impact of Racism on Child and Adolescent Health” (<https://doi.org/10.1542/peds.2019-1765>), and “Preventing Childhood Toxic Stress: Partnering With Families and Communities to Promote Relational Health” (<https://doi.org/10.1542/peds.2021-052582>).
15. A recommended tool to assess use of alcohol, tobacco and nicotine, marijuana, and other substances, including opioids is available at <http://craftt.org>. If there is a concern for substance or opioid use, providers should consider recommending or prescribing Naloxone (see <https://www.cdc.gov/ore/search/pages/2018-evidence-based-strategies.html> and <https://nida.nih.gov/publications/drugfacts/naloxone>).
16. Screen adolescents for depression and suicide risk, making every effort to preserve confidentiality of the adolescent. See “Guidelines for Adolescent Depression in Primary Care (GLAD-PC): Part I. Practice Preparation, Identification, Assessment, and Initial Management” (<https://doi.org/10.1542/peds.2017-4081>), “Mental Health Competencies for Pediatric Practice” (<https://doi.org/10.1542/peds.2019-2757>), “Suicide and Suicide Attempts in Adolescents” (<https://doi.org/10.1542/peds.2016-1420>), and “The 21st Century Cures Act & Adolescent Confidentiality” ([https://adolescenthealth.org/press\\_release/naspag-sahm-statement-the-21st-century-cures-act-adolescent-confidentiality/](https://adolescenthealth.org/press_release/naspag-sahm-statement-the-21st-century-cures-act-adolescent-confidentiality/)).
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” (<https://doi.org/10.1542/peds.2011-0322>).
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/>) establish the criteria for and coverage of newborn screening procedures and programs.
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 “Clinical Practice Guideline Revision: Management of Hyperbilirubinemia in the Newborn Infant 35 or More Weeks of Gestation” (<https://doi.org/10.1542/peds.2022-058859>).
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” (<https://doi.org/10.1542/peds.2011-3211>).
23. Schedules, per the AAP Committee on Infectious Diseases, are available at <https://publications.aap.org/redbook/pages/immunization-schedules>. 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” (<https://doi.org/10.1542/peds.2016-1493>) and “Low Level Lead Exposure Harms Children: A Renewed Call for Primary Prevention” (<https://stacks.cdc.gov/view/cdc/11859>).

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](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. Screen adolescents for HIV at least once between the ages of 15 and 21, making every effort to preserve confidentiality of the adolescent, as per “Human Immunodeficiency Virus (HIV) Infection: Screening” (<https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/human-immunodeficiency-virus-hiv-infection-screening>); after initial screening, youth at increased risk of HIV infection should be retested annually or more frequently, as per “Adolescents and Young Adults: The Pediatrician’s Role in HIV Testing and Pre- and Postexposure HIV Prophylaxis” (<https://doi.org/10.1542/peds.2021-055207>).
31. Perform a risk assessment for hepatitis B virus (HBV) infection according to recommendations per the USPSTF (<https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/hepatitis-b-virus-infection-screening>) and in the 2021–2024 edition of the AAP *Red Book: Report of the Committee on Infectious Diseases*, making every effort to preserve confidentiality of the patient.
32. 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.
33. Perform a risk assessment, as appropriate, per “Sudden Death in the Young: Information for the Primary Care Provider” (<https://doi.org/10.1542/peds.2021-052044>).
34. 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” (<https://doi.org/10.1542/peds.2010-1564>).
35. Assess whether the child has a dental home. If no dental home is identified, perform a risk assessment (<https://www.aap.org/en/patient-care/oral-health/oral-health-practice-tools/>) 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” (<https://doi.org/10.1542/peds.2022-060417>).
36. Perform a risk assessment (<https://www.aap.org/en/patient-care/oral-health/oral-health-practice-tools/>). See “Maintaining and Improving the Oral Health of Young Children” (<https://doi.org/10.1542/peds.2022-060417>).
37. The USPSTF recommends that primary care clinicians apply fluoride varnish to the primary teeth of all infants and children starting at the age of primary tooth eruption (<https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/prevention-of-dental-caries-in-children-younger-than-age-5-years-screening-and-interventions1>). Once teeth are present, apply fluoride varnish to all children every 3 to 6 months in the primary care or dental office based on caries risk. Indications for fluoride use are noted in “Fluoride Use in Caries Prevention in the Primary Care Setting” (<https://doi.org/10.1542/peds.2020-034637>).
38. If primary water source is deficient in fluoride, consider oral fluoride supplementation. See “Fluoride Use in Caries Prevention in the Primary Care Setting” (<https://doi.org/10.1542/peds.2020-034637>).

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

This schedule reflects changes approved in December 2023 and published in June 2024. For updates and a list of previous changes made, visit [www.aap.org/periodicityschedule](http://www.aap.org/periodicityschedule).

### FOOTNOTE CHANGES MADE IN DECEMBER 2023

#### • 3-5 DAY VISIT (Footnote 4)

This footnote reflects the AAP “[Policy Statement: Breastfeeding and the Use of Human Milk](#)”, published June 2022.

#### • BODY MASS INDEX (Footnote 5)

This footnote reflects the AAP “[Clinical Practice Guideline for the Evaluation and Treatment of Children and Adolescents with Obesity](#)”, published January 2023.

#### • BEHAVIORAL/SOCIAL/EMOTIONAL SCREENING (Footnote 14)

This footnote reflects the USPSTF “[Anxiety in Children and Adolescents: Screening](#)” recommendations, published October 2022.

#### • TOBACCO, ALCOHOL, OR DRUG USE ASSESSMENT (Footnote 15)

This footnote reflects the Centers for Disease Control (CDC) and National Institute of Drug Abuse (NIDA) guidance related to recommending and prescribing Naloxone.

#### • NEWBORN BILIRUBIN SCREENING (Footnote 21)

This footnote reflects the AAP “[Clinical Practice Guideline Revision: Management of Hyperbilirubinemia in the Newborn Infant 35 or More Weeks of Gestation](#)”, published August 2022.

#### • ORAL HEALTH (Footnotes 35 and 36)

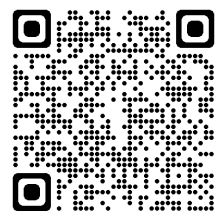
These footnotes reflect the AAP clinical report, “[Maintaining and Improving the Oral Health of Young Children](#)”, published December 2022.

### CHANGES MADE IN DECEMBER 2022

#### HIV

The HIV screening recommendation has been updated to extend the upper age limit from 18 to 21 years (to account for the range in which the screening can take place) to align with recommendations of the US Preventive Services Task Force and AAP policy (“Adolescents and Young Adults: The Pediatrician’s Role in HIV Testing and Pre- and Postexposure HIV Prophylaxis”).

- Footnote 30 has been updated to read as follows: “Screen adolescents for HIV at least once between the ages of 15 and 21, making every effort to preserve confidentiality of the adolescent, as per ‘Human Immunodeficiency Virus (HIV) Infection: Screening’ (<https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/human-immunodeficiency-virus-hiv-infection-screening>); after initial screening, youth at increased risk of HIV infection should be retested annually or more frequently, as per ‘Adolescents and Young Adults: The Pediatrician’s Role in HIV Testing and Pre- and Postexposure HIV Prophylaxis’ (<https://doi.org/10.1542/peds.2021-055207>)”



**HRSA**  
Health Resources & Services Administration

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## Rules of Thumb for Growth

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

1. Weight loss in 1<sup>st</sup> few days—5-10% of birthweight.
2. Return to birthweight @ 7-10 days
3. Double birthweight @ 4 months
4. Triple birthweight @ 1 year
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, 5 cm 3-9 mo, 5 cm from 9 mo-3 yrs, and 5 cm 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 bands—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 51 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 2<sup>nd</sup> 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; Maryland uses 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 and posted on an Excel spread sheet prior to making it into MHSB 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..

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### ***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 fontanel, 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](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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](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"

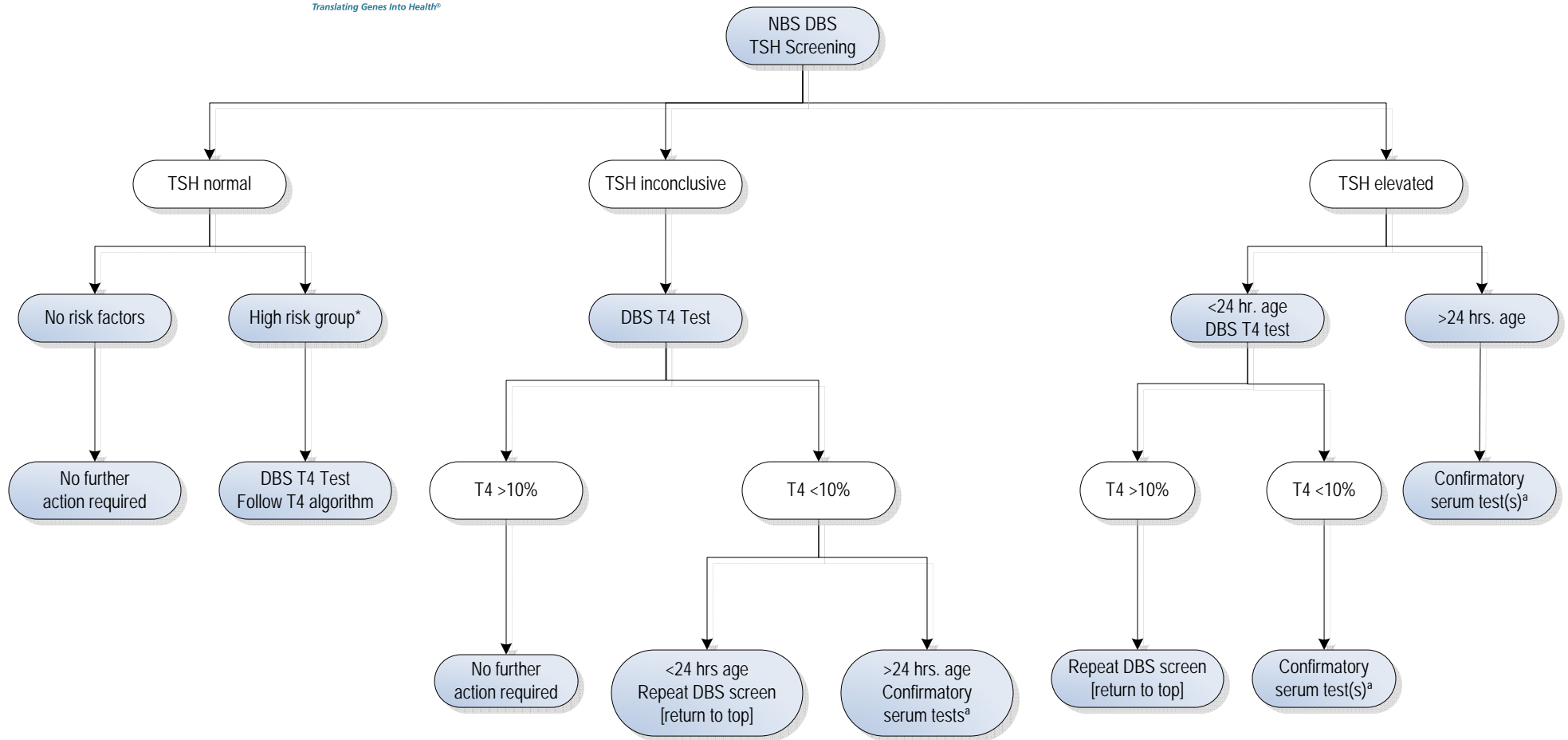
<http://lwpes.org>

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

<sup>a</sup> Confirmatory Serum Tests

Free T4 [or]  
 4 and T3 resin uptake (T3RU)  
 TSH

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## 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.

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***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.

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**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](#)

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LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Reviews

<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=pk>

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](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

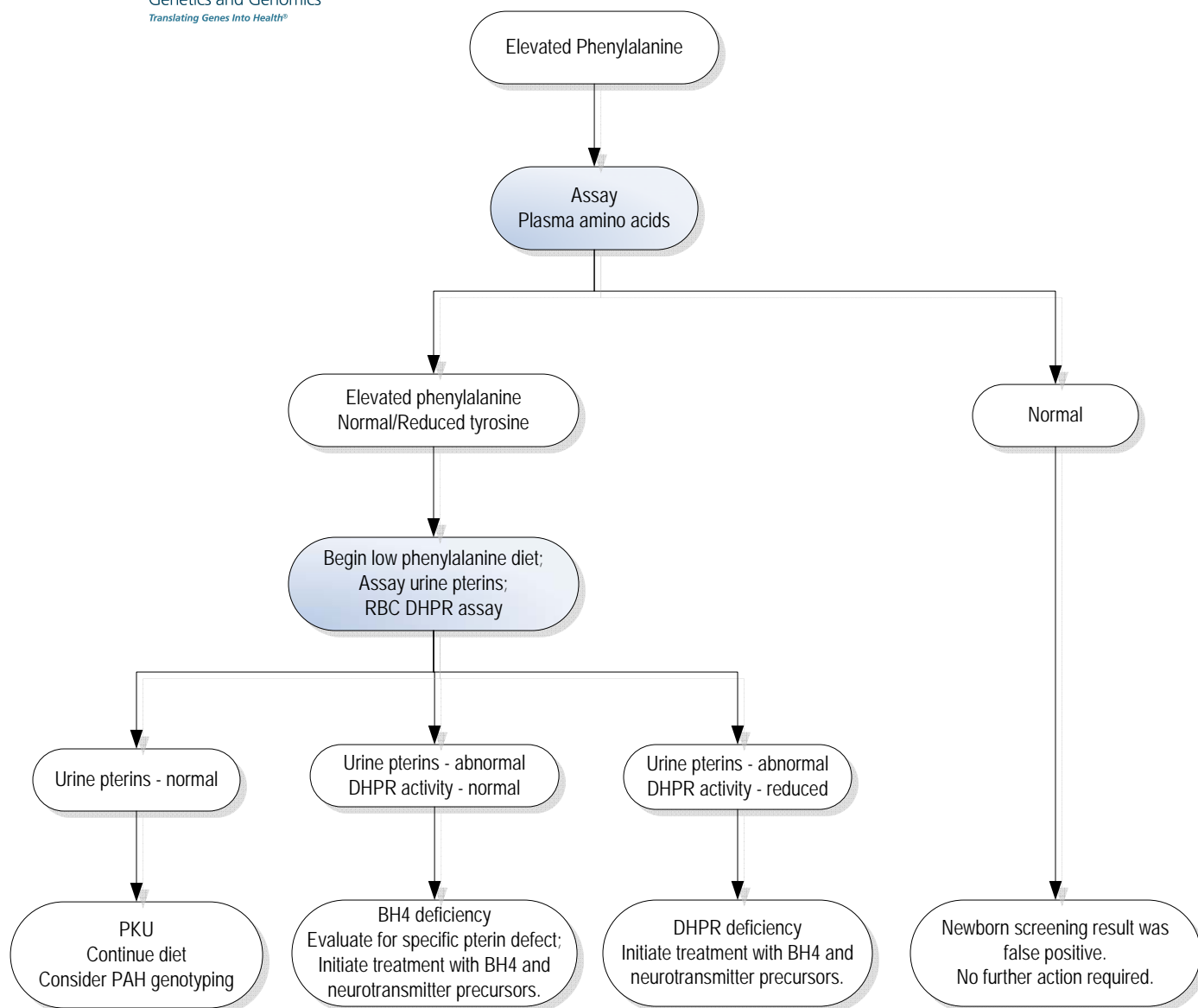
<http://www.acmg.net/GIS/Disclaimer.aspx>

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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

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## 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  $\geq 38.5^{\circ}$  C ( $101^{\circ}$  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](#)

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Comments	<input type="text"/>

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](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](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](http://www.scinfo.org/index.php?option=com_content&view=article&id=197&Itemid=34)

Sickle Cell Information Center

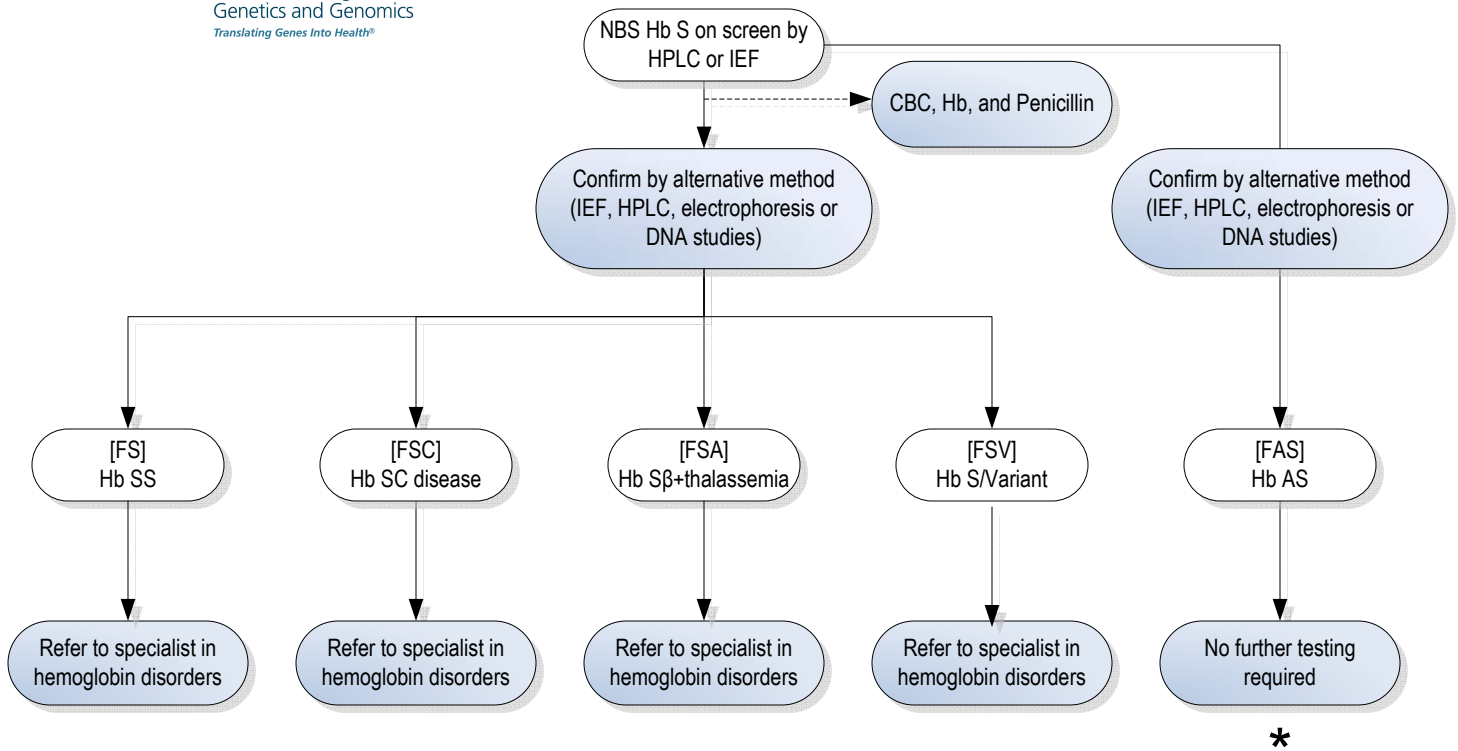
<http://www.scinfo.org/>

Find Genetic Services

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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.

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## Health Maintenance I Quiz:

1. Match the screening test or referral below with the age it should be performed at:

- A. Blood pressure
- B. STI screening
- C. Developmental screening

2. Match the following ages with the cut-off for “poor weight gain”:

- A. 1 month-old: Less than \_\_\_\_\_ grams/day
- B. 4 month-old: Less than \_\_\_\_\_ grams/day
- C. 10 month-old: Less than \_\_\_\_\_ grams/day

3. The average infant will \_\_\_\_\_ (**double, triple, or quadruple**) their birthweight and grow \_\_\_\_\_ **inches** and increase head circumference by \_\_\_\_\_ **cm**, 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
<b>When to Use</b>		
<b>Reference Population</b>		
<b>Cut-off Points</b>		
<b>Infant Growth</b>		
<b>Prevalence of:</b>		
* Low weight-for-age & low W/L (FTT)		
* Low length-for-age (short stature)		
* High weight/length (overweight)		

5. Maryland State Newborn Metabolic Screening is performed by \_\_\_\_\_, which tests for \_\_\_\_\_ metabolic disorders. On the ACMG website, these tests are broken down into \_\_\_\_\_ major categories. In Maryland, each baby is tested \_\_\_\_\_ times, first at \_\_\_\_\_ old and then at \_\_\_\_\_ old.

6. When and how should you check a patient's newborn screen results?

## Health Maintenance I Cases

**Case 1:** Dr. Greenwald hands you 2 cryptically folded pieces of paper, prior to your 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?**

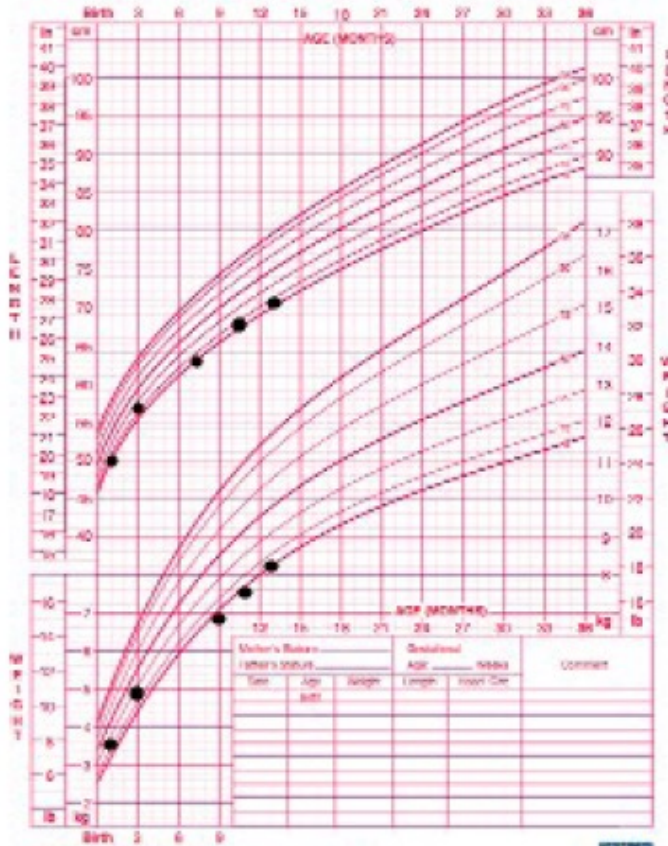
On the second, it shows that your 0930 appointment, Baby Folling has an abnormal PKU screen. **What do you do now?**

**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?**

**Birth to 36 months: Girls**  
Length-for-age and Weight-for-age percentiles

NAME \_\_\_\_\_ RECORD # \_\_\_\_\_

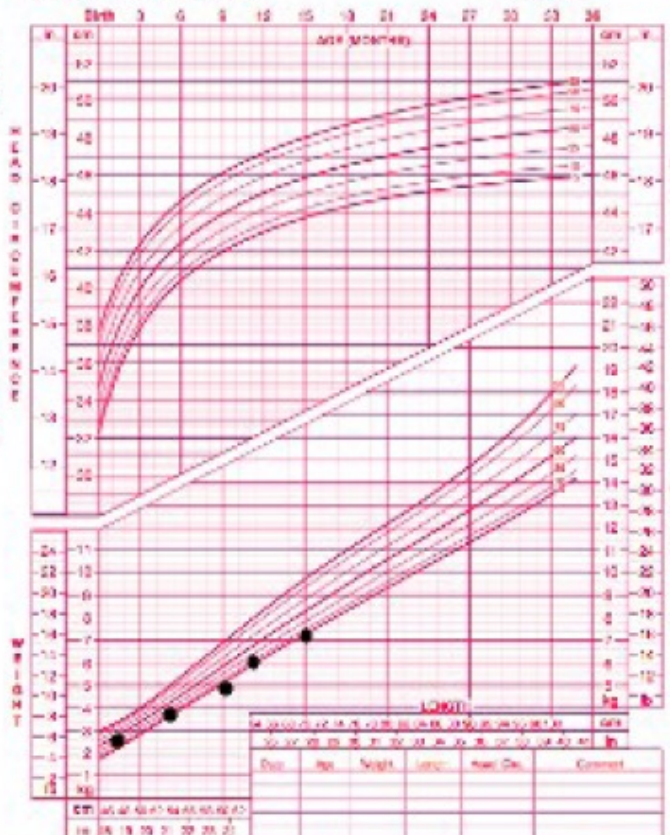


Published May 20, 2000 (revised August 2000).  
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**Birth to 36 months: Girls**  
Head circumference-for-age and Weight-for-length percentiles

NAME \_\_\_\_\_ RECORD # \_\_\_\_\_

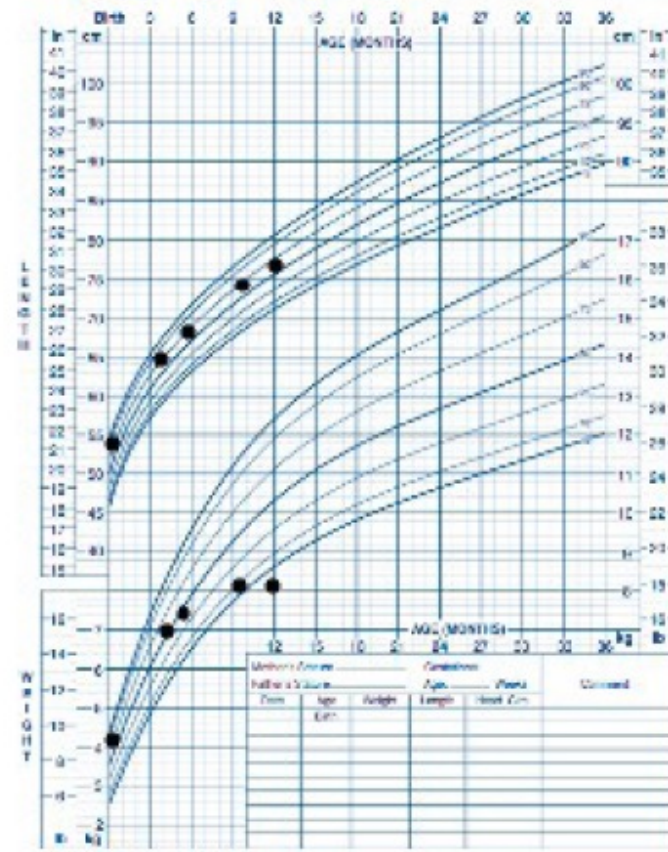


Published May 20, 2000 (revised August 2000).



**Birth to 36 months: Boys**  
Length-for-age and Weight-for-age percentiles

NAME \_\_\_\_\_ RECORD # \_\_\_\_\_

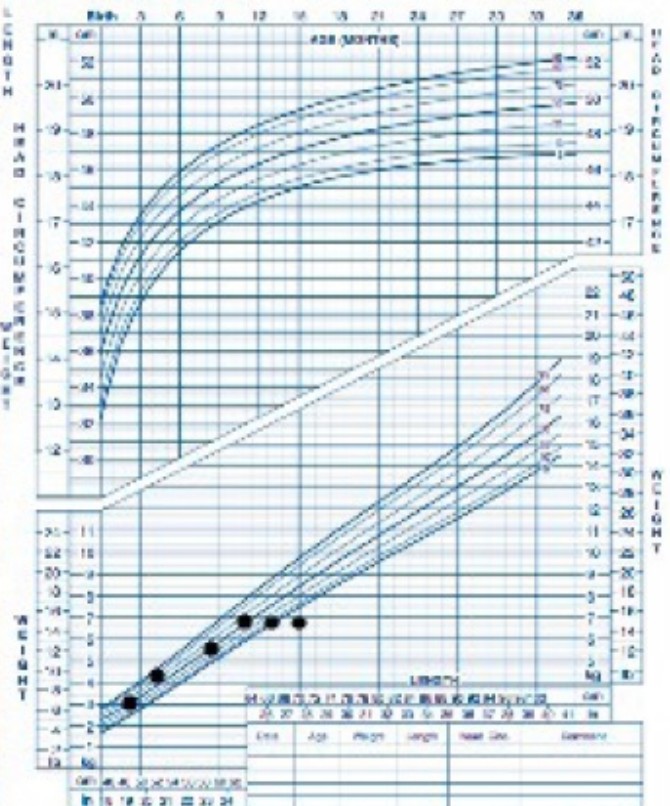


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**Birth to 36 months: Boys**  
Head circumference-for-age and Weight-for-length percentiles

NAME \_\_\_\_\_ RECORD # \_\_\_\_\_



Published May 20, 2000 (revised August 2000).





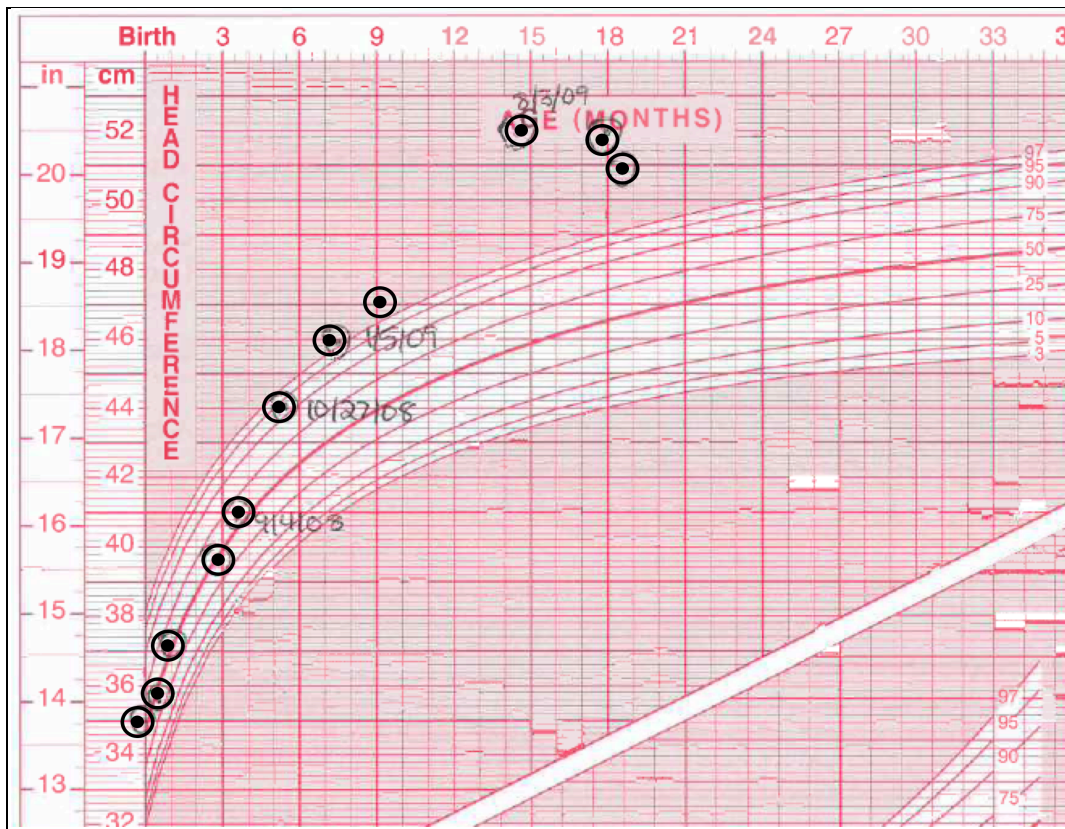
**Case 3:** The following growth chart plots the head circumference of a 19mo female with poor weight gain and mild hypotonia. Please answer the following questions:

**A. Is this normal or abnormal?**

**B. What is your differential diagnosis?**

**C. How would you evaluate this patient?**

**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?**



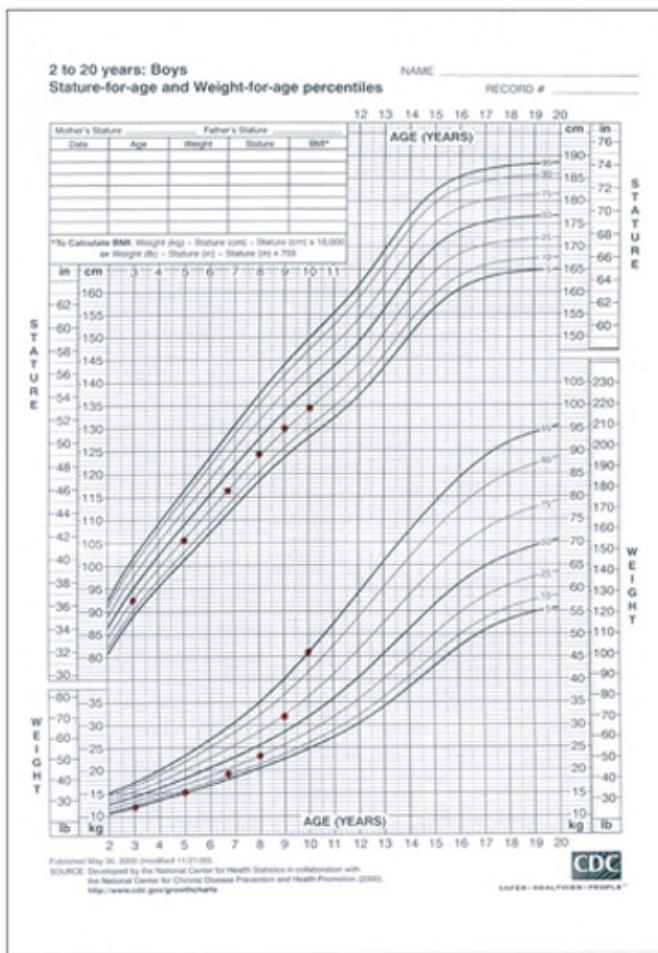


## Health Maintenance I Board Review

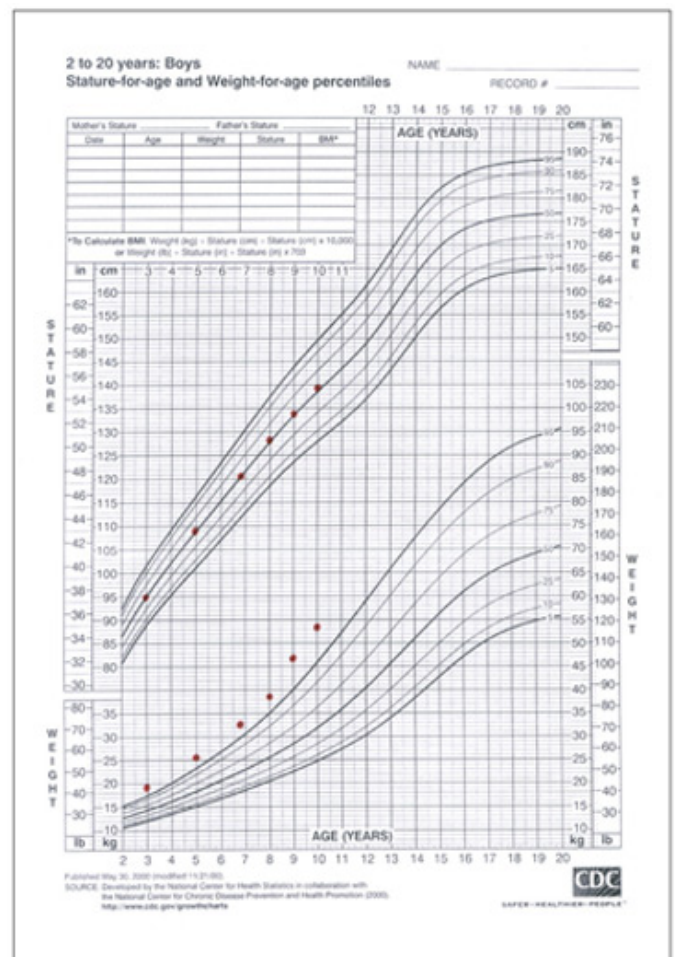
1. The mother of a 10-year-old boy, whom you have been following since he was 3 years old, complains that he is always hungry and is gaining weight. The mother, who is overweight, reports that the boy refuses to exercise, and she cannot control his diet. She just read an article in a magazine about weight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

Of the following, the growth chart that suggests Cushing syndrome is

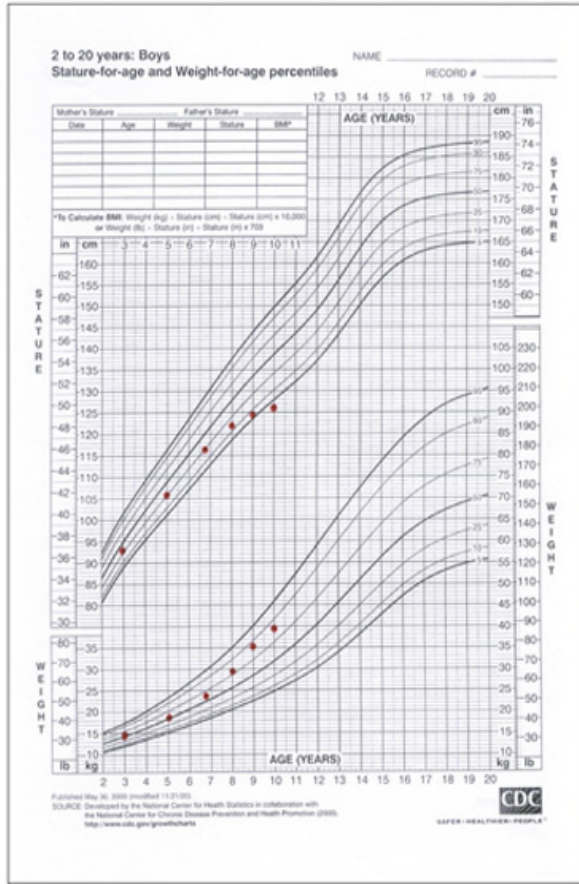
- A. Growth chart A
- B. Growth chart B
- C. Growth chart C
- D. Growth chart D
- E. Growth chart E



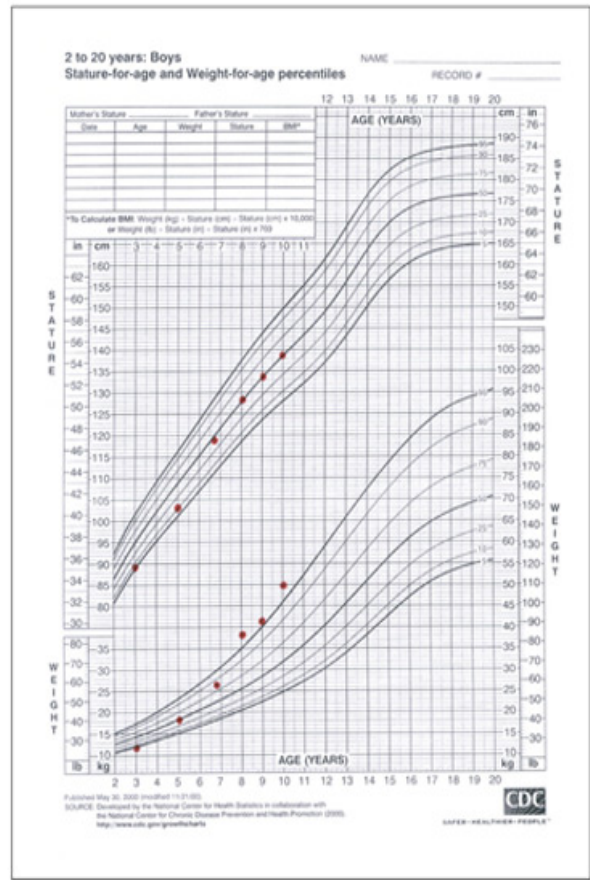
Growth Chart A



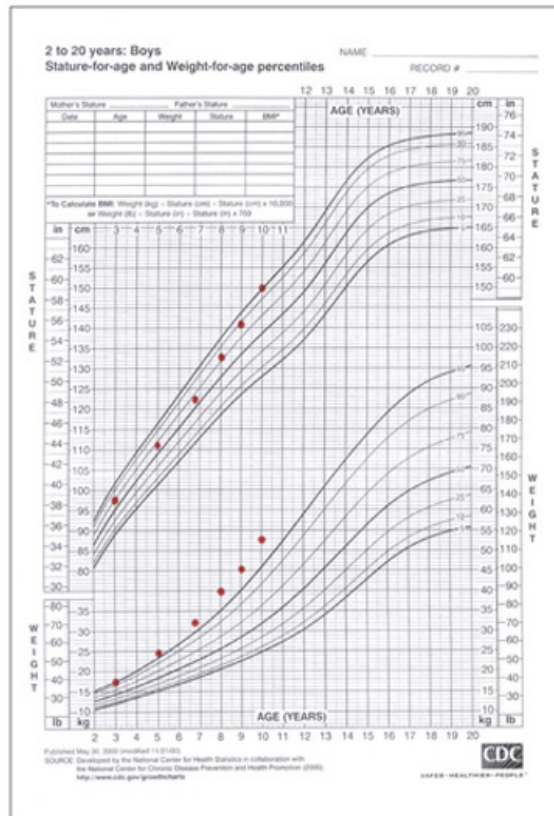
Growth Chart B



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

