

Follow-up and Treatment Workgroup for ACHDNC

**Stephen McDonough, MD
Pediatrician**

Bismarck, North Dakota February 9, 2017

New members to workgroup

- December 14, 2016 Dr. Kus and myself participated in a conference call with GSB to review 11 potential candidates for 4 open positions on LTFU workgroup
- 4 recommendations with 2 alternates were sent to chair of ACHDNC and the following excellent professionals were chosen
 - Dr. Nancy Doan Leslie
 - Dr. Joseph Schneider
 - Dr. Margie Ream
 - Dr. Debra Freedenberg

Medical Foods sub-workgroup

- Chair- Susan Berry MD, Director, Division of Genetics and Metabolism, University of Minnesota Department of Pediatrics
- Co-chairs
 - Christine S. Brown, MS, Executive Director, National PKU Alliance
 - Kathryn Camp, MS, RD, Retiree
 - Carol Greene, MD Professor of Pediatrics, University of Maryland Medical Center, Clinical Medical Genetics

Medical Foods sub-workgroup

- Members
 - Kathryn L. Hassell, MD, University of Colorado School of Medicine, Professor of Medicine
 - Christopher A. Kus, MD, MPH, Pediatric Director, Associate Medical Director, Division of Family Health, New York State Department of Health
 - Stephen McDonough, MD, pediatrician, Bismarck, North Dakota
 - Kamila B. Mistry, PhD, MPH, Senior Advisor, Child Health and Quality Improvement, Agency for Healthcare Research and Quality (AHRQ)
 - Jana Monaco, Advocacy Liaison, Organic Acidemia Association

Medical Foods sub-workgroup

- Members
 - Robert Ostrander MD, Valley View Family Practice Associates, Rushville, New York
 - Joseph Schneider, MD, Retiree
 - Rani Singh, Ph.D., Professor, Director, Division of Medical Genetics, Nutrition Section, Emory University School of Medicine
 - Janet Thomas MD, Pediatrics and Genetics, Colorado Children's Hospital

Medical Foods sub-workgroup

- HRSA Members
 - Joan A. Scott, M.S., C.G.C., Department of Health and Human Services, Health Resources and Services Administration, Maternal and Child Health Bureau, Division of Services for Children with Special Healthcare Needs, Acting Chief, DSCSHN.
 - Debi Sarkar, M.P.H., Health Resources and Services Administration, Maternal and Child Health Bureau, Chief, Genetic Services Branch, Designated Federal Official
 - Jill F. Shuger, ScM, Health Resources and Services Administration, Maternal and Child Health Bureau, Genetic Services Branch, Project Officer

Medical Foods sub-workgroup

- Phone call meetings
 - June 9, 2016
 - July 14, 2016
 - August 11, 2016
 - October 13, 2016
 - December 12, 2016 co-chairs
 - January 12, 2017 full workgroup

Quality Measures sub-workgroup

- Chair- Alan E. Zuckerman , M.D., Assistant Professor, Community and Family Medicine; Assistant Professor, Department of Pediatrics, Georgetown University Medical Center
- Co-chairs
 - Amy Brower, PhD, National Coordinating Center, Newborn Screening Translational Research Network, American College of Medical Genetics
 - Jeffrey P. Brosco MD PhD Professor of Clinical Pediatrics University of Miami School of Medicine Department of Pediatrics
 - Kathryn L. Hassell, M.D. University of Colorado School of Medicine, Professor of Medicine
 - Jana Monaco, Advocacy Liaison, Organic Acidemia Association

Quality Measures sub-workgroup

- Members
 - John Eichwald, M.A., branch chief Child Development and Disability (CDD) Division of Human Development and Disability (DHDD) National Center on Birth Defects and Developmental Disabilities (NCBDDD) at the Centers for Disease Control and Prevention (CDC)
 - Terese Finitzo, PhD, clinical pediatric audiologist and CEO of OZ Systems
 - David Flannery, MD, FACMG, Medical Director, American College of Medical Genetics and Genomics
 - Debra Freedenberg, MD, PhD, Medical Director, Newborn Screening & Genetics, family & Community Health Service Division, Department of State Health Services, Texas.
 - Rebecca Goodwin, JD Project Manager and Special Assistant to the Director of the Lister Hill National Center for Biomedical Communications

Quality Measures sub-workgroup

- Members

- Celia Kaye, MD, PhD, Professor Emerita, Pediatrics-CHA/PA Program Consultant, Education Office, UCD School of Medicine Senior Genetics Advisor
- Christopher Kus, M.D., M.P.H., Associate Medical Director, Division of Family Health, New York State Department of Health
- Sylvia Mann, Supervisor, Genomics Section, State Genetics Coordinator, Hawaii Department of Health
- Stephen McDonough, MD, pediatrician, Bismarck, North Dakota, Retiree
- Kamila B. Mistry, PhD, MPH, Senior Advisor, Child Health and Quality Improvement, Agency for Healthcare Research and Quality (AHRQ)
- Robert Ostrander MD, Valley View Family Practice Associates, Rushville, New York

Quality Measures sub-workgroup

- Members

- Marci Sontag, PhD, Associate Professor, MPH, Epidemiology, Colorado School of Public Health
- Annamarie Saarinen, Co-Founder, CEO, Newborn Foundation
- Joseph Schneider, MD, Retiree
- Stanley Sciortino, PhD, California Department of Public Health
- Janet Thomas MD, Pediatrics and Genetics, Colorado Children's Hospital

Quality Measures sub-workgroup

- HRSA Members

- Joan A. Scott, M.S., C.G.C., Department of Health and Human Services, Health Resources and Services Administration, Maternal and Child Health Bureau, Division of Services for Children with Special Healthcare Needs, Acting Chief, DSCSHN
- Debi Sarkar, M.P.H., Health Resources and Services Administration, Maternal and Child Health Bureau, Chief, Genetic Services Branch, Designated Federal Official
- Jill F. Shuger, ScM, Department of Health and Human Services, Health Resources and Services Administration, Maternal and Child Health Bureau, Division of Services for Children with Special Healthcare Needs, Project Officer
- Catharine Riley, PhD, MPH, Department of Health and Human Services, Health Resources and Services Administration, Maternal and Child Health Bureau, Division of Services for Children with Special Healthcare Needs, Newborn Screening Lead

Quality Measures sub-workgroup

- Phone call meetings
 - July 21, 2016
 - August 18, 2016
 - September 15, 2016
 - October 20, 2016
 - November 17, 2016
 - December 15, 2016
 - January 19, 2017

Quality Measures Progress Report

**February 9, 2017
Alan E Zuckerman MD FAAP**

What are Quality Measures?

- Quality measures are standardized quantitative assessment tools that are a first step and an essential part of quality improvement activities or designing proactive clinical decision tools.
- They are typically ratios such as the percentage of children with sickle cell disease prescribed penicillin.
- Each expresses a definition of quality as a goal for care delivery.
- Some measures are a subset of more comprehensive research databases that are used for new knowledge discovery and that may require informed consent.
- Quality assessment/improvement is part of routine care and can be embedded in electronic health records (EHR) to eliminate the need for separate data entry or manual chart review.
- New HIT standards guide electronic definition, measurement, and reporting of quality measures.
- The interest and need to use and report quality measures creates an opportunity to apply them to LFTU of NBS.

Our Definition of Long Term Follow-up is Linked to Quality Assessment

- Fundamentally, long-term follow-up comprises the assurance and provision of **quality chronic disease management, condition-specific treatment, and age-appropriate preventive care** throughout the lifespan of individuals identified with a condition included in newborn screening. Integral to assuring appropriate long-term follow-up are activities related to improving care delivery, including engagement of affected individuals and their families as effective partners in care management, **continuous quality improvement through the medical home**, research into pathophysiology and treatment options, and active surveillance and **evaluation of data related to care and outcomes**.

Three Types of Approaches to Quality Measurement for LTFU of NBS

- An important finding of our work to date is that there are three types of approaches to quality for LTFU of NBS that use different quality goals and methods:
 - The most familiar are the **disease-specific measures** of process and physiologic outcomes.
 - The second are **public health services** after children are identified through screening and these may be provided outside of the NBS program to connect children to care they require.
 - The third are **patient/child-specific measures** of well being and access to medical homes and services that are best measured directly from consumers.
- All three overlap in the populations they are applied to and the settings (provider organizations, health departments, consumers) where they are measured.

Case Studies Illustrate the Value and Feasibility of Using Quality Measures

- Case studies of the use of quality measures for follow-up of newborn screening reveal the value of their use and barriers we seek to overcome:
 - EHDI measures we certified by NQF after considerable work.
 - Sickle Cell measures have revealed failures to start recommended preventive care.
 - Cystic Fibrosis care has evolved by comparing best practices in centers of excellence.
 - An EHR embedded check list for MCADD has helped find gaps in care and alert providers to emergency needs.
 - The National Survey of Children with Special Healthcare Needs reveals gaps in access and use of medical homes.

Activities and Timeline

- The sub-workgroup has held monthly calls since July 2016.
- We have a clear work program and have been developing and reviewing draft materials.
- We have three co-chairs (Amy Brower, Kathryn Hassell, and Jana Monaco) representing informatics, clinicians, and consumers to assure including all stakeholder perspectives.
- We have public health participation from several states.
- Jeff Brosco, ACHDNC member and bioethicist, has provided important guidance and insight to the group.
- We anticipate a final report by August 2017 that we would like to share with the ADHDNC to enlist their help in promoting quality measures as an important strategy for assuring long term follow-up of newborn screening.