



Newborn Screening Advisory Review Committee

Meeting Minutes

Virtual Meeting Conducted via TEAMS / Tuesday, November 15th, 2022

9:00AM-12:30PM

- I. **Call to order:** The New Jersey Open Public Meeting Law was enacted to ensure the rights of the public to have advance notice of and to attend the meetings of public bodies at which any business affecting their interests is discussed or acted upon. In accordance with the provisions of this act, NSARC has caused notice of this meeting to be published by having the date, time and place thereof posted in the *Courier Post*, November 9, 2022 and the *Star Ledger*, November 9, 2022. This notice is also posted on the State of New Jersey official website see link below: <https://www.nj.gov/health/fhs/nbs/> and filed with the Secretary of State. Members of the public are scheduled to address the committee at the beginning portion of the meeting.

II. **Roll Call**

Absent/ Excused/ Present	Members/Name	Role/Affiliation
P	Sharon Anderson, DNP, NNP-BC, APNG	Neonatal Nurse: Rutgers School of Nursing Rutgers RWJ Medical School
	(vacant)	Nurse
P	Jennifer Barrett Sryfi, MHA	NJ Hospital Association (Resource Representative): Department of Health
P	Dalya Chefitz, MD	General Pediatrician: Rutgers
A	Mary Coogan, Esq.	Advocates for Children of NJ
P	Debra Day-Salvatore, MD, PhD	Geneticist: Saint Peter's University Hospital Chair of Metabolic and Genetic Subcommittee
P	Steven Diamond, MD	Pediatric Hematologist: Hackensack University Medical Center Chair of Hematology Subcommittee
A	Patrick Hill, PhD	Ethicist: Rutgers
	(vacant)	March of Dimes Representative
P	Thomas Lind, MD, FAAP	Medical Director: NJ Dept of Human Services (resource representative)
P	Michael McCormack, PhD, FACMG	Genetics Professor (ad hoc member): Cell Biology and Neuroscience, Rowan-SOM



A	Jeannette Mejias	Family Representative
A	Konstantinos Petritis, PhD	Centers for Disease Control and Prevention (CDC): Laboratory Chief, Biochemical Mass Spectrometry Laboratory, Newborn Screening and Molecular
P	Ernest Post, MD	Chair of NSARC: Chair of Endocrinology Subcommittee
A	Nicole Pratt	Family representative
	(vacant)	NJ Association of Health Plans
A	Geneve Romond	Family Representative
A	Christiana R. Farkouh-Karoleski, MD	Neonatal Physician: American Academy of Pediatrics
P	Andrea Siering, MS, RD, CSP	Nutritionist: Saint Peter's University Hospital
P	Michael Katz, MD	Pediatric Neurologist: Hackensack Meridian
P	Alan Weller, MD, PhD	Pediatrician: Rutgers, The State University of NJ
A	Thomas Westover, MD	Obstetrician-Gynecologist: American College of OB/Gyn
A	Gwen Orlowski, JD	Disability Rights NJ: Executive Director
P	Maryrose McInerney, PhD	Audiologist: Chair of EHDI Advisory Committee
P	Jennifer Heimall, MD	Chair of Immunology Advisory Committee: Allergist-Immunologist CHOP
A	Al Gillio, MD	Chair of Immunology Advisory Committee: Director, Children's Cancer Institute
A	Robert Zanni, MD	Pulmonologist, Monmouth Medical Center Chair: Pulmonology Specialty Group
Absent/ Excused/ Present	Guests/Name	Affiliation
P	Thomas Scanlin, MD	MD Rutgers RWJ Medical School, Chair of Pediatric Cystic Fibrosis Subcommittee
Absent/ Excused/ Present	DOH Employees	Position
P	Mary Carayannopoulos, PhD	Technical Specialist: NBS Lab
P	Miriam Schachter, PhD	Program Manager/Research Scientist 1: NBS Lab
A	Donna McCourt	Microbiologist 4/Quality Assurance: NBS Lab
A	Karyn Dynak	NBS Follow-up Program Coordinator
P	Hui Xing	Research Scientist
A	Diane Driver	Nurse Consultant
P	Michelle Seminara	NBS Public Health Representative 3
P	Suzanne Canuso	Public Health Consultant 1: Nursing, Interim Program Manager for NBS Follow-Up
A	Caitlin Russo	Research Scientist 3: NBS Lab
P	Sarah Eroh	Quality Assurance Specialist



A	Joy Rende, MSA, RNC-E-MNN, NE-BC	Nurse Consultant 1
P	Victoria Floriani	Research Scientist 3
	Jen Hopkins, MPH, CHES	Program Specialist 1
P	Kathryn Aveni	RNC, MPH, Research Scientist 1, Data Systems Coordinator
	Public Attendees	Affiliation
P	Miranda McAuliffe	ALD Alliance: Parent
P	Keely Harris	President Founder: G6PD Deficiency Foundation
A	Anita M. Giddens	Bio-Techne, Inc.
A	Atiyah Abdurraheem	Public Attendee
A	Michelle Sachs	Parent
P	Kellyn Madden	Genetic Counseling student for the University of PA
A	Diane Kane	Public Attendee
P	John Frieri	Public Attendee

Dr. Post called the meeting to order at 9:05 am

Approval of May 17th, 2022, Minutes: APPROVED

	Actions/Resolved
Introductions of Members and Guests: Attendees introduced themselves and their affiliations.	

III. Public Comment on Agenda Items Only:

Motion: To open public comment on agenda items

Call to Order by: Suzanne Canuso **Time:** 9:14 am

Time will be allocated for public comment at this meeting. Members of the public wishing to address NSARC agenda items must state their name, municipality, and the group, if any, they represent. A member of the public shall not be permitted to speak until they are recognized by the presiding officer. Each comment shall be limited to three (3) minutes. No participant may speak more than once. No dialogue between a speaker and the Committee shall extend the three (3) minutes time limit provided herein.

If the meeting is held virtually, participants must place their name and municipality only within the chat section of the virtual platform



(MicrosoftTeamsMeeting). No written questions will be read from this section. Participants must have audio and video capability to present comments and questions during public comment. Participants will be called upon to present in the order their names are received. Although NSARC encourages public comment, the Chair of the Newborn Screening Advisory Review Committee can interrupt, warn and/or terminate a participant’s statement, if question or inquiry is abusive, obscene, or may be defamatory. The Chair of NSARC can request any person to leave the meeting when that person does not observe reasonable decorum. NSARC will use this public comment period as an opportunity to listen to resident concerns, but not to debate issues or to enter into a question-and-answer session. Issues that are raised by the public may require review and investigation and may or may not be responded to by NSARC during the meeting. All comments will be considered, and a response will be forthcoming, if and when appropriate.

Speakers:

- 1) **Keely Harris** (President Founder - g6pd Deficiency Foundation) – Keely is the President and Founder of the G6PD foundation. Keely was first introduced to this disorder when she learned that her grandson was diagnosed with G6PD. Keely believes if detected early enough her grandson could have lived a life with little to no limitations. As of right now he has hearing and speaking issues, problems with walking and has been diagnosed with cerebral palsy. Keely states that G6PD is an enzyme disorder where an estimated 4%-5% of people each year are affected with this deficiency. Keely also discussed the issue of jaundice found in babies as a possible early detector of this disorder. However, since babies do not stay in the hospital as long as they did in the past, this disorder is something that can be missed. It is an important mission of Keely’s to get G6PD to be screening on the Newborn Screening panel. Keely provided her web address for further reference. www.g6pddf.org



Dr. Post thanked the public speaker for her comments.

IV.

Old Business	Actions/Resolved
Subcommittee Reports	
<p><u>Endocrinology Meeting</u> – Dr. Post shared with the group what was discussed at the last endo meeting. At the meeting the group was able to come to a consensus about their management of ALD. The group also spoke about urgent congenital hypothyroidism results stating the lab and follow up are to always report those results out to no matter what day of the week in order to provide timely care. Therefore, protocol will remain the same when reporting out urgent results. When a presumptive result comes through the lab will inform follow up and follow up then get in contact with the clinicians to address the matter with the families.</p> <p><u>Immunology (SCID)</u> – Based on the last meeting Dr. Heimall discussed the development of the graphic imagery project, which would help educate patients about newborn screening targeted towards families who have difficulty with literature and the English language. Dr. Hemaill proposed to the group if they were interested in participating and that volunteers are welcomed to join. The people interest at this time are Jen Hopkins, Dr. Choen and Michelle Seminara. The meeting will be held in January and would like for this project to be comepleted in 2023.</p> <p>The potential of adding an ADA screening as a part of a clarification of a positive newborn screening with trek based testing. The lab informed the group that this is already part of an existing kit and would be there to clarify the data and would not be a new disorder.</p> <p>Lastly, SCID noticed in their data that they are not detecting the expected incidence of cases based on national numbers. Mary is going to analyze these numbers at the lab and see if we need to consider adjusting the cut offs and will be able to present her findings at the next Spring meeting.</p>	



Metabolic and Genetic Subcommittee – As per Dr. Day-Salvatore there are three points she would like to bring up. First, amongst other metabolic and genetics units in the state they were able to combine all of the positive nbs of lysosomal storage disease to try and begin a registry. This data helped her team gain a better sense of what values were of concern, and what values might be more indicative of a true positive. Dr. Day-Salvatore’s team was able to collect a substantial amount of information and wishes to continue with the exercise as they found it to be very valuable.

Secondly, the team worked with the lab regarding the concern of the number of Pompe disease cases and looked at altering the cut off.

Lastly, the group discussed the concern of lack of responsiveness from pediatricians’ offices as it relates to positive newborn screens. Either trying to figure out how to educate all pediatricians about appropriate response times or to determine if we need to rethink how we want to handle positive newborn screens overall.

Hematology (G6PD) – Dr. Diamond informed the committee the Hematology group met to discussed the possibility of adding glucose-6-phosphate dehydrogenase (G6PD) to the New Jersey Newborn Screening panel. However, although this is a continuous topic of interest, no decision has come to fruition as of yet.

Cystic Fibrosis Meeting – Dr. Scanlin spoke about major steps forward in the NBS department. A grant was written for the CF Foundation in 2016 and Dr. Mary Carayannopoulos did most of the heavy lifting. Due to Covid it was difficult to launch but this summer July 27th, 2022, we launched the Illumina 139 which tests for 139 CF mutations. This screening is done on any infant who has an IRT above the cutoff of 70. In order to improve NBS, the lab reduced the cutoff from 90 to 70. As of now 60 is the recommended cut off however, an even stronger



<p>recommendation is to have a floating cut off which as per Miriam we will be able to launch the first half of 2023.</p> <p>Within the last few years more and more of the CFTR modulators are available and patients have expressed that this is a game changer for their care. For patients who have some mutations the modulators are available at four months of age for those who have Kalydeco and the most powerful one, Trikafta is only available to children who are 6 and over. However, the CF foundation just applied to the FDA for approval for Trikafta from two to five years of age. The trial is complete and looks good and now CF is just waiting for FDA approval. This would be extremely helpful to not miss children with CF and be able to diagnose them early so that they are able to take the benefit of the CFTR modulator.</p> <p>In the last 20 years about 200 women with CF would have a baby every year. In 2020 after the modulators were put out there were 615 successful births. In 2021 675 women had successful births. The median survival reached 50 in 2020 and in 2021 it was up to 53.3 which was the largest single year jump ever. Therefore, there is more reason to diagnose these patients early because specialist are able to provide these patients with a really strong treatment.</p> <p><u>EHDI</u> – There was a meeting in October and will have more information for the next meeting in the Spring. As per Dr. Post hearing screening has the most positive NBS results.</p> <p><u>CCHD</u> – As per Dr. Post critical congenital heart disease (CCHD) is not yet a subcommittee for the NBS program because it is an issue that is generally handled at the hospital however, it is a subcommittee we are looking to have in the future.</p>	
<p>New Business</p>	
<p>Overall Incidence and Program to Date Cases–</p> <p>Hui presented the program to date data and discussed with the group.</p>	



<p><u>Ernie announced that the Open portion of the session was completed. The guests were asked to disconnect from TEAMS and Michelle assured that they had done so.</u></p>	
---	--

IV. Adjournment

Meeting Adjourned By: Dr. Post **Time:** 12:10 pm

Next Meeting Date: May 9th, 2023

Minutes submitted by Michelle Seminara