Advisory Committee on Heritable Disorders in Newborns and Children

Meeting Summary
September 24, 2019

The Advisory Committee on Heritable Disorders in Newborns and Children (Committee) meeting was convened on September 24, 2019, and adjourned on September 24. In accordance with the provisions of Public Law 92-463, the meeting was open for public comment.

Committee Members

Mei Baker, M.D.
Professor of Pediatrics
University of Wisconsin School of Medicine and Public Health
Co-Director, Newborn Screening Laboratory
Wisconsin State Laboratory of Hygiene

Susan A. Berry, M.D.
Professor and Director
Division of Genetics and Metabolism
Departments of Pediatrics and Genetics, Cell Biology & Development
University of Minnesota

Jeffrey P. Brosco, M.D., Ph.D.
Professor of Clinical Pediatrics
University of Miami School of Medicine
Department of Pediatrics
Deputy Secretary, Children's Medical Services
Florida State Department of Health

Kyle Brothers, M.D., Ph.D.
Endowed Chair of Pediatric Clinical and Translational Research
Associate Professor of Pediatrics
University of Louisville School of Medicine

Jane M. DeLuca, Ph.D., R.N.
Associate Professor
Clemson University School of Nursing

Cynthia M. Powell, M.D., FACMG, FAAP (Chairperson)
Professor of Pediatrics and Genetics
Director, Medical Genetics Residency Program
Pediatric Genetics and Metabolism
The University of North Carolina at Chapel Hill

Annamarie Saarinen
Co-founder, CEO
Newborn Foundation

Scott M. Shone, Ph.D., HCLD(ABB)
Director
North Carolina State Laboratory of Public Health

Beth Tarini, M.D., M.S., FAAP
Associate Director, Center for Translational Research
Children's National Health System

Ex-Officio Members

Agency for Healthcare Research & Quality
Kamila B. Mistry, Ph.D., M.P.H.
Senior Advisor
Child Health and Quality Improvement

Centers for Disease Control & Prevention
Carla Cuthbert, Ph.D.
Chief, Newborn Screening and Molecular Biology Branch
Division of Laboratory Sciences

Food and Drug Administration
Kellie B. Kelm, Ph.D.
Deputy Director
Division of Chemistry and Toxicology Devices
Office of In Vitro Diagnostics and Radiological Health

Health Resources & Services Administration
Michael Warren, M.D., M.P.H., FAAP
Associate Administrator,
Maternal and Child Health Bureau

National Institutes of Health
Diana W. Bianchi, M.D.
Director
Eunice Kennedy Shriver National Institute

Designated Federal Official

Catharine Riley, Ph.D., M.P.H.
Health Resources and Services Administration
Genetic Services Branch
Maternal and Child Health Bureau

Organizational Representatives

American Academy of Family Physicians
Robert Ostrander, M.D.
Valley View Family Practice

American Academy of Pediatrics
Debra Freedenberg, M.D., Ph.D.
Medical Director, Newborn Screening and Genetics, Community Health Improvement
Texas Department of State Health Services

American College of Medical Genetics & Genomics
Michael S. Watson, Ph.D., FACMG
Executive Director

American College of Obstetricians & Gynecologists
Steven J. Ralston, M.D., M.P.H.
Chair, OB/GYN
Pennsylvania Hospital

Association of Maternal & Child Health Programs
Jed L. Miller, M.D., M.P.H.
Director, Office for Genetics and People with Special Health Care Needs
Maryland Department of Health
Prevention & Health Promotion Administration

Association of Public Health Laboratories
Susan M. Tanksley, Ph.D.
Manager, Laboratory Operations Unit Texas Department of State Health Services

Association of State & Territorial Health Officials
Christopher Kus, M.D., M.P.H.
Associate Medical Director
Division of Family Health
New York State Department of Health

Association of Women's Health, Obstetric & Neonatal Nurses
Jacqueline Rychnovsky, Ph.D., R.N., CPNP, FAANP
Vice President, Research, Policy and Strategic Initiatives

Child Neurology Society
Jennifer M. Kwon, M.D., M.P.H, FAAN
Director, Pediatric Neuromuscular Program
American Family Children's Hospital
Professor of Child Neurology, University of Wisconsin School of Medicine & Public Health

Department of Defense
Jacob Hogue, M.D.
Lieutenant Colonel, Medical Corps, US Army
Chief, Genetics, Madigan Army Medical Center

Genetic Alliance
Natasha F. Bonhomme
Vice President of Strategic Development

March of Dimes
Siobhan Dolan, M.D., M.P.H.
Professor and Vice Chair for Research
Department of Obstetrics & Gynecology and Women's Health
Albert Einstein College of Medicine and Montefiore Medical Center

National Society of Genetic Counselors
Cate Walsh Vockley, M.S., LCGC
Senior Genetic Counselor
Division of Medical Genetics
UPMC Children's Hospital of Pittsburgh

Society for Inherited Metabolic Disorders
Georgianne Arnold, M.D.
Clinical Research Director, Division of Medical Genetics
UPMC Children's Hospital of Pittsburgh

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I. Administrative Business

Cynthia M. Powell, M.D., M.S., FACMG, FAAP
Committee Chair
Professor of Pediatrics and Genetics
Director, Medical Genetics Residency Program
Pediatric Genetics and Metabolism, The University of North Carolina at Chapel Hill

Catharine Riley, Ph.D., M.P.H.
Designated Federal Official
Health Resources and Services Administration (HRSA)

A. Welcome and Roll Call

Dr. Powell welcomed participants to the fourth meeting in 2019 of the Advisory Committee on Heritable Disorders in Newborns and Children.

Dr. Powell then conducted the roll call. The Committee members in attendance were:

- Dr. Kamila Mistry (Agency for Healthcare Research & Quality)
- Dr. Susan Berry
- Dr. Jeff Brosco
- Dr. Kyle Brothers (joined later in the meeting)
- Dr. Jane DeLuca
- Dr. Kellie Kelm (Food and Drug Administration)
- Ms. Joan Scott (Heath Resources & Services Administration)
- Dr. Cynthia Powell
- Dr. Scott Shone
- Dr. Catharine Riley (Designated Federal Official)

Organizational representatives in attendance were:

- American Academy of Family Physicians, Dr. Robert Ostrander
- American Academy of Pediatrics, Dr. Debra Freedenberg
- American College of Medical Genetics & Genomics, Dr. Michael Watson
- Association of Maternal & Child Health Programs, Dr. Jed Miller
- Association of Public Health Laboratories, Dr. Susan Tanksley
- Association of State & Territorial Health, Dr. Christopher Kus
- Association of Women's Health, Obstetric & Neonatal Nurses, Dr. Jacqueline Rychnovsky
- Department of Defense, Dr. Jacob Hogue
- Genetic Alliance, Ms. Natasha Bonhomme
- March of Dimes, Dr. Siobhan Dolan
- National Society of Genetic Counselors, Dr. Cate Walsh Vockley
- Society for Inherited Metabolic Disorders, Dr. Georgianne Arnold

B. Vote on August 2019 Meeting Minutes

The Committee members received a draft of the minutes of the August meeting to review prior to this meeting. As no revisions were to be heard, the Committee voted unanimously to approve the minutes.

C. Opening Remarks

Dr. Powell provided an update on the medical foods report that was previously accepted by the Committee. A reply was received on September 9th from HRSA's Acting Administrator on behalf of Health and Human Services thanking the Committee.

Joan Scott was introduced to speak and she commented that the legislative authority for the Committee expires on September 30th and at that time committee operations will halt. There is an option to establish a discretionary committee which is being considered. All information and any future dates will be on the Committee's website.

Dr. Riley introduced herself as the Designated Federal Official and provided the Committee with standard reminders. She stated that this Advisory Committee is governed by the Federal Advisory Committee Act and all Committee members are subject to the rules and regulations for special government employees. Also, all Committee members must recuse themselves from participation in all matters that affect the financial interest of any organization with which you serve as an officer, director, trustee, or general partner, unless employed by the organization, or received a waiver from HHS authorizing you to participate.

II. Interoperability for Newborn Screening: State Experiences

Amy Gaviglio, M.S., CGC
Follow-Up Supervisor/Genetic Counselor
Minnesota Department of Health Newborn Screening Program

Brendan Reilly
Program Specialist
Texas Department of State Health Services

Ms. Gaviglio provided a quick disclaimer, stating that some of the work she would present was completed during her tenure at the Minnesota Department of Health. She then asked why newborn screening programs should build more data. The answer was broken down into three phases: preanalytical, analytical, post-analytical.

It was then explained how connecting vital records, for instance matching specimens received with filed birth certificates, can provide a better understanding of who has or has not been screened and monitor the refusal rate. However, in order to implement this connection of data, the importance of state statutory requirements, limitations of out-of-hospital births or births attended by relatives, and the timing of the connection in order to be effective must all be understood.

Ms. Gaviglio stated that the Minnesota Newborn Screening Program utilized this connection beginning

in August 2016. The program's Office of Vital Records each day sends a file from the prior day through the Internal Exchange Hub, which a staff member manually imports into the LIMS system. Once imported, a query is run within the LIMS which looks to match the birth certificate information with the specimens received. The most accurate matches are found when using four criteria: infant date of birth, infant time of birth, mother's first and last name. If a match is found, the birth certificate number and any other associated information is automatically added to the patient's case within the LMS. If a match is not found, it is manually reviewed by a program staff member who can deselect or select other demographic criteria to determine if a match does exist.

By using this process, the Minnesota program is able to perform a match in approximately five days from date of birth or even sooner once the birth certificate is filed. It also generates information regarding screening refusals, in-state births that were transferred out, lost specimens needing a follow-up, and number of infants remaining unscreened.

Vital record connections can be used to obtain state-level denominators from birth facilities with EMRs called Newborn Admission Notification Information, or NANI, which connects to the birth facility's EMR using an HLT ADT feed. In Minnesota, four specific ADT messages are received from birth facilities: A01 for patient admission, A08 for an update of patient records, A03 for a patient discharge, and A31 for personal information updates. These are just two projects involving the connection of vital records in order to improve outcomes. Other connection possibilities are with birth defect registries to other programs such as WIC, clinical laboratories and subspecialist EMRS.

Dr. Gaviglio then concluded that interoperability can potentially save staff time and money as well as improve the newborn screening process and system. Unfortunately, time and money are needed up front to facilitate this.

Mr. Reilly was then introduced to talk about electronic test orders. It was explained that traditionally test orders are received on a demographic form with the blood spot specimen. This is all on a handwritten form which limits the number of fields collected as it needs to fit a certain size. This is then transcribed by data-entry operators by hand into a system leading to various transcription issues.

Comparatively, electronic orders are sent electronically and are more intricate. Orders can be sent to LIMS or as previously described using the NANI system which can send admission messages to web applications or even the LIMS. Integration of this can take the necessary information, place it in the proper format and send it to the public health laboratory.

Mr. Reilly elaborated that when dealing with electronic results, when transmitted the results are integrated into the system without the need of transcription. By using electronic orders, the Texas Newborn Screening Program has been able to increase efficiency by reducing the time of manually entering results by 130,000 hours and improving both data accuracy and data completeness. Yet, when dealing with electronic results there are also technological difficulties or issues. Also, implementing these efficiencies is not an easy effort. For example, in Texas, separate connections require separate interfaces and can cause deficiencies.

Newborn screening is different than a standard lab test order. The information that is needed is typically not available in the LIMS which causes a challenge of needing custom coding to get all that information

into the message going to the laboratory as the majority of LIS and EMR vendors do not have specific solutions just for newborn screening.

A. Discussion

The Committee discussion began with questions concerning the number of required links within the NANI program. It was stated that nothing different is required and that nothing additional was required for two-screen programs. As there are only about 15 to 20 data entry operators it was discussed how efficient the system can be. Further, system ownership came into questions and a discussion on how 90% of the ownership comes from the program which also involves implementation and system maintenance. However, it was suggested to have a project manager and supervisor during setup. Lastly, the Committee Chairperson questioned if or how the two or three large electronic health record providers were integrated. The Committee members then discussed the flexibility of the system but pointed out that approaching the EMR and LIMS vendors in order to advance them on the development of a newborn screening-specific solution is an alternate approach of integrating.

III. Public Comments

A. Rebecca Abbot, March of Dimes

Rebecca Abbott is the Deputy Director of Federal Affairs for Public Health at the March of Dimes. The coalition has focused efforts on reauthorization of the Newborn Screening Saves Lives Act. The legislation moved through the House quickly; however, progress in the Senate has been much slower. Sen. Maggie Hassan (D-N.H.) and Sen. Cory Gardner (R-Colo.) introduced legislation in July, and since then the coalition has been working with them and staff on the Senate Health Committee on refinements to the bill and language to address concerns from other lawmakers. The coalition continues to pursue all legislative options to reauthorize the Newborn Screening Saves Lives Act as soon as possible.

IV. RUSP Condition Nomination and Evidence Review Process: Public Health System Impact Assessment

Cynthia M. Powell, M.D., M.S., FACMG, FAAP
Committee Chair
Professor of Pediatrics and Genetics
Director, Medical Genetics Residency Program
Pediatric Genetics and Metabolism, The University of North Carolina at Chapel Hill

Dr. Powell quickly reviewed the four focus points of the Condition Nomination and Evidence Review and Decision-Making Processes: the nomination process, the systematic evidence-based review process, the decision matrix and decision-making process, and a possible review of current conditions on the RUSP.

Prior April and August discussions were reviewed, and then the topic of how the Committee assesses the impact of adding new conditions on the public health system was introduced.

The assessment of state newborn screening programs is intended to evaluate the entire integrated system needed for implementation of comprehensive newborn screening includes authority, laboratory testing, interpretation, reporting, tracking, and systems for assurance of diagnostic evaluations, and evaluation of outcome in order to inform the Committee on the feasibility of screening, state readiness and to describe the cost of implementing a new condition screening.

In order to assess the impact of adding new conditions on the public health system, there needs to be feasibility and readiness of the condition in the form of resource availability and valid and reliable tests. A review of the current approach of population modeling and surveys was then provided. Some of the feedback that was received was how surveys may not capture implementation difficulties nor can account for possible impacts on primary care physicians, specialists and other providers. This feedback was used in order to help revise the survey. However, the Committee has not sent a condition forward for an evidence review since the surveys were revised.

A. Discussion

The Committee discussion began with a statement on how great the changes have been over the last couple of years. However, in order to get broader information views on the public health impact will be incorporated from data gathered through the use of the Readiness tool as well as through conversations initiated by the surveys. Pilot programs were further discussed along with the use of case studies to further receive data. An example of two-stage surveys was provided in partnership with APHL. However, the pilot programs are limited in terms of how much can be done within the newborn screening program. It was pointed out that as the surveys are completed, information is provided on who provided input, thus allowing access to the appropriate staff in order to potentially receive additional feedback and data. It was then mentioned by the Committee Chairperson that the new survey is estimated to take about 10 hours on average to complete.

V. Adjourn

Dr. Powell adjourned the meeting at 11:27 a.m.