One Regional Collaborative's Activities to Improve Access to Services for CYSHCN

Katharine B. Harris, MBA
NYMAC Project Manager
AMCHP Conference, 2/13/12
HRSA/MCHB’s Mandate

“The primary goal of the RCs is to ensure that individuals with heritable disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home that provides accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally effective care.”
The Regional Collaboratives
NYMAC: New York — Mid-Atlantic Consortium for Genetic and Newborn Screening Services
Standardization of Newborn Screening Panels

Newborn Screening Panels 2004 and 2012

- Other (6/7)*
- Hemoglobin (3/1)
- Endocrine (2/0)
- Amino Acid (6/8)
- Organic Acid (9/6)
- Fatty Acid (5/8)

* (core conditions/secondary conditions)
# NYMAC Diagnostic Guidelines for Confirmation of Screen-Positive Newborn Screening Results

## Phenylketonuria (PKU) and Hyperphenylalaninemia (H-PHE) (Amino Acidemia)

<table>
<thead>
<tr>
<th>Information</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Disease (common abbreviation)</strong></td>
<td>Phenylketonuria (PKU) and Hyperphenylalaninemia (H-PHE)</td>
</tr>
<tr>
<td><strong>MIM #</strong></td>
<td>261600</td>
</tr>
<tr>
<td><strong>SNOMED Code / ICD-10-CM Code</strong></td>
<td>7573000 / E70.0</td>
</tr>
<tr>
<td><strong>Enzyme or other abnormality</strong></td>
<td>Phenylalanine hydroxylase</td>
</tr>
<tr>
<td><strong>MIM # / Enzyme Commission #</strong></td>
<td>261600 / 1.14.16.1</td>
</tr>
<tr>
<td><strong>Abnormal Newborn Screening Metabolite(s)</strong></td>
<td>Elevated phenylalanine</td>
</tr>
<tr>
<td><strong>LOINC Number(s)</strong></td>
<td>29573-3</td>
</tr>
<tr>
<td><strong>Initial Diagnostics at Referral Center</strong></td>
<td>Plasma amino acids (PAA)</td>
</tr>
<tr>
<td><strong>Recommended additional testing to consider at time of initial consultation</strong></td>
<td>Urine biopterin and neopterin, Dihydropteridine reductase activity</td>
</tr>
<tr>
<td><strong>Abnormal Metabolites Expected</strong></td>
<td>Elevated phenylalanine levels, Normal/low tyrosine (PAA), Normal pterin studies</td>
</tr>
<tr>
<td><strong>If initial testing is negative has the disorder been ruled out?</strong></td>
<td>Yes</td>
</tr>
<tr>
<td><strong>Diagnostic Confirmation</strong></td>
<td>Elevated phenylalanine levels</td>
</tr>
<tr>
<td><strong>Differential Diagnosis</strong></td>
<td>Defects of Biopterin Metabolism, Neonates on Total Parenteral Nutrition (TPN)</td>
</tr>
<tr>
<td><strong>Specific Testing Laboratories as listed in GeneTests (accesses link to GeneReviews if available)</strong></td>
<td><a href="http://www.genetests.com/servlet/access?prg=i&amp;db=genetests&amp;site=gt&amp;id=8888891&amp;fcn=c&amp;qry=2273&amp;res=rous&amp;res=nointl&amp;key=fmnoQgemOKULd&amp;show_flag=c">www.genetests.com</a></td>
</tr>
<tr>
<td><strong>American College of Medical Genetics ACT Sheet</strong></td>
<td><a href="http://www.acmg.net/StaticContent/ACT/Phenylalanine.pdf">http://www.acmg.net/StaticContent/ACT/Phenylalanine.pdf</a></td>
</tr>
<tr>
<td><strong>American College of Medical Genetics Algorithm</strong></td>
<td><a href="http://www.acmg.net/StaticContent/ACT/Algorithms/Visio-Phenylalanine.pdf">http://www.acmg.net/StaticContent/ACT/Algorithms/Visio-Phenylalanine.pdf</a></td>
</tr>
<tr>
<td><strong>American College of Medical Genetics Panel</strong></td>
<td>Core Panel</td>
</tr>
</tbody>
</table>

## Tests to be done by the specialist and expected results for diagnosis

- Elevated phenylalanine levels
- Normal/low tyrosine (PAA)
- Normal pterin studies

## What other conditions might have this NBS abnormality

- Defects of Biopterin Metabolism
- Neonates on Total Parenteral Nutrition (TPN)

## Other Resources

- [American College of Medical Genetics ACT Sheet](http://www.acmg.net/StaticContent/ACT/Phenylalanine.pdf)
- [American College of Medical Genetics Algorithm](http://www.acmg.net/StaticContent/ACT/Algorithms/Visio-Phenylalanine.pdf)
- [Core Panel](#)
Standardization among Newborn Screening Laboratories and Clinical Centers

- All 7 NYMAC states report results to the Region 4 Genetics Collaborative MS/MS database housed at the Mayo Clinic. (D.C. isn’t a state and it doesn’t report; their testing is done by a commercial NBS laboratory)

- 13 laboratory technologists from the 7 states attended MS/MS training at Mayo Clinic

- NYMAC provides funding to 4 Metabolic Centers to report data to the Region 4 Genetics Inborn Errors of Metabolism Information System. We will add more centers in the near future.
Access to Genetic Counseling

Clinical Genetics Providers
(1.5 hour drive-time)
Access to Specialty Care

Metabolic Genetics Providers

Cystic Fibrosis Providers

Endocrine Providers

Hemoglobin Providers
Consumer Knowledge about Newborn Screening and Genetic Conditions

Improving Maternal and Child Health Across the Life Span: Acting Today for Healthy Tomorrows
Genetics and Your Health Brochures

- Prepregnancy
- Prenatal
- Adulthood
- Adolescence
- Pediatrics

Improving Maternal and Child Health Across the Life Span: Acting Today for Healthy Tomorrows
Medical Home Systems

NYMAC funded specialty care centers to develop systems to improve communication with the primary and emergency care providers of their patients

- One metabolic program developed a web-based, encrypted and password-protected personal health history page that could be accessed by approved providers
- A sickle cell program held grand rounds with local pediatricians about the benefits of communication among them and the patient and family
- Another metabolic program used pre- and post-intervention surveys to assess their effectiveness in defining each person’s role in providing care

Generally, there was very little change in communication among providers 😞
NYMAC funded a Teen Transition Program for PKU at Children’s Hospital of Pittsburgh that used modules developed by the Cristine Trahms PKU Program at University of Washington

- This program started with only a few teens, but added more as they progressed. CHP has acquired additional funding to further modify the modules to best meet their program’s needs and to work with more teens with PKU and other metabolic conditions.

Nicole Payne, then the metabolic dietician at CHP, presented this work on the radio program, *Patient Power*, in 2010. The program can be accessed at the *Patient Power* website [www.patientpower.info](http://www.patientpower.info).
Transition from Pediatric to Adult Care

NYMAC is currently funding three transition programs

- Children’s Center for Cancer and Blood Disorders of Northern Virginia for young adults with sickle cell disease
- University of Maryland for young adults with complex genetic diseases
- A.I. duPont Hospital for Children in Delaware for severely disabled young adults

Each program employs a Transition Navigators who works with the young adults and their families

- Transition is a process for both: the young adult must learn self-care and advocacy and the family must learn to let go as much as possible.

They all work with the medical community to identify adult care providers. UMD is using Med-Peds residents who often stay in the Baltimore area after graduation – providing a natural “transition.”
New York State Sickle Cell Transition Program

In 2011, the Sickle Cell Service grants were refocused from case management to Transition.

- Each applicant must have two principal investigators — one a pediatric hematologist and the other an adult hematologist who takes medical responsibility for the young adult upon transition
- The funding is used, primarily, for a Transition Navigator
- Grantees are Brookdale University Medical Center, New York Methodist Hospital and Interfaith Medical Center in Brooklyn, Harlem Hospital in Manhattan and Bronx-Lebanon Medical Center in the Bronx
Do You Remember Her?
Emergency Preparedness for Newborn Screening Laboratories

In 2009 Katrina put Louisiana’s Newborn Screening Program out of service for three years!

- Without a precedent to follow, Iowa Hygienic Laboratory ensured that Louisiana’s newborns were screened
- NYMAC has been working with our state NBS laboratories to get prepared
  - After two table-top drills and a lot of discussion, Specimen Exchange drills will start this spring
Emergency Preparedness for Specialty Care Centers

In the upcoming cycle NYMAC will work with clinicians to ensure that they are prepared

- Accessible patient databases
  - Electronic Medical Record should have all the information the clinician needs to contact the family or to provide current information to another specialist if the family is forced to relocate
- Medical supplies, prescriptions, foods and formulas in case usual access is not possible
Emergency Preparedness for Families

• We will work with families to ensure that they have access to medical records, especially emergency information about current health status, medications and diet requirements, how to contact the specialist

• We will also try to find ways that families can maintain an emergency supply of special foods and formulas as well as prescriptions
  • This is not always possible, since there are restrictions on dispensing, as well as reimbursing, for “excess” medications.
Emergency Preparedness for Families

How to find a specialist in another city or state?

- NYMAC has posted maps of our region that includes the location and contact information for specialists
- The other RCs have similar resources
NYMAC
(New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services)

NYMAC Products

* General Information
  * NYMAC Brochure
  * NYMAC Needs Assessment and Plan
    * NYMAC Directory of Genetic and Specialty Care Services
  * Distance Strategies
    Distance infants and their families must travel to a treatment center for appropriate care with the following conditions:
    * Sickle Cell Disease (SCD)
    * Congenital Primary Hypothyroidism (CH)
    * Phenylketonuria (PKU)
  * Newborn Screening Standardization Guidelines for the clinical evaluation of infants who screen positive by newborn screening:
    * NYMAC Diagnostic Guidelines
    * State Newborn Screening Program Notification Protocols
  * Consumer Education
    * Genetics and Your Health Brochures
      * Pregpearnacy (English) (Spanish)
      * Prenatal (English) (Spanish)
      * Pediatrics (English) (Spanish)
      * Adolescence 11-21 (English) (Spanish)
      * Adulthood (English) (Spanish)

NYMAC States

* Home
* About NYMAC
* Leadership
* National MGC Transition Work Group
* Current Projects
* Subcontracts
* NYMAC Products
* Additional Resources
* Educational Events
* Contact NYMAC
* Funding Opportunities
* State Specific Resources

Improving Maternal and Child Health Across the Life Span: Acting Today for Healthy Tomorrows
NYMAC Genetic and Specialty Care Centers

Clinical Genetic Counseling and Treatment Centers for diseases detected through state Newborn Screening Programs including Metabolic Diseases, Endocrine Disorders, Cystic Fibrosis, Sickle Cell Disease

Public: 444 views
Created on Oct 19, 2009 - By NYMAC - Updated Jan 12, 2011
Rate this map  Write a comment  KML

Al duPont Hospital for Children
1600 Rockland Road, Wilmington, DE 19899 Website: www.nduports.org Genetics (302) 651-5916 Metabolic (302) 651-5916 Cystic Fibrosis (302) 651-8400 Endocrine (302) 651-4000 Hemoglobinopathy (302) 651-5...

Children's National Medical Center
111 Michigan Avenue, NW, Washington, DC 20010 Website: www.dccchildrens.com Genetics (202) 476-2187 Metabolic (202) 476-2187 Cystic Fibrosis (202) 476-2187 Endocrine (202) 344...

Georgetown University Hospital
3800 Reservoir Road, NW, 2 PHC, Washington, DC 20007 Web www.georgetownuniversityhospital.org Genetics (202) 444-8518 Cystic Fibrosis (202) 444-0011 Endocrine (201)...

Howard University Hospital
2041 Georgia Avenues, Washington, DC 20060 Website: www.huhcare.com Genetics (202) 885-3022

Herman and Walter Samuelson Children's Hospital at:
2101 West Belvedere Avenue, Baltimore, MD 21215 Website: www.lifebridgehealth.org/chs.cfm?id=1039 Endocrine (410) 691 8331 Hemoglobinopathy (410) 691-5854

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1600 Rockland Road, Wilmington, DE 19899 Website: www.nemours.org Genetics (302) 651-6515 Metabolic (302) 651-6515 Cystic Fibrosis (302) 651-6400 Endocrine (302) 651-6400 Hemoglobinopathy (302) 651-6515

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Georgetown University Hospital
3800 Reservoir Road, NW, 2 PHG, Washington, DC 20007 Website: www.georgetown.universityhospital.org Genetics (202) 444-8518 Cystic Fibrosis (202) 444-6011 Endocrine (202) 444-6011

Howard University Hospital
2041 Georgia Avenue, Washington, DC 20050 Website: www.huh.org Genetics (202) 866-3022

Herman and Walter Samuelson Children's Hospital at:
2461 West Belvedere Avenue, Baltimore, MD 21215 Website: www.lifefh.org/chs2fhs Website: www.lifesbridgehealth.org Website: www.lifesharehealth.org Genetics (410) 361-8334 Children's National Medical Center (202) 476-2187

Improving Maternal and Child Health Across the Life Span: Acting Today for Healthy Tomorrows
Diagnosis: Classic Galactosemia

In an emergency, call the Metabolic Specialist immediately.

Metabolic Specialist: ________________________________
Phone: (__) -____; Fax discharge summary (__) -____
Primary Physician: ________________________________
Phone: (__) -____; Fax discharge summary (__) -____
Patient Name: ____________________________________
Address: _______________________________________
City, State, Zip: _________________ Phone: (__) -_____  

NYMAC - New York – Mid-Atlantic Consortium for Genetic and Newborn Screening Services

Diagnosis: Classic Galactosemia

Galactosemia is an inherited disorder that prevents the body from breaking galactose and lactose (found in milk products) into glucose. Untreated galactosemia causes liver damage, failure to thrive, seizures, sepsis, and mental and growth retardation.

If hospitalized, do not provide foods containing milk products.

American College of Medical Genetics (ACMG) ACT Sheets.
www.acmg.net/resources/policies/ACT/condition-analyte-links.htm
Insurance Coverage for Formulas and Metabolic Foods

NYMAC participated in a multi-regional study of use and insurance coverage of medical foods, supplements and low-protein foods by children with metabolic conditions for the SACHDNC Follow-up and Treatment Subcommittee

• The data show that nearly all children had insurance coverage
• Most children use medical foods and low-protein foods
• Generally medical foods are covered by insurance, but low-protein foods are not
Some of NYMAC’s Future Activities

• Develop collaborations between safety-net providers (FQHCs and SBHCs) and specialty centers
• As mentioned earlier, fund pilot telemedicine projects for NBS follow-up and medical management
• Engage insurers – Medicaid programs and private companies – in consideration of the benefits to insurers and insured of adequate coverage
• Continue Transition activities as well as engage additional adult care providers
• Ensure that genetics and NBS are integrated into the state departments of health
• Assist the NBS programs and health departments to implement the SACHDNC-recommended NBS panel
• Continue working with the NBS labs to standardize and expand to include NBS follow-up programs
• Continue and expand Emergency Preparedness activities
• Strengthen partnerships with other HRSA-funded programs such as LEND, Hemophilia and Thalassemia Centers
Questions?
Thank you to HRSA/MCHB

Thank you to all the NYMAC Champions and other Partners

Thank you to AMCHP for this opportunity to share Information about NYMAC
Transition Intervention Program

Mitzi K. Glass, LCSW
Sickle Cell Transition Coordinator
Children’s Hospital Of The King’s Daughters

AMCHP
February 13, 2012
Introduction

• “Transition is a process that addresses the medical, social, educational and vocational needs of patients as they leave pediatrics to adult medical care.”

• This presentation will briefly describe the Transition Intervention Program. (T.I.P.)
Comprehensive Sickle Cell Program

- Located in the CCBDC at CHKD
- Program began approximately 20-25 years ago
- We serve approximately 500 patients between the ages of birth-21 years old
- Referrals received from Newborn Screening, community pediatricians, self referrals
- We have the largest catchments area of sickle cell patients in Virginia. 50% of referrals
- Staff includes medical director, physician, nurse practitioner, two nurse coordinators, social worker, educational consultant and a program manager and transition coordinator
Transition Intervention Program

“Healthy People 2010 established the goal that all young people with special health care needs will receive the services needed to make necessary transitions to all aspects of adult life.”

- Dr. William Owen submitted a grant proposal to The Virginia Department of Health in May 2007

- A grant was awarded to hire a Transition Coordinator and develop a Sickle Cell Transition Program
“The Comprehensive Sickle Cell Program at Children’s Hospital Of The King’s Daughters needs a more formalized program to facilitate the successful transition of young adults (ages 17-21) from pediatric to adult care, currently estimated at 100 patients over the next four years. To meet these needs, we propose to establish a full time transition coordinator position and to develop a transition pathway to coordinate the multidisciplinary efforts in the transition process.”
Transition Program Targets

• A plan to partner with adult care professionals, community based sickle cell programs, the patient and the family in an effort to develop a transition focused intervention program that addresses:
  • Medical
  • Social
  • Employment and Vocational Training
  • Educational
The Model

- Health Care Skills Checklist/Teen Self Assessment Form
- Transition Coordinator Assessment Healthcare Skills Checklist; tracks transition readiness
- Adaptations from Massachusetts Initiative for Youth with Disabilities and Healthy and Ready to Work Project.
Transition Intervention Program

Age 15

• The program is explained to the parent and patient

• The teen self assessment is completed during comprehensive clinic appointment

• The Transition coordinator assessment is started

• Educational materials are distributed to the patient
Age 16 – 21

- Transition Coordinator Assessment updated at every clinic appointment; team updated on transition issues

- Interface with TC: inpatient, outpatient follow up, comprehensive clinic, emergency room and telephone

- Patients seen without parent(s) at age 18

- Patients seen every 6 months at age 17
Age 20-21

- Patient staffing led by transition coordinator
- Adult provider identified with patient
- Appointment with new provider made by or with TC; transition summary completed by provider
- TC accompanies patient to first appointment
Follow up

• Phone call made to patient

• Patient contact TC for additional services

• TC receives dictated progress note from new provider
Challenges

Preparing patients for the differences between pediatric and adult care

• Narcotic use; Medication compliance
• Keeping appointments
• Accessing care
• Transportation
• Life expectations and responsibilities

• How is transition success defined?
Conclusion

“Your problem is to bridge the gap which exists between where you are now and the goal you intend to reach.”

Earl Nightingale
Thank You!

Katharine Harris, MBA

Association of Maternal and Child Health Programs

Questions????