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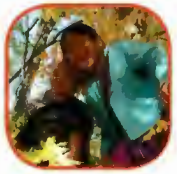
The mission of the New England Genetics Collaborative is to improve the health and well-being of those living with genetic conditions in New England and nationally.



www.negenetics.org

U.S. Department of Health and Human Services
HRSA
Health Resources and Services Administration

Institute on Disability / UCED
A University Center for Excellence on Disability

University of New Hampshire

Family Contributors to GEMSS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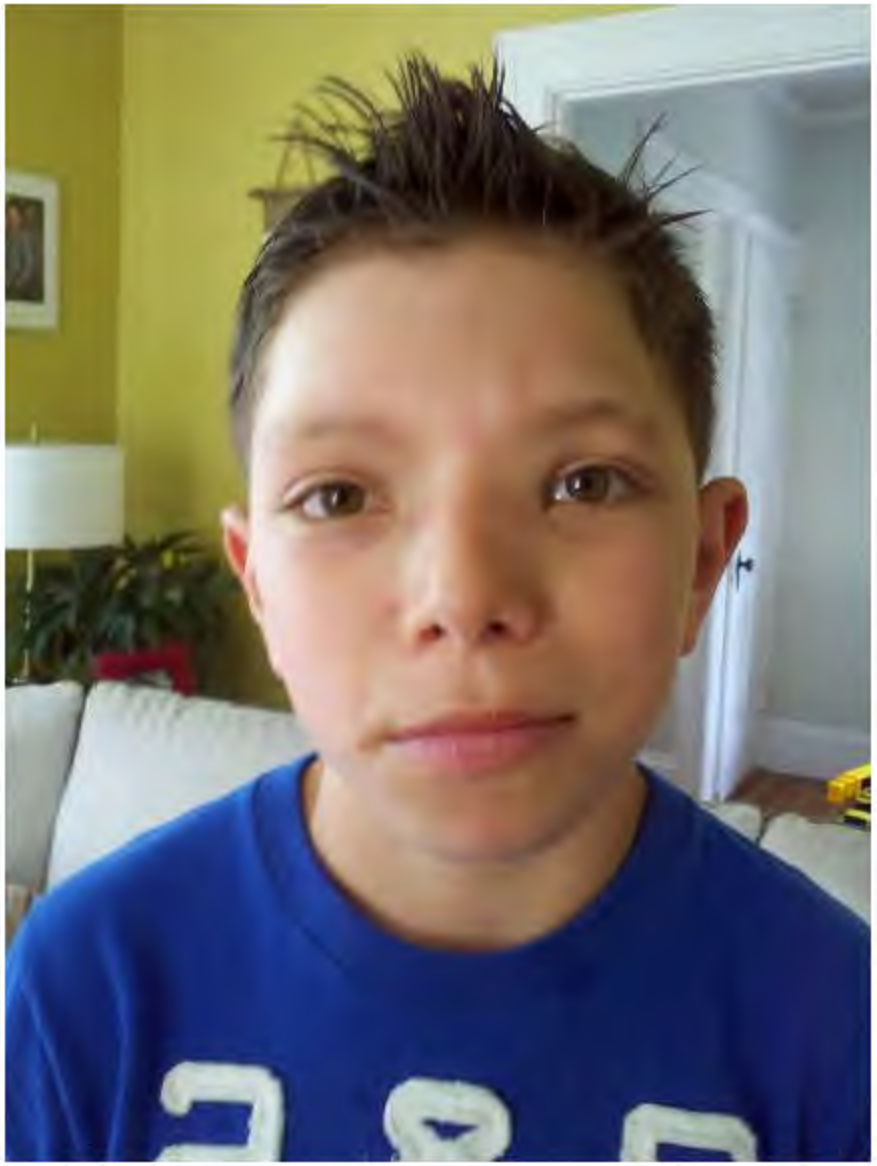

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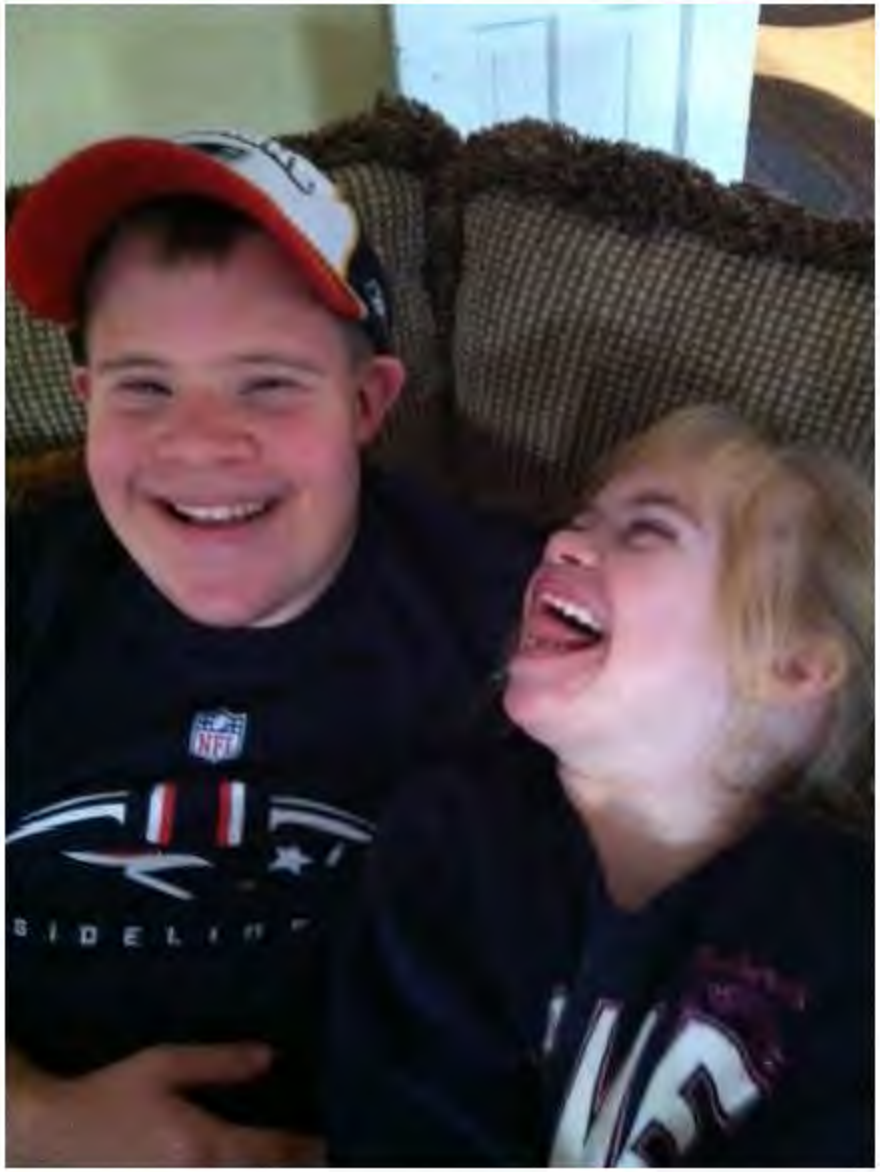

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Belated
Happy 9th
Birthday
Sara!


1st day
of

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OLDNAVY
Athletics

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Ancient History
(prior to 2012)

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"Newborn Screening Saves Lives Act"

April 24, 2008

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QUALITY ASSURANCE WORK GROUP

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Commonwealth Medicine

UMASS MEDICAL SCHOOL

Applied Knowledge in Public Service

Activities & Accomplishments:

Establishment of the Laboratory Quality Assurance Collaborative (LQAC):

- Participants from the two NBS laboratories in Region 1: (Connecticut and the NENSP, serving MA, ME, NH, RI, and VT)
- NBS laboratories in New York and Wisconsin (inter-regional collaborators)

Data Collection and Analysis:

- NENSP has created templates that convert each program's natural data exports into a standard format to facilitate analysis and further comparisons.
- Each state has contributed data (comprising of values of all markers analyzed by their programs) from babies with a confirmed diagnosis of one of the disorders associated with propionylcarnitine (C3) elevations (propionic acidemia, methylmalonic acidemia, mixed carboxylase deficiency, and the cobalamin defects).
- Wisconsin has provided data on all babies determined by their laboratory to have a "not normal" C3 concentration but on further confirmatory testing were concluded to be false-positives. Other collaborators are in the process of assembling the data on their "false-positives."
- The applicability of indices developed by NENSP to improve sensitivity of C3 screening has been tested on metabolic profiles of true positives.

Next Steps:

- A similar process will be applied to metabolic profiles of the false positives and for out-of-range initial newborn screens for C0, C4, C5/1, C5, C5OH, C5DC, C5-3M-DC, C8, C14, C14:1, C16OH, C18:1 OH, C16, C18:1, PHE, LEU, MET, ARG, ORN, TYR, and CIT.
- The algorithm followed to derive the indices at the NENSP will be recreated using raw laboratory data from each participating laboratory to account for any differences in instrumentation, methodologies, reagents, controls, and environment that will result in different analytic parameters (means of normals, standard deviations, dose-response, etc.).

Working Group Description & Mission

Focus 1 Award entitled: Multicenter Validation of Algorithms to Improve Communications of Positive Newborn Screening Results to the Medical Home

The Quality Assurance Workgroup, led by Dr. Roger Eaton, Director of the NENSP is working on a multicenter validation of algorithms to improve communications of positive NBS results to the medical home. A universal challenge for newborn screening programs nation- (and world-) wide is to strike an appropriate balance between sensitivity and specificity, so that 1) babies with actual disorders are not missed by screening and 2) parental stress and medical system overload due to false positive screen notifications is minimized. Dr. Indumeti Sahai, Chief Medical Officer of the NENSP, has developed algorithms to categorize tandem mass spectrometry (MSMS) results to better discriminate between false positives and true cases, improve the clarity of communications to the medical home, and to better target the use of scarce specialty care resources.

The aim of the Workgroup is to gain objective evidence as to the general applicability of the NENSP algorithms to other NBS laboratories in Region 1 and across the country. Validation of the universality of the application of the NENSP algorithms is only possible through collaborative studies with other NBS laboratories, so that the algorithms may be tested against independent data sets acquired using the variety of MSMS methodologies in use today.

Why it's important:

NBS demands systems-wide approaches that seamlessly link multiple components. One key link is the communication that translates raw laboratory results into effective medical care. The workgroup is helping to provide an objective means for NBS follow-up staff to appropriately communicate lab results to the provider.

Goals & Objectives for 2008-2009

- Continue to explore additional indices that may be examined for general usefulness
- Apply a similar approach to the analysis of CAH data with the goal of developing new indices to improve specificity and quality of communications of out-of-range 17-hydroxyprogesterone results to the medical home.
- Collaborative Conference to be held in Boston with representatives from each collaborating site (NENSP, Connecticut, New York and Wisconsin)

Collaborations

- Connecticut Public Health Laboratory
- New York State Department of Health, Wadsworth Center
- Wisconsin State Laboratory of Hygiene

Contact Information

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MD, WI NBS
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Gary Hoffman, B.S., WI NBS

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LONG TERM FOLLOW UP

PURPOSE & DESCRIPTION

The LTFU Workgroup is working to establish a sustainable regional approach to ensuring 1) that infants and children with NBS conditions continue to be engaged in optimized, state-of-the-art lifespan and family-centered care, 2) that newborn screening systems have evidence in hand for quality improvements in the care of these clients and 3) that the same evidence is available for sound policy decision-making that benefits the population at large. The approach builds upon the success and infrastructure of existing public health NBS systems, inclusive of the New England Newborn Screening Program's regional database and collaboration among 5 New England state Departments of Health.

IMPORTANCE

The purpose of a NBS program is to identify all infants who will benefit from early intervention to reduce death, mental retardation and other significant health problems through their continued engagement in effective care. We know that many usual life circumstances (family move, change in employment and provider) can have significant impact on continued care. We also know that the spectrum of long-term outcomes for children identified by NBS has yet to be defined for many conditions or linked to a particular approach in care. A sustainable system to address these issues is needed.

WORK GROUP MEMBERS

Anne Marie Comeau, PhD, CHAIR
New England Newborn Screening Program
University of Massachusetts Medical School

Roger B. Eaton, PhD
New England Newborn Screening Program
University of Massachusetts Medical School

Elle Mikkelsen
Maine Newborn Screening

Indermeet Sahai, MD
New England Newborn Screening Program
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Cindy Ingram
Vermont Newborn Screening

Marcia Lavochkin
New Hampshire Newborn Screening

Barbara McNeely
Rhode Island Newborn Screening

Janet Panek
Massachusetts Department of Public Health

Kristen Campagna
RI Newborn Screening

Collaborating with

The New England Hemoglobin Clinics

The New England Cystic Fibrosis Centers

The New England Metabolic and Genetics Clinics

FEATURED ACTIVITIES

- Meetings of "condition" specific NBS workgroups continued over the course of the year in order to engage specialists caring for infants and children diagnosed with newborn screening conditions to develop and refine data collection tools and variables.
- Completed data collection and analysis for a LTFU project on children diagnosed with long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD) by NBS (by Dr. Indermeet Sahai)
- Under the direction of Dr. Sahai, began data collection for a project to evaluate the long term metabolic outcomes of children identified with Short chain acyl-CoA dehydrogenase (SCAD)
- The Hemoglobin Workgroup hosted the "Surviving to Thriving: Improving Long-term Outcomes in Sickle Cell Disease" conference in September 2010. The event was attended by over 100 people and brought together experts from around the county to identify best practices for improvements to patient care.

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Health Care Transition

Exploring the Role of the School Nurse as a Facilitator of Health Care Transition

University of Massachusetts Boston
Carol Orton, RN

UMASS BOSTON

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Genetics, Genomics and Public Health

Established Programs and New Frontiers

Amy Schwartz, MPH

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Haiku Contest Entries:

oh GINA, GINA
our insurance and our jobs
are secure with you

Eye shadow, lipstick
and hair dye cannot disguise
Genetic Makeup

Nurture and nature
Effects double stranded forms
Thus, evolution

Genetics we have
Throughout our family tree
the future may be

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American Journal of Medical Genetics Part C (Seminars in Medical Genetics) 151C:241–254 (2009)

ARTICLE

Improving Genetic Health Care:
A Northern New England Pilot Project Addressing the Genetic Evaluation of the Child With Developmental Delays or Intellectual Disability

JOHN B. MOESCHLER,* R. STEPHEN AMATO, THOMAS BREWSTER, LEAH BURKE, MARY BETH DINULOS, ROSEMARIE SMITH, WENDY SMITH, AND PATRICK MILLER

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MAKING COLLABORATIVE CO-MANAGEMENT EXPLICIT AMONG PARENTS, PRIMARY CARE PROVIDERS, AND SPECIALISTS

INTRODUCTION

When primary care providers (PCPs) refer children to specialists, communication between physicians and with parents is frequently a problem. For children with metabolic or other genetic conditions, this problem is particularly important, because these complex disorders are lifelong and may require frequent specialty referrals and careful management. Most parents are willing to take an active role in information transfer between their child's providers. However, studies have not described how to include parents in this process. This project proposes a care planning tool intended to improve the collaboration and teamwork among physicians and families.

METHODS

The two specific aims of this study are:

1. To complete the development of an intervention to improve communication among parents, specialists, and the primary care medical home.
2. To test the feasibility of this intervention in two pediatric practices.

RESULTS & DISCUSSION

Five physician focus groups and four parent experts were used to develop the intervention form. The Evaluation phase was launched in two PCP practices and three pediatric specialties at UMass (genetics and metabolism, neurology, and endocrinology). Only one patient was enrolled in this study because there were very few new referrals who were eligible. While acknowledging the value of the communication tool, PCPs would forget to use the form, and PCPs were reluctant to take the extra time necessary to complete the form.

Because Dr. Stille left UMass on August 1, 2010, Susan Waisbren, PhD, has adapted the form to test its use at Children's Hospital Boston. That clinic continues to pilot the form and provide recommendations for modifications.

A number of systemic barriers must be overcome before this care plan tool can be practical in busy practice settings. Integration of this plan into an electronic medical record and ensuring that practices have a care coordinator available are two strategies that are recommended for future testing. A quality improvement approach to implementation may improve uptake and efficacy.

Commonwealth Medicine
UMASS MEDICAL SCHOOL

Principal Investigator:
Christopher J. Stille, MD, MPH
UMass Medical School

Co-Investigators:
W. Carl Cooley, MD, Ctr. for Medical Home Improvement
Susan Waisbren, PhD, Children's Hospital Boston
Beth Dworetzky, MS, Parent Consultant

STATE LAWS OF NEW ENGLAND: USE AND DISCLOSURE OF GENETIC AND NEWBORN SCREENING INFORMATION

FOR THE PURPOSES OF TREATMENT, A REGISTRY, AND RESEARCH

MICHELLE M. WINCHESTER, J.D.

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U.S. Department of Health and Human Services
HRSA
Health Resources and Services Administration

HRSA Update; December 2010

Sara Copeland, MD
Medical Officer, Genetics Services Branch
Health Resources and Services Administration
Department of Health and Human Services

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An Educational Training Facility
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529 Chestnut Street

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Strategic Planning Recommendations

From HRSA's Regional Genetics and Newborn Screening Service Collaboratives and their National Coordinating Center

Facilitated by Peter Antal, Ph.D.
Institute on Disability, UNH

Presentation to the NEGC Collaborative Council
April 20, 2011

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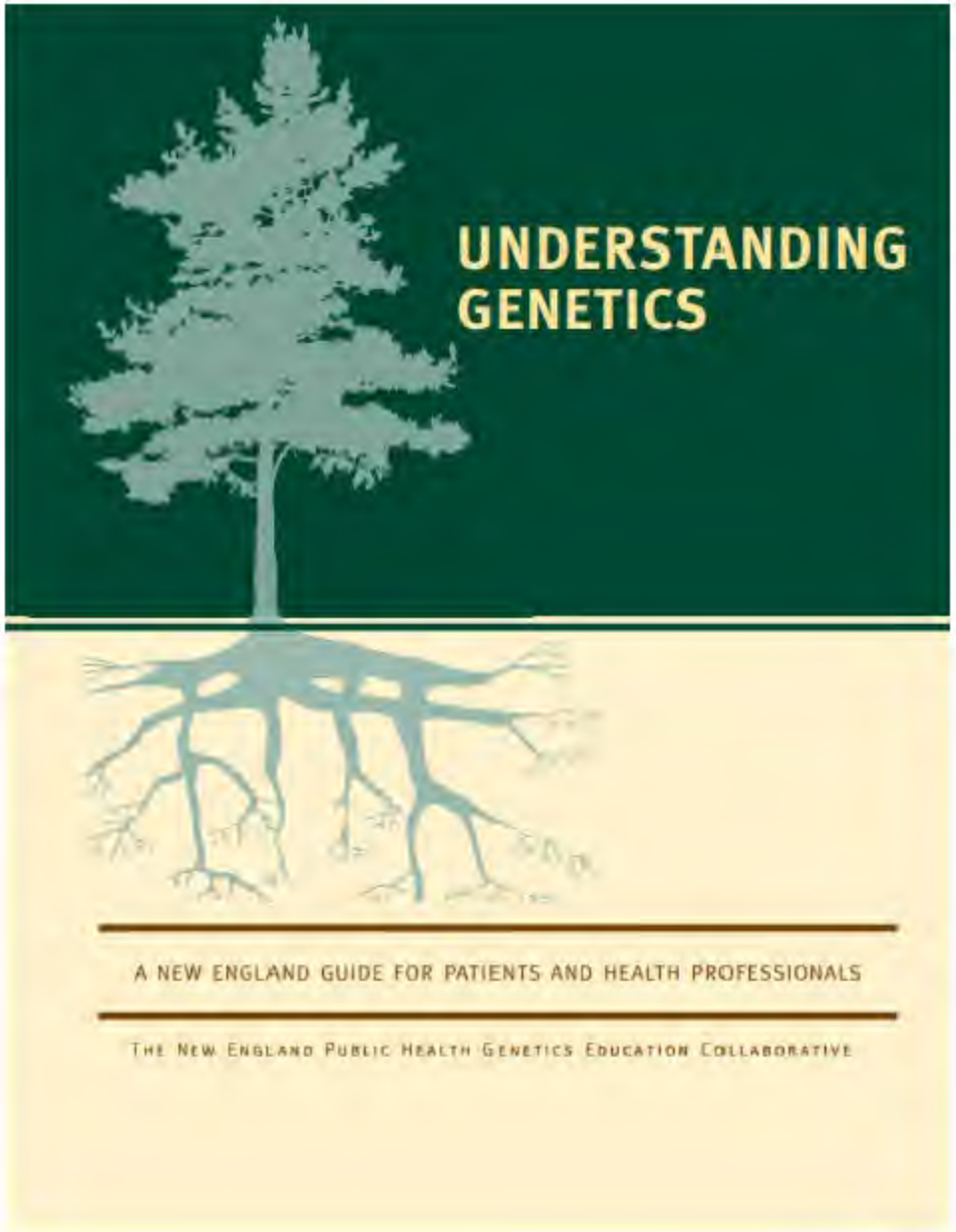

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اختبارات فحص الاطفال حديثي الولادة:
قد تنقذ حياة طفلك

Sponsored in part by a grant from the Genetic Services Branch of the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA) and the New England Regional Genetics and Newborn Screening Collaborative, HRSA Grant #1U22MC03959

2008

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Newborn Screening

A Guide for Prenatal Educators

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PKU TOOLKIT

Diet for Life!

Supported by Children's Hospital Boston, Applied Nutrition & The New England Consortium of Metabolic Programs

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- Lifestyle
- Maternal PKU
- Checklists
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Welcome to the PKU Toolkit

Congratulations! If you are a teen or a young adult with PKU, then you have just taken a step in the right direction. Whether you are already managing your diet or just returning to it, this PKU Toolkit will help you on your way to better PKU control and better health!

Here are some links to get you started:

- PKU
- Diet
- Insurance
- Transition
- Lifestyle
- Maternal PKU

About the ToolKit

This Toolkit is designed to be your personal guide for managing PKU. In the Toolkit you'll find information and resources that we think are important for young adults with PKU, and which can make your life easier.

You'll find information about diet, exercise, travel, and living a healthy lifestyle. And also about transitioning to life as an adult, including how to handle doctor visits and health insurance.

For young women who want to become pregnant some day, there's a section on maternal PKU, and how to keep your baby healthy.

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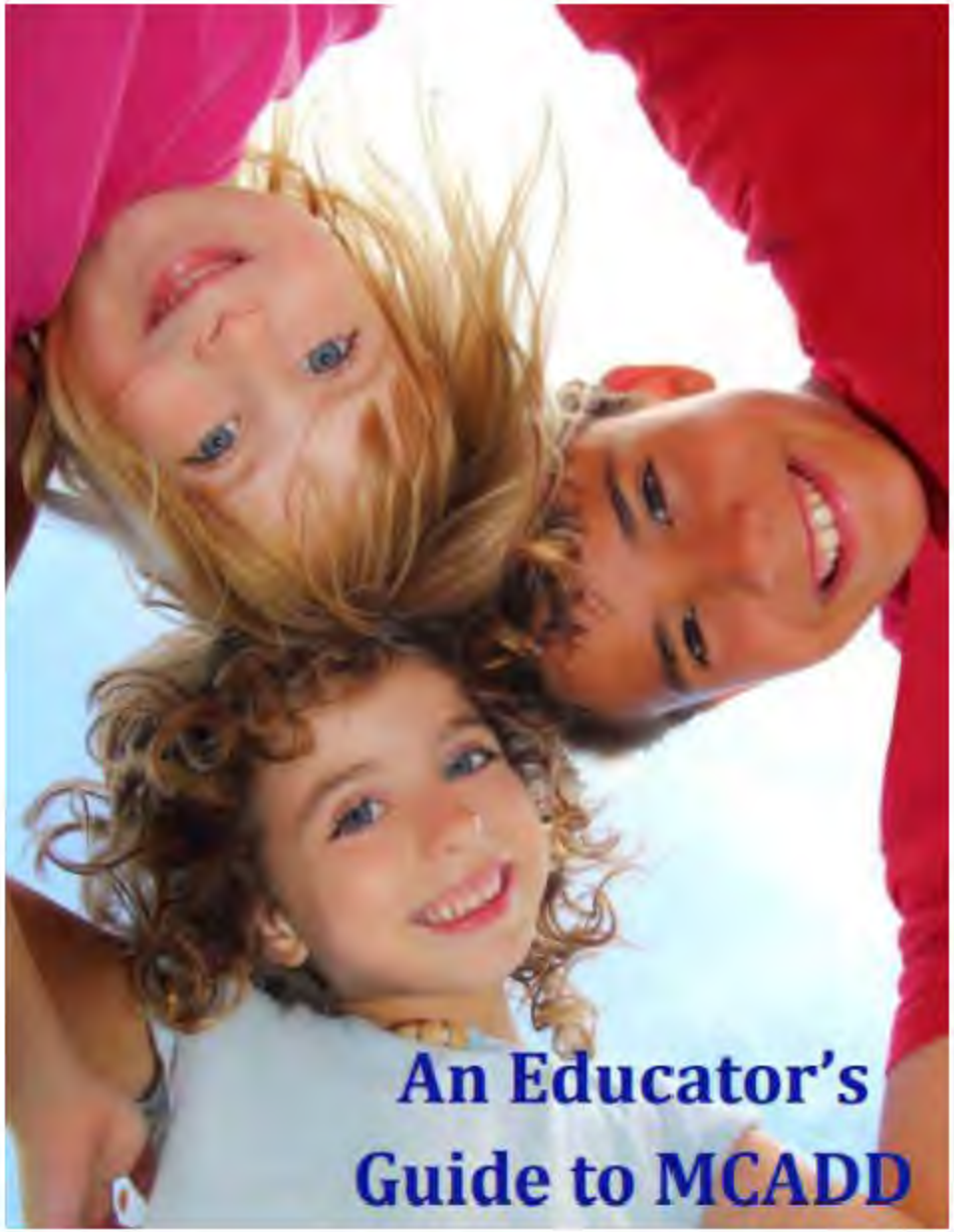

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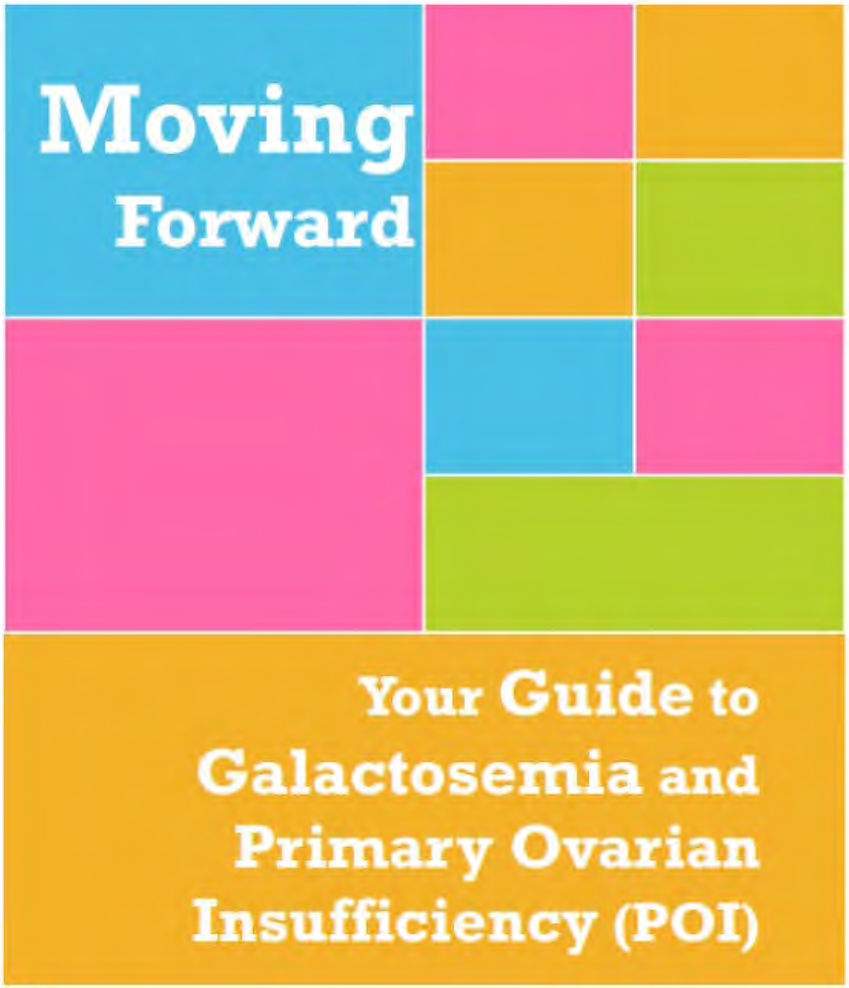

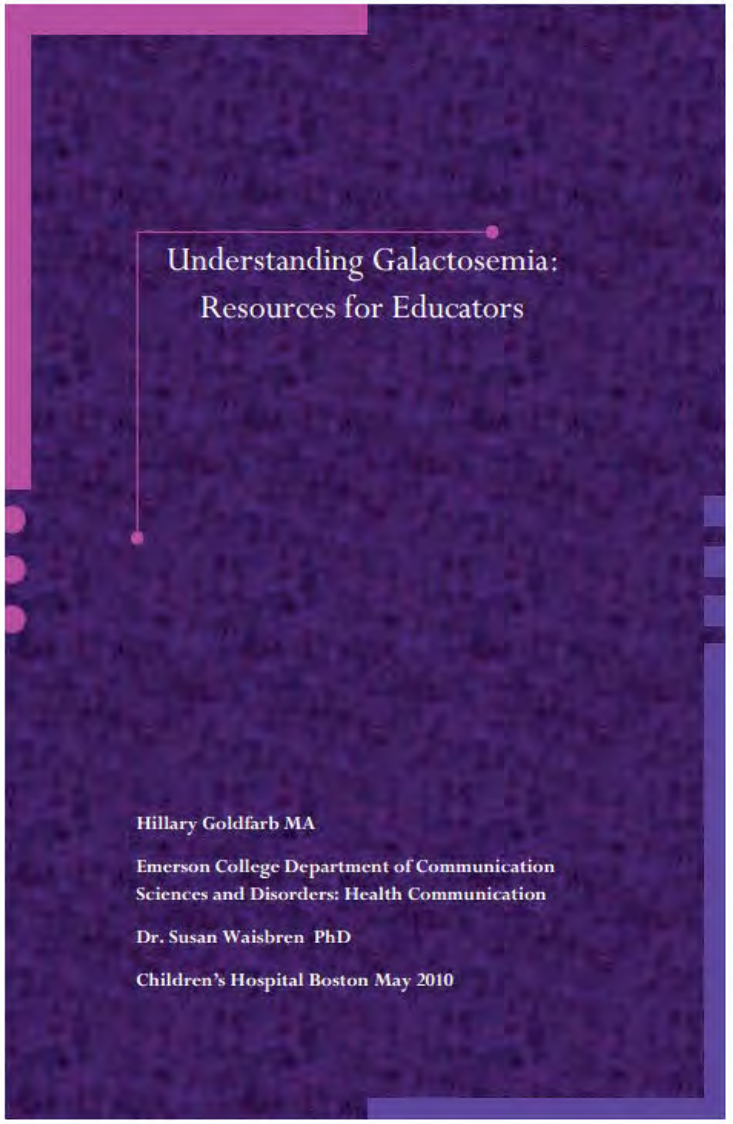

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Galactosemia

Understanding Galactosemia

Hillary Goldfarb, MA, Emerson College
Dr. Susan Waisbren, PhD,
Children's Hospital Boston

Children's Hospital Boston

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PREVENTION OF BIRTH DEFECTS IN NEW ENGLAND THROUGH REGIONAL COLLABORATION

INTRODUCTION

Initiated in 2009, the New England Birth Defects Consortium (NEBDC) is a regional collaboration of New England states (Connecticut, New Hampshire, Maine, Massachusetts, Rhode Island, Vermont) with the shared mission of improving services for infants and children in New England with birth defects.

The goals of the NEBDC are to promote regional collaboration through: (1) data sharing, (2) research activities, (3) prevention activities, and (4) health care quality improvement.

METHODS

The NEBDC used evidence-based methodology to initiate a prevention initiative through an organized campaign of folic acid containing, multivitamin distribution for the purpose of:

- Increasing awareness about folic acid for women of childbearing age in the New England states
- Increasing the number of women of childbearing age who take a multivitamin with 400 micrograms of folic acid
- Evaluating the implementation and impact of a standardized prevention activity across states

To implement this project, the NEBDC proposed a target population from the WIC (Women, Infants and Children) programs in each state.

The protocol included:

- Proposed implementation in at least 1 WIC site per state
- Provide "goodie bags" to each woman (*not currently pregnant*) with free 3-month supply of multivitamins containing 400 micrograms of folic acid
- MA, CT and RI targeted Hispanic women
- One page pre and post intervention surveys in English and Spanish
- Educational materials consisting of pre-printed brochures from the CDC
- Pilot program would last for 12 months from January through December 2011

RESULTS & DISCUSSION

To date, all six New England states have implemented multivitamin distribution sites from their state WIC program offices. Valuable alliances have been created in states that had not previously worked with their state WIC offices for birth defects prevention activities. The project is still ongoing, but as of September 2011, over 1500 surveys have been received for analysis. Preliminary data shows that the target population (women of childbearing age) were reached with this initiative and that use of both a Spanish and English survey have contributed to gathering data on the success of this pilot program from a diverse population. Full data analysis will be complete in January 2012 and it is the intention of the NEBDC to publish results in a peer reviewed journal.

www.nebirthdefects.org

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A QUALITATIVE STUDY TO EXPLORE THE KNOWLEDGE AND ATTITUDES OF SELECTED DIVERSE POPULATIONS ABOUT GENETICS & GENETIC SERVICES

INTRODUCTION

Background: This project was based on the assumption that there is a lack of knowledge and awareness about genetics and genetic services in diverse populations which can contribute to lack of utilization of genetic services and thus decrease opportunity for identifying and preventing diseases and conditions including cancer, cardiovascular disease, some chronic illness, genetic disease and birth defects.

Goal: The goal of the study was to explore the knowledge and attitudes of a sample of diverse clients about genetics and genetic services.

METHODS

Design: Qualitative Descriptive Study using Focus Group Methodology

A community facilitator from each program (Haitian Public Health Initiative (HAPHI) in Mattapan, Massachusetts and the Somali Development Center (SDC) in Boston, Massachusetts) was identified and assisted in recruitment of participants and acted as an interpreter for the focus groups

RESULTS & DISCUSSION

Participants

The participants at HAPHI and the SDC were very different. The HAPI group consisted of middle age men and women who had a developmentally disabled child or adolescent.

The women in both focus groups at the Somali center were younger and did not disclose whether or not they had a child with a disability. There were no men in the Somali group.

1. Overall the most obvious finding is that the participants from both communities did not have any knowledge about genetics or the role genetics plays in health and disease prevention.
2. Since attitudes are formed based on subjective evaluations of an object or concept, no attitudes either positive or negative were identified.
3. The Haitian group was made up of parents that had a child with a developmental disability. Several participants asked the focus group leader specific information about their child and diagnosis. Several were still seeking a reason for the disability.
4. No one in any of the three groups had, to their knowledge, any genetic workup or referrals to genetic services. The majority of participants in both groups did have health insurance and a primary care provider.
5. Participants in all three groups requested more information on genetics and how to access services.
6. The Somali women were aware of newborn screening but did not know the role it played on prevention and health care.

Recommendations

1. The education about genetics needs to be culturally targeted and sensitive.
2. The education needs to be developed in collaboration with members of the diverse communities, based on a well-developed and culturally sensitive needs assessment tailored to each of the communities.
3. There is a need for education about the role of genetics in disease prevention.
4. There is a need for education about the availability and role of genetic services for these populations including location, when to request them, and insurance coverage.

Principal Investigator
Patricia Rissmiller RN, DNSc, PNP
Associate Professor, Simmons College

David T. Helm, PhD
ICI/LEND Director, Children's Hospital Boston

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NEW PARADIGM OF INTEGRATED HOME CARE MANAGEMENT FOR ADULTS WITH SICKLE CELL DISEASE

INTRODUCTION

- Sickle cell disease (SCD) is a lifelong complex disorder is a chronic illness that lends itself to the Chronic Care Model (CCM)
- SCD complications including acute and chronic pain, acute chest syndrome, stroke, transient ischemic attacks, asepbc necrosis of the hip, renal failure, and blindness have resulted in organ damage, debilitating disabilities and untimely death
- Adults with SCD often suffer from fragmented care and are transitioned to the adult health care system that is usually ill prepared for their complex health care needs
- The negative impact on the adult's level of self-efficacy and sufficiency, coping and self-care management skills fosters episodic care leading to poor health outcomes and poor health-related quality of life (HRQOL
- Several studies have demonstrated successful chronic disease management utilizing the CCM to enhance health outcomes

PURPOSE

The goal for this project is to facilitate and improve the home health care, functional status and health related quality of life of adults 16 years and older with sickle cell disease (SCD) in Connecticut.

AIMS

1. Appraise and pilot the home care service needs of adults with SCD both episodic and chronic
2. Demonstrate a seamless sharing of client health information concerning acute-on-chronic care and coordination needs through Electronic Health Records (EHRs) or paper/fax to promote continuity of care and HRQOL
3. Evaluate the impact of the pilot integrative home health care project on HRQOL and disseminate finding to key stakeholders and at local, regional, and national forums

METHOD

- A quasi- experimental cohort study guided by the Community Based Participatory Research methodologies
- Identified and interested adults were consented by the CBOs with authority to share information with identified service/study partners, the participants complete baseline HRQOL survey
- Medical providers write prescriptions for home health care services
- Enrolled participants receive integrative home care health assessments from one of the two home health care agency partners close to their residence
- Home health care agency develops care plan based on the Outcome and Assessment Information Set (OASIS) with participants and medical providers, sends de-identified form to PI
- Recommended services such as wound care, IV fluid, port flush are billed to insurance company and uncovered services are catalogued
- HIPPA compliant password protected data are available for clinical decision making through shared access.
- Enrolled patients will receive a quarterly $20 gift cards (up to $75) as incentive as well as the four consumers (two each from the North and the South) on the project team for attendance at the quarterly team meetings.

References

RESULTS & DISCUSSION

- Obtained IRB approval from the University of Connecticut Health Center and approved by the UNH
- The two CBOs have identified a total of 18 adults with SCD who are interested in participating in this project.
- Recruitment began at the end of October after all the contracts were signed with the project partners
- Eight adults with SCD have been consented for the study and are awaiting prescriptions from their medical providers.
- The participants will have varying periods of time in the program based on time of enrollment, assessments and service needs

This pilot study is an integrative home health care service model with a new paradigm of care that is holistic and promotes continuity of care as proof of concept for adults with SCD. The ultimate goal is that this model will maintain or improve HRQOL, prevent or mitigate health problems and reduce ED admission and urgent ambulatory care. We anticipate an improvement between the participants' prior year hospital emergency admissions, day treatment and inpatient trends; functional status and HRQOL on enrollment and over the course of the project will be collected and analyzed.

Principal Investigator: Victoria Osborne, APRN, BNP
Co-Investigator: Virginia Pontillo

Partners: Citizens for Quality Sickle Cell Care-Sickle Cell Disease Association of America, Northern CT-Chapter, Inc., Sickle Cell Disease Association of America, Southern CT Chapter, The University of Connecticut Health Center, Farmington; The Masonicare Partners Home Health & Hospice (Hartford); The Visiting Nurses Association of South Central Connecticut (New Haven) and Health Education and Research (HLER) consultant (Boston).

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Proverb Contest Entries:

Ask not what your genes have done to you, but what you can do with them?
Amy Schwartz

Genes are not destiny, but suggestions.
Amy Schwartz

Don't get the blues if your pair of genes don't show up soon.
Marinell Newton

Beware the shallow pool.
Wendy Smith

Confidentiality: What is discussed with the patient, stays with the patient.
Mary-Frances Garber

If we don't cultivate genetic newborn screening, babies may die from this complication.
Vine Samuels

2012-2013

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Laboratory and Clinical Challenges of Whole Genome/Exome Sequencing

David T. Miller, MD, PhD

Assistant Director, Genetic Diagnostic Lab, *Children's Hospital, Boston*

Clinical Geneticist, Division of Genetics, *Children's Hospital, Boston*

New England Genetics Collaborative

April 5th, 2013

Children's Hospital Boston

Harvard Medical School

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Implications of Genome Sequencing on Public Health: Promise and Pitfalls

Susan Estabrooks Hahn, MS, CGC

John P. Hussman Institute for Human Genomics,
University of Miami, Miller School of Medicine

HUMAN GENETICS

UNIVERSITY OF MIAMI
MILLER SCHOOL OF MEDICINE
HUSSMAN INSTITUTE
for HUMAN GENOMICS

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Boston Children's Hospital Transition Toolkit

Welcome to the Boston Children's Hospital Transition Toolkit!

As a teenager getting ready for adulthood, or as a young adult, you can begin to take control of how you handle your metabolic condition and your health in general. This Toolkit is designed to help you! To start, read through these materials and fill out the forms. You can also begin to look for an adult-focused doctor to replace your pediatrician who only deals with childhood problems. This process of switching to an adult-focused doctor is called *medical care transition* and takes time and planning—but once it's done, it will be great for your future health and success.

Use the following forms to help achieve your medical care transition

Health Readiness Assessment

Metabolic Condition Basics

Medical Health Summary

Transition Plan

What am I supposed to do with these forms?

Measure your health independence by taking the Health Readiness Assessment and over time try to answer yes to every question.

Read up on your Metabolic Condition Basics and discuss possible adult-health problems with your doctors or nurses.

Ask a doctor or social worker for help filling out the Medical Health Summary and Transition Plan at your next appointment.

Save these forms on your computer and USB flash drive and bring the Transition Toolkit to every medical appointment.

Keep the flash drive in your wallet or purse and save any changes or updates.

Save these forms and bring them to future medical appointments

Ask a doctor to help you fill out your forms

How will The Transition Toolkit help me? With the Toolkit you can –

Decide when you're ready to act as your own health spokesperson.

Prepare for doctor's appointments and fill out health forms.

Talk to doctors and nurses about your medical condition and what roles they should play in your medical care.

Keep track of all of your important medical and condition information.

To get started, fill out the Health Readiness form.

This project is supported by the New England Genetics Collaborative and their cooperative agreement with the US Health Resources and Services Administration (HRSA), grant # U22MC16960.

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2013-2014

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Children with Genetic/Metabolic Conditions in Educational Settings

negc
the new england genetics collaborative

DRAFT
December 4, 2008

gemss gemssforschools.org

genetics education materials for school success

WEBSITE OVERVIEW

Genetics education materials for school success (GEMSS) is a new website for parents, educators, nurses, therapists, counselors, and others featuring information that schools can use to support students with genetic conditions. The site (www.gemssforschools.org) houses a library of useful tools and tips for educational supports, dietary needs, field trips, sports, and much more.

GEMSS IN SCHOOLS

About one in 20 children has a genetic condition, even without a prior family history. Teachers and parents can use GEMSS to better understand the needs of students who have genetic conditions. The aim of GEMSS is to make school a successful experience for all students who have genetic conditions.

"The GEMSS site is a wealth of information and a valuable asset to teachers and parents in helping them to develop comprehensive educational programs for children who have genetic disorders."

– Laurie Lambert, NH educator, inclusion facilitator

CONDITIONS

Visit the website to find information on supporting students with:

- 22q deletion velocardiofacial
- Angelman syndrome
- Down syndrome
- Fragile X
- MCAD and VLCAD
- PKU
- Prader-Willi syndrome
- Sickle Cell Disease
- Neurofibromatosis 1
- Williams syndrome
- Undiagnosed / other
- More to come!

SUPPORTED BY

GEMSS was developed by the Education & Outreach work group within the New England Genetics Collaborative (www.negenetics.org). The NEGC is funded by grant no. H46MC24093 with the Health Resources and Services Administration/Maternal and Child Health Bureau/Genetic Services Branch.

Available in alternative formats upon request.

Karen Smith | NEGC Project Coordinator
Institute on Disability | University of New Hampshire
karen.smith@unh.edu | 603.862.3454 | relay 711

GEMSS is a project of the

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www.gemssforschools.org

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gemss genetics education materials for school success

GEMSS - Genetics Education Materials for School Success: *Prader-Willi Syndrome*

www.gemssforschools.org

The AIM of GEMSS

To assure all children with genetic health conditions:

- Succeed in school-life
- Are members of neighborhood schools across the country

25+ Conditions in GEMSS

including...

Angelman	Prader-Willi
Cornelia de Lange	Rhett
Cystic Fibrosis	Sickle Cell
Down Syndrome	Turner
Fragile X	Urea Cycle
Fetal Alcohol	MCAD & VLCAD
Klinefelter	Williams
Marfan	22q Deletion
Neuromuscular	More!

GEMSS is for Teachers, Parents, School Nurses, and Genetic Counselors

Teachers and parents:

- Use GEMSS to better understand the needs of students who have genetic conditions
- Explore GEMSS to see if there are strategies you can implement
- Share the link with other families and groups

Nurses/Genetic Counselors:

- Refer families and educators to this site
- Provide feedback on new conditions and suggestions to the GEMSS team
- Provide printable handouts and flyers in your office waiting areas for families

To find the GEMSS website, go to
www.gemssforschools.org
Now available on your mobile device!

GEMSS is a web based resource for families and school personnel that offers:

- Genetic conditions explained in plain language
- Helpful strategies for field trips, diet, communication, instruction, transition, and more
- General tips and strategies for other conditions and undiagnosed

Parent Ambassadors are invited to help us spread the word!

GEMSS was created by geneticists, genetic counselors, teachers and parents as part of the New England Genetics Collaborative.

Institute on Disability
University of New Hampshire

www.negenetics.org

This project is supported by the Eunice Kennedy Shriver National Institute of Child Health and Human Development of the National Institutes of Health under Award Number U54HD090216. The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

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NEW ENGLAND CHILDREN WITH GENETIC DISORDERS & HEALTH CARE REFORM
Information and Recommendations for State Policymakers
negc THE NEW ENGLAND GENETICS COLLABORATIVE
This document is available in alternative formats upon request.
May 2014

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GPCI

GENETICS IN PRIMARY CARE INSTITUTE

geneticsinprimarycare.org

A cooperative agreement between the American Academy of Pediatrics and the Health Resources and Services Administration, Maternal and Child Health Bureau

Natalie Mikat-Stevens, MPH

Manager, Genetics in Primary Care Institute

New England Genetics Collaborative Meeting

April 11, 2014

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Implementing a Whole Genome Sequencing Clinical Research Study

Janet L. Williams, M.S., LGC
Marc S. Williams, M.D.

Research
Geisinger Health System

NEGC April 11, 2014

GEISINGER
REDEFINING BOUNDARIES

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A Regional Approach to Critical Congenital Heart Disease Newborn Screening Implementation

Monica McClain, MS, PhD
Research Associate Professor
Institute on Disability
University of New Hampshire

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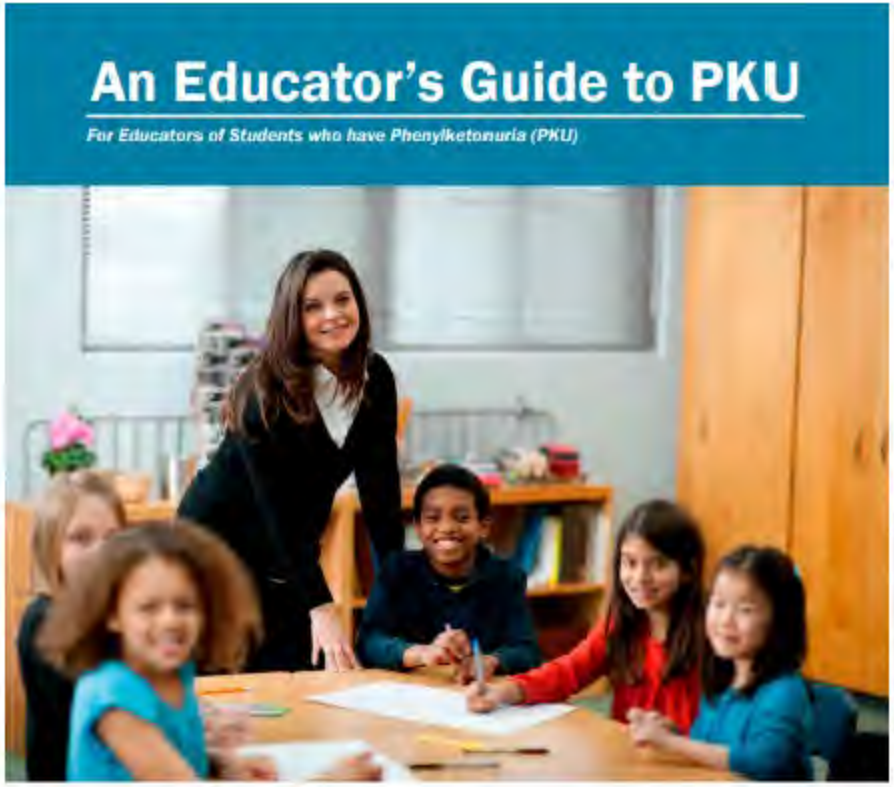
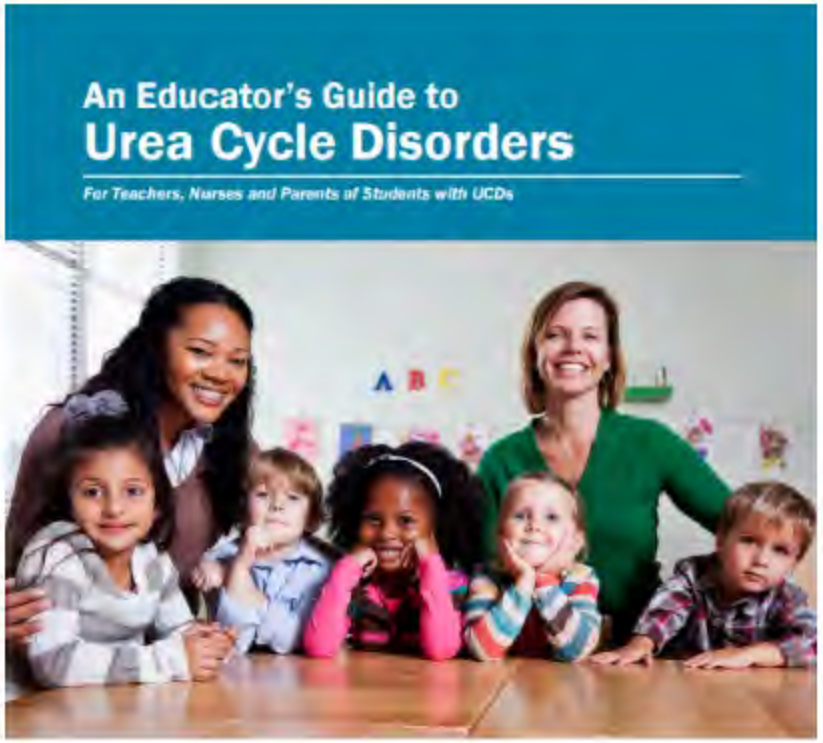


Boston Children's Hospital
Until every child is well



Boston Children's Hospital
Until every child is well

NUCDF

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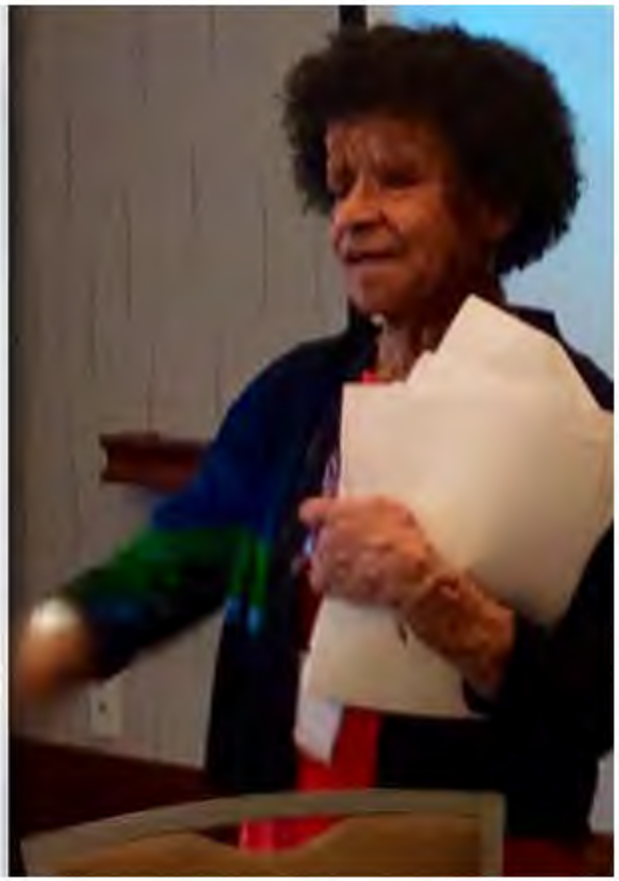
2014-2015


Absolute
Compassion is the
Only Thing that
Works



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Evaluation
Meeting

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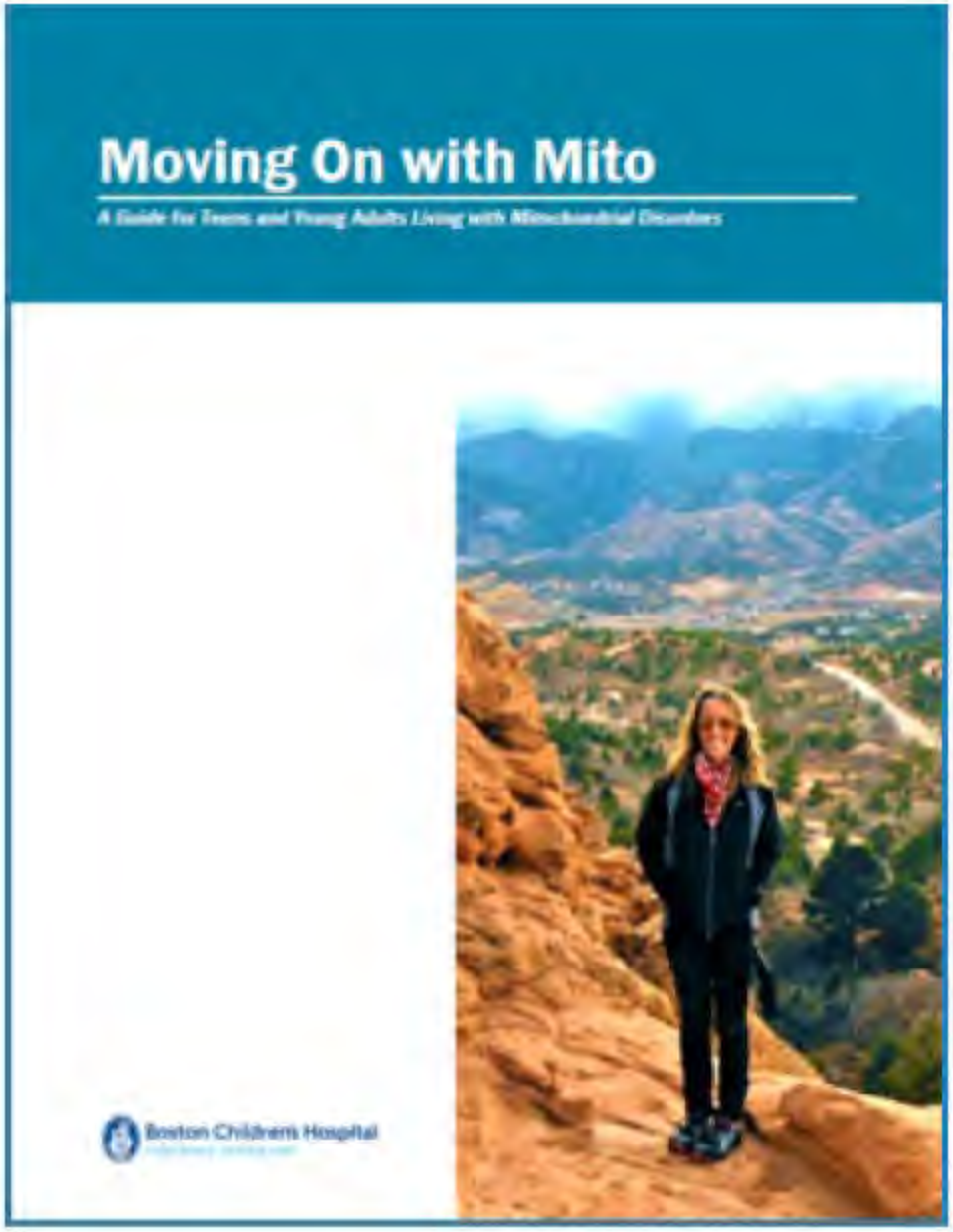

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New England Consortium of Metabolic Programs

Connecting professionals to provide the best patient care

Home | About Us | News | Membership | Annual Meeting | Upcoming Events | Contact | A To Z Index

Mitochondrial Disorders

Medical information you need to know as an adult with mitochondrial disorders

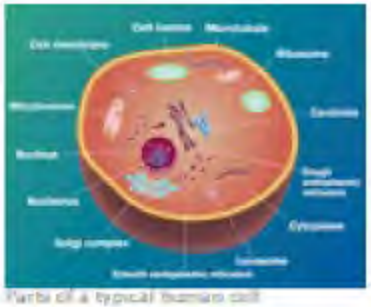
Print complete Transition Toolkit

This overview provides an introduction to mitochondrial disorders, their symptoms, and treatment. You can show it to friends, teachers, school nurses, coaches, new doctors, family members, and anyone else who you feel needs to understand your condition.

What are Mitochondrial Disorders?

In our bodies, mitochondria are the parts of cells that make energy from food and oxygen. With a mitochondrial disorder, something goes wrong with that energy-making process. When a cell is unable to produce enough energy, it may lose some of its ability to function. When enough cells in a certain part of the body are weakened that body part may not work correctly.

Mitochondria are found in almost all the cells in our bodies. In different people who have a mitochondrial disorder, different sets of cells, different body organs, or different body parts may be affected. Even people from the same family, with the same disorder, can have different health problems.



Parts of a typical human cell

Depending on which cells are affected, major symptoms might be:

- Muscle weakness or "heaviness"
- Vision or hearing problems
- Liver or kidney disease
- Diabetes
- Gastrointestinal problems
- Brain problems such as seizures

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School can be a great experience for EVERYONE!

About one in 20 children has a genetic condition.

Visit GEMSS:

Genetics Education Materials for School Success

gemssforschools.org

Product of the New England Genetics Collaborative and HRSA

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School can be a GREAT experience for EVERYONE!

About 1 in 20 children have a genetic condition that affects learning. The GEMSS website can help children with genetic conditions have more success in school.

Genetics Education Materials for School Success

gemssforschools.org

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GENETIC ALLIANCE

Schools camps & RESIDENCES

finding the GEMSS in your school

BY ANN DONOGHUE DILLON, M.ED., OTR/L • PHOTOS COURTESY THE GEMSS WEBSITE

If you are like me, I always had my eyes and ears open to learn about any information to help my daughter receive a good education! Having a genetic condition that was both rare and new for her school, she made all of us on the team try our best and then hope! I wish GEMSS was available when she was starting out in school! It would have been a great source of information that we could have used as a foundation, and then branched out as needed!

– Ann Donoghue Dillon

Are you the parent of a child who has a genetic condition such as Down syndrome, Fragile X, or Marfan syndrome? Have you searched for a base of knowledge that is comprehensive and reliable? Do you spend energy wondering HOW your child should be included in typical school programs, not questioning IF he/she should be included? You may be surprised to know that there is a new website receiving national and international attention! It is called GEMSS – Genetics Education materials for School Success www.gemssforschools.org

BACKGROUND

Launched in 2012, GEMSS now numbers over 20 conditions on its site. GEMSS has relied on feedback from parents, teachers, and viewers to help shape it. About seven conditions per year are being added and the site has expanded to include stories of children and adults who have many of the conditions.

Originally, a grant through the New England Genetics Collaborative encouraged workgroups to form within the Collaborative. The Education & Outreach Work Group began to dream about using the WEB to educate parents and teachers about the possibilities, cautions, and supports that are necessary to make a child's education more successful! Knowing that education can help alleviate the fears that can block acceptance and inclusion, they aimed to strengthen and reinforce that belief that education for ALL children, including those who happen to have a genetic condition, can occur in the typical classroom alongside their peers if they have the right supports.

gemss genetics education materials for school success

GEMSS was developed by the New England Regional Genetics Collaborative, one of seven HRSA-funded regional genetics collaboratives. Genetic Alliance is a nonprofit health advocacy organization that works closely with these regional collaboratives on engaging families and individuals and improving access to genetic services. For more information on Genetic Alliance and available resources for individuals and families around genetics and health, visit www.geneticalliance.org.

The content for each condition is created by a genetics counselor, and then travels to a parent reviewer, a geneticist,

GENETIC ALLIANCE

The world's leading nonprofit health advocacy organization committed to transforming health through genetics and promoting an environment of openness centered on the health of individuals, families, and communities.

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Health Policy and Advocacy Workshop

Gabrielle Orbaek White
State Advocacy Manager, Community Catalyst Alliance for Children's Health

Healthcare Access and Financing Workgroup Meeting
Thursday, April 9, 2015

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Face Forward Conference Report

July 12 – July 14, 2013

A Program of Next Step

Next Step's Face2Face Conference Report

July 9 – July 12, 2015

Next Step's Face2Face Program
www.nextstepnet.org

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"Welcome to Holland:" The Impact on Parents of a Diagnosis of CCHD

Joanna Fanos[1,2], Christopher Landon[3], Monica McClain[4]

[1]Department of Pediatrics, Geisel School of Medicine at Dartmouth

[2]Department of Psychology, San Jose State University

[3]Department of Pediatrics, Ventura County Medical Center

[4]Institute on Disability, University of New Hampshire

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ELSI Considerations and IRB Responses to Genomic Sequencing in the General Newborn Population

Caroline Weipert, MS, CGC and Meghan Towne, MS, CGC
Brigham and Women's Hospital and Boston Children's Hospital
New England Genetics Collaborative Annual Meeting
Portsmouth, NH – April 10, 2015

Boston Children's Hospital
Until every child is well

BWH BRIGHAM AND WOMEN'S HOSPITAL

HARVARD MEDICAL SCHOOL

BCM Baylor College of Medicine

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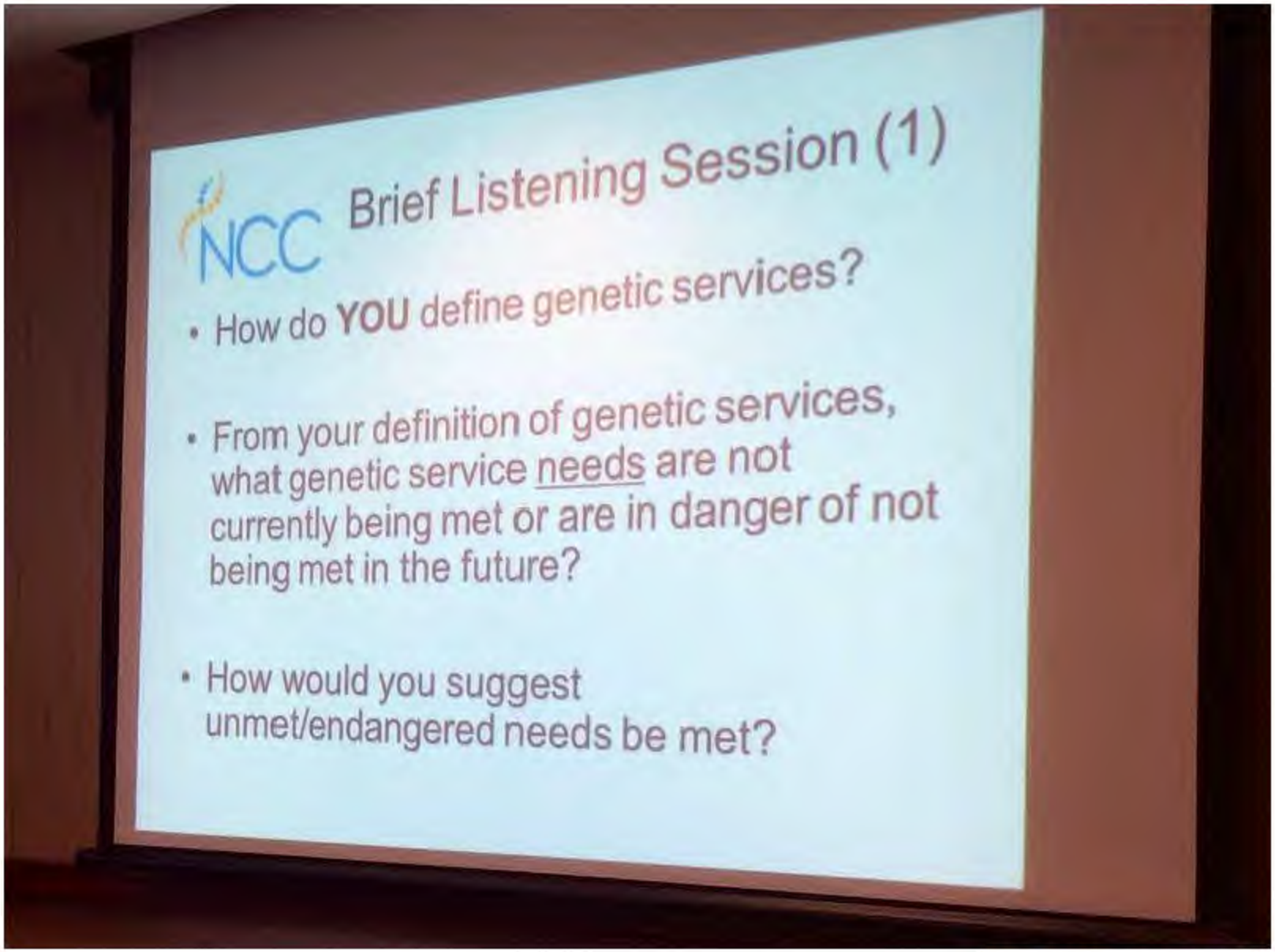
NCC
Brief Listening Session (1)
• How do YOU define genetic services?
• From your definition of genetic services, what genetic service needs are not currently being met or are in danger of not being met in the future?
• How would you suggest unmet/endangered needs be met?

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Point of Care CCHD Screening: Lessons from EHDI trenches

TERESE FINITZO, OZ SYSTEMS
ELIZABETH BRADSHAW, CHILDREN'S NATIONAL MEDICAL CENTER

JUNE 7, 2012

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Preliminary Findings of a Regional Approach to Critical Congenital Heart Disease Newborn Screening Implementation

Monica R. McClain MS, PhD *Institute on Disability, Health Management and Policy, University of New Hampshire*
Adelaide Murray *Health Management and Policy, University of New Hampshire*

Introduction

Pulse oximetry as a screening test to detect critical congenital heart disease (CCHD), has been recommended for universal newborn screening. This project examines a regional approach for five New England states to support the development, dissemination and validation of screening protocols and newborn screening infrastructure needs for CCHD. This study presents results through two years of this three year project.

Methods

A coordinating council comprising representatives from public health, pediatrics, pediatric cardiology, health educators, March of Dimes, family advocates, perinatal nurses and screening was formed to guide and evaluate this project. Seven birthing facilities have provided CCHD newborn screening data. An education work group was formed to develop an educational brochure.

Figure 1. Front of educational Brochure.

Results

States are in various stages of implementing CCHD newborn screening, and differences in public health authority to oversee programs exist. A Tier 2 educational brochure (Figure 1) has been developed for parents of a baby who receives a positive screen. To date, 32,747 babies have been screened (Figure 2); there have been 16 with positive screens (7.3 per 10,000) and 2 have been diagnosed with CCHD (1.2 per 10,000).

Site	# Screened	# Positive	Positive Rate (per 10,000)	# of CCHD cases	Diagnosis
Site A	5,001	6	12	1	TAPVR
Site B	1,314	2	15.2	1	TAPVR
Site C	1,965	1	5.1	0	
Site D	1,905	0	0	0	
Site E	1,481	2	13.5	0	
Site F	14,586	5	3.4	0	
Site G	6,495	0	0	0	
Total	**32,747**	**16**	**7.3 (4.5 – 11.7)**	**2**	

Figure 2. Data are from seven birthing facilities through 7/31/14. (Data that appears in red are from incomplete data sets)

Conclusions

Differences among states in the way disorders are added to their screening panel, and the wording of legislation has impeded public oversight of CCHD newborn screening programs. A regional approach to implementing CCHD screening allows the sharing of resources and expertise across states. Initial analyses show that the false positive rate is acceptable and the number of CCHD cases identified by newborn screening is slightly lower than expected.

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Their


Implications of the Critical Congenital
fect Screening Program
Emily Reddy, OTR
ELSI Results
ELSI Questions
Leadership
Through this Leadership in Action placement, I learned that it is vital that we assess the impacts of both the good and potentially harmful effects that programs have on people. Through our actions as leaders, it is crucial that we remain ethical, and do no harm to the people affected by our programs.
Through this project, I gained a deeper understanding of program evaluation. I learned the importance of communication when collaborating on portions of a larger project.
MAINE


Placement with GEMSS
nyon
n on the Autism condition web
webpage regarding it's
s/supports
website purpose thr
found at gemssfo
S office, I had never ha
ons. Furthermore, I ha

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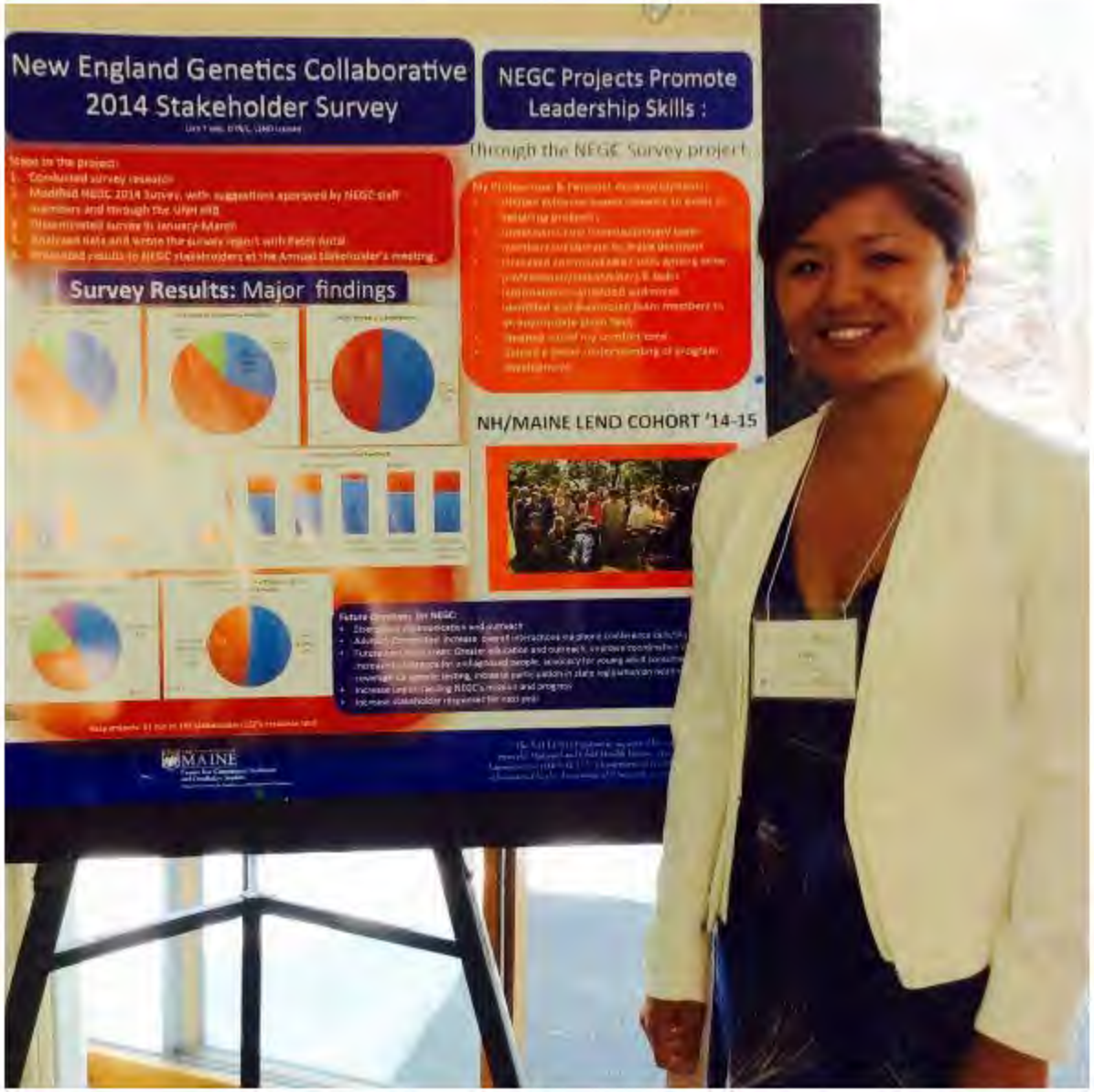

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Children with Genetic Disorders & Health Care Reform

The Health Care Access and Financing (HCAF) Workgroup

The Health Care Access and Financing (HCAF) Workgroup of the New England Genetics Collaborative (NEGC) was created in response to a Stakeholder Survey Report that identified "addressing financial barriers to care" as a high priority issue. The workgroup is made up of family-advocate leaders and a select number of professional partners.

The Policy Brief

The regional policy brief was created to inform stakeholders of the opportunities for improving health care access and financing for children with genetic disorders under the Affordable Care Act (ACA). It provides:

- A description of children with genetic disorders
- An overview of the current impacts of un- and underinsurance
- The results of a survey of New England families
- Legislative analysis related to health reform
- Recommendations for additional state policy options
- Additional resources in the appendix

"Children with genetic disorders experience gaps in insurance coverage and benefits that put their health and well-being in jeopardy and their families at risk for overwhelming medical debt."

Dissemination

The brief was distributed to over 1,600 New England state and federal policymakers, as well as the NEGC's partners. A webinar was hosted on 09/02/14 featuring highlights from the brief; the archive of which can be found on the NEGC website.

2015-2016

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"I HAVE A COMPLAINT"

What the Maine Bureau of Insurance
can do to assist you.

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IMPROVING THE NEWBORN SCREENING SYSTEM IN THE GENOMIC ERA

Aaron Goldenberg, PhD, MPH
Case Western Reserve University

Beth Tarini, MD, MS
University of Michigan

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2014 Updates from the National Coordinating Center for the Regional Genetic Service Collaboratives (NCC)

Providing Resources for Bridging Genetics, Primary Care, and Public Health, and for Bringing Genetics to Local Communities

The NCC is funded by U22MC24100, awarded as a cooperative agreement between the Maternal and Child Health Bureau/Health Resources and Services Administration, Genetic Services Branch, and the American College of Medical Genetics.

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QC for Kids

nhpip

VCHIP

Webinar Series Announcement:

"It's All About Teamwork: Incorporating Genetics and Family History into the Work of the Patient Centered Medical Home (PCMH)."

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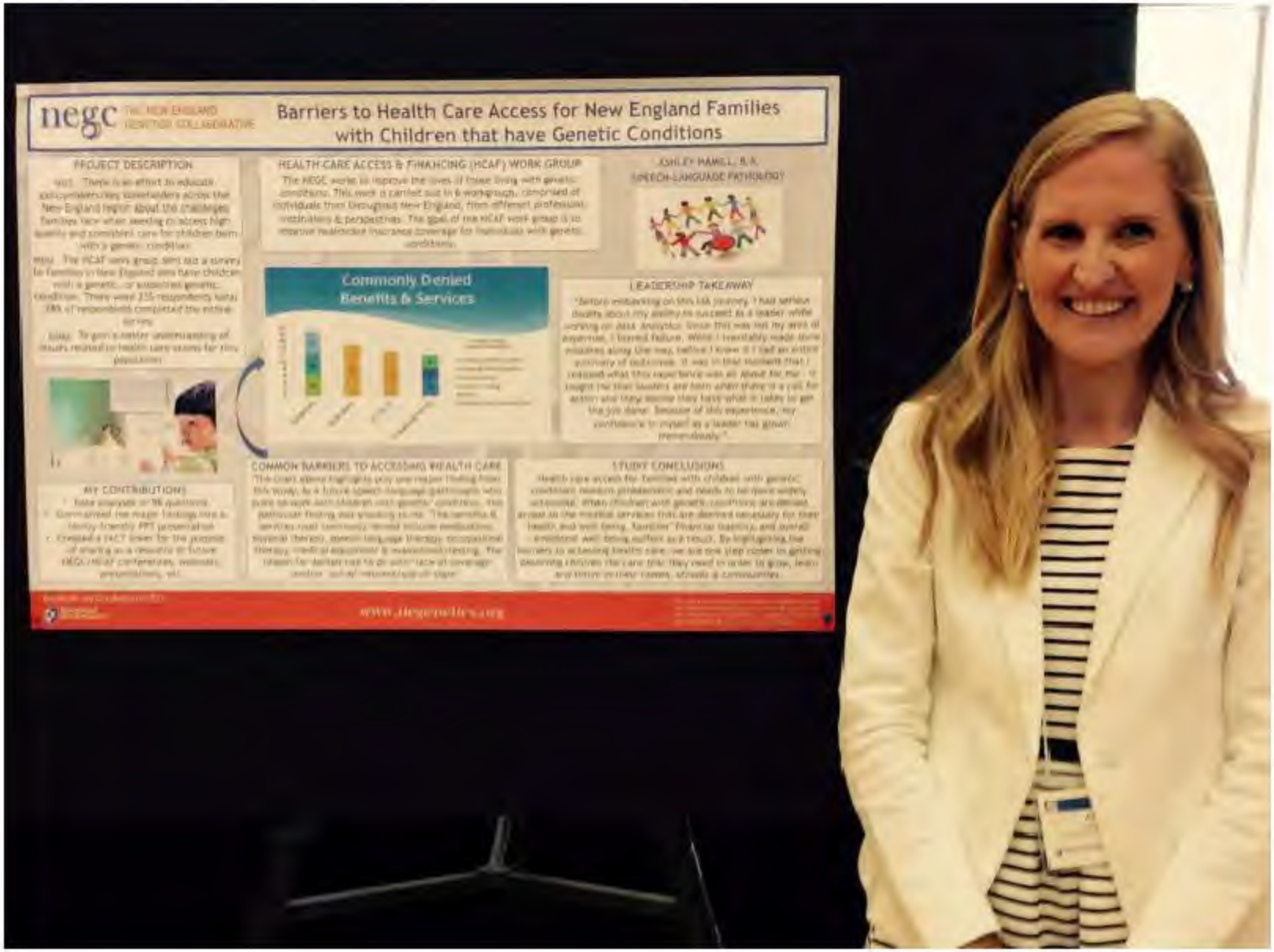
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Barriers to Health Care Access for New England Families with Children that have Genetic Conditions
PROJECT DESCRIPTION
HEALTH CARE ACCESS & FINANCING (HCAF) WORK GROUP
The NEGC works to improve the lives of those living with genetic conditions. This work is carried out in 6 workgroups, comprised of individuals from throughout New England, from different professional institutions & perspectives. The goal of the HCAF work group is to improve reasonable insurance coverage for individuals with genetic conditions.
ASHLEY HAMEL, M.A.
SPEECH-LANGUAGE PATHOLOGY
Commonly Denied Benefits & Services
LEADERSHIP TAKEAWAY
"Before embarking on this LEA journey, I had serious doubts about my ability to succeed as a leader while working on data analytics. Since this was not my area of expertise, I feared failure. While I inevitably made some missteps along the way, before I knew it I had an entire summary of outcomes. It was in that moment that I realized what this experience was all about for me - it taught me that leaders are born when there is a call for action and they decide they have what it takes to get the job done. Because of this experience, my confidence in myself as a leader has grown tremendously."
MY CONTRIBUTIONS
COMMON BARRIERS TO ACCESSING HEALTH CARE
STUDY CONCLUSIONS
www.negcwikis.org
THE NEW ENGLAND GENETICS COLLABORATIVE

negc
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Institute on Disability
RESEARCH

April 2016

Promoting and Improving the Health and Well-Being of People with Inherited Conditions

Written by: Ashley Hamill

The mission of the New England Genetics Collaborative (NEGC) is to promote and improve health and social well-being of those with inherited conditions through collaborations among public health professionals, private health professionals, educators, consumers and advocates throughout New England.

The NEGC is housed at the Institute on Disability, at the University of New Hampshire. To access the NEGC's website, please visit www.negenetics.org. One of the work groups of the NEGC is the Health Care Access and Financing (HCAF) work group, with an overarching goal of improving healthcare insurance coverage for individuals with genetic conditions.

Surveys of New England Families of Children with Genetic Conditions

In 2012, the HCAF work group designed an online survey of families of children with genetic disorders living in the six New England states. The questions in the survey were intended to identify health insurance coverage and benefits gaps for children with genetic disorders. Key findings suggested that particularly challenging areas of underinsurance included outpatient services, care for emotional, behavioral or substance abuse issues, prescription drug coverage, rehabilitative and habilitative therapies, medical devices, pediatric services like developmental screenings, and prescribed medical foods. Furthermore, families reported cost as a major problem—high deductibles, co-pays and co-insurance were noted in almost every category. Overall, these survey results emphasized the critical need for policymakers to take action to reduce underinsurance for children with genetic disorders. A link to the 2014 policy brief highlighting specific policy implications and recommendations can be found here.

There is an effort to educate policymakers and key stakeholders across the New England region about the challenges families face when seeking to access high quality and consistent care for children born with a genetic condition. A second survey was sent out to families in New England who have children with a genetic, or suspected genetic, condition. The goal of this survey was to gain a better understanding of issues related to health care access and coverage/reimbursement for this population.

Survey results were analyzed both quantitatively and qualitatively and major findings are summarized below.

Methodology & Demographics

Survey respondents were parents or guardians (ages 18+) of children living throughout New England who were under the age of 21 and who had been diagnosed with a genetic condition, or who had been told by a health care professional that there may be a genetic link to their child's condition.

All questions were voluntary and respondents were able to enter into a raffle for a $50 Target gift card. The survey was open from September 8, 2015 until October 31, 2015.

There were a total of 255 responses, from MA, ME, NH, CT, RI, VT, with the majority of responses being from NH; 72 completed the survey in its entirety. The most common average age of children was between 8 – 11 years old. The seven most common genetic diagnoses represented included Down Syndrome,


9

gemss genetics education materials for school success

from the New England Genetics Collaborative (NEGC)

Quick Links

Forward to a friend

View in browser

Visit GEMSS website

Visit NEGC website

Follow NEGC on Twitter

Contact us

The NEGC is one of seven regional organizations across the United States dedicated to narrowing the gap between what is, and what can be, for individuals with genetic disorders. The NEGC coordinates collaboration between representatives of public health, metabolic and genetic clinics, medical homes, academia and parent groups to support innovation in genetics and improve access to genetic services.

January 2016

Dear instructor (university, community college or other),

Whether in general or special education, every school teacher will have some students with complex needs. Sometimes those needs will be related to a genetic condition, as about 1 in 20 children have a genetic condition.

We want to put a resource in *your* hands to help future teachers meet these needs, and we want to make it easy for *you*.

The Learning Module for GEMSS (Genetics Education Materials for School Success) builds on the GEMSS website and is a free public resource. The website itself has:

- Vetted information and resources for 30+ genetic conditions
- Practical tips & strategies for learning

The ***Learning Module*** has suggested activities - choose one or all:

- Pre-work for students
- In or out-of-class assignments (works well online)
- Slide presentation for whole group activity
- Independent or small group activity
- Discussion questions to encourage critical thinking

We encourage you to incorporate the Learning Module into your class, or share this with others who might.

Teachers can be a great support for all students - GEMSS can help!

Best regards,

The New England Genetics Collaborative (NEGC)
Education & Outreach Work Group

The New England Genetics Collaborative Tel: (603) 862-4320 | Fax: (603) 862-0555

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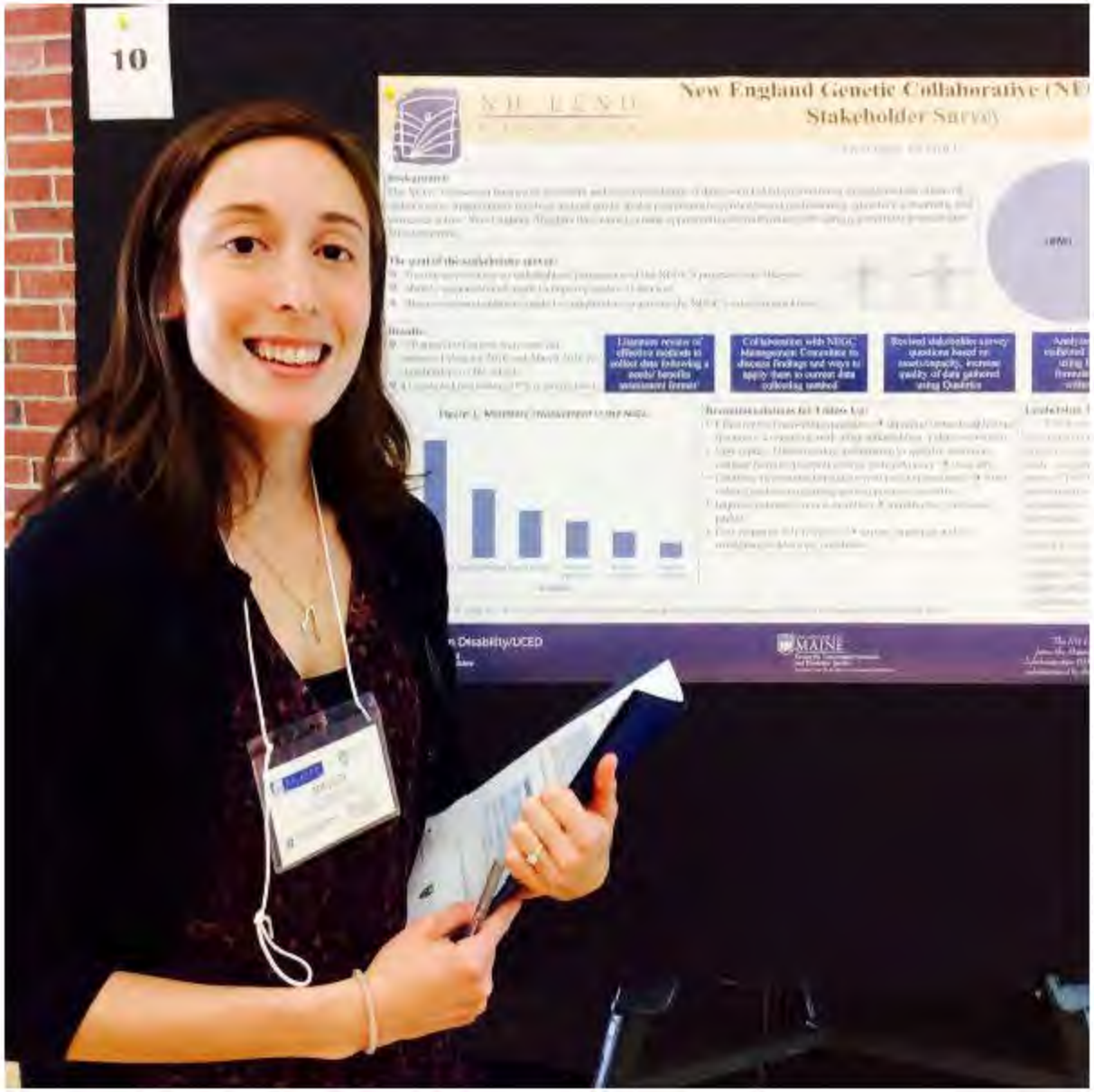
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