New Jersey Department of Health
Family Health Services

Newborn Screening Advisory Review Committee

Meeting Minutes

Hybrid Meeting at Saint Peter's University Hospital, *CARES Building 3rd Floor Conference Room,* 254 Easton Avenue, New Brunswick, NJ 08901 and via TEAMS

Tuesday, May 20th, 2025 - 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* and the *Star Ledger*, on May 5th, 2025. 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. The meeting will be recorded for quality assurance of the minutes and the committee has been made aware.

II. Roll Call

Absent/ Excused/ Present	Members/Name	Role/Affiliation
A	Sharon Anderson, DNP, NNP-BC, APNG	Neonatal Nurse: Rutgers School of Nursing Rutgers RWJ Medical School
	(vacant)	Neonatal Nurse
P	Jennifer Barrett Sryfi, MHA	NJ Hospital Association (Resource Representative): Department of Health
P	Dalya Chefitz, MD	General Pediatrician: Rutgers, RWJ Medical School
P	Mary Coogan, Esq.	Advocates for Children of NJ
P	Debra Day-Salvatore, MD, PhD	Geneticist: Saint Peter's University Hospital Chair of Metabolic and Complex Disorder Specialty Groups
P	Stacey Rifkin-Zenenberg, MD	Pediatric Hematologist: Pediatric Hematology-Oncology Specialist: Children's Hospital of NJ, Newark Beth Israel Medical Center
A	Patrick Hill, PhD	Ethicist: Rutgers

P	Shakira Williams-Linzey, MPH	March of Dimes Representative
A	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
P	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
P	Dorian Minond, JD	Family Representative
P	Deborah Johnson-Rothe, MD	NJ Association of Health Plans
A	Genene Romond	Family Representative
P	Christiana R. Farkouh-Karoleski, MD	Neonatal Physician: American Academy of Pediatrics
P	Andrea Siering, MS, RD, CSP	Nutritionist: Saint Peter’s University Hospital
A	Michael Katz, MD	Pediatric Neurologist: Hackensack Meridian
P	Alan Weller, MD, PhD	Pediatrician: Rutgers, RWJ Medical School
P	Thomas Westover, MD	Obstetrician-Gynecologist: NJ Chapter, American College of OB/Gyn
A	Gwen Orlowski, JD	Disability Rights NJ: Executive Director
P	Maryrose McInerney, PhD	Audiologist: Chair of EHDI Advisory Committee
A	Jennifer Heimall, MD	Co-Chair of Immunology Advisory Committee: Allergist-Immunologist, CHOP
A	Barbara Spitzer, MD	Co-Chair of Immunology Advisory Committee: Pediatric Hematologist/Oncologist, Hackensack University Medical Center
P	Robert Zanni, MD	Chair: Pulmonology Specialty Group; Pulmonologist, Monmouth Medical Center
Absent/ Excused/ Present	**Guests/Name**	**Affiliation**
A	Mary Ciccone	Director of Policy at Disability Rights New Jersey
P	Vanessa Wills	NJ Division of Medical Assistance and Health Services
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	Sandra Howell, PhD	Executive director for Special Child Health Services
P	Karyn Dynak	NBS Follow-up Program Coordinator: NBS Follow up
A	Hui Xing	Data/Research Scientist: NBS Follow Up
A	Kathy Aveni	Data/Research Scientist 1: Special Child Health Services

P	Michelle Seminara	Meeting Coordinator/NBS Support Program Specialist: NBS Follow up
P	Suzanne Canuso	Program Manager: NBS Follow-Up
P	Sarah Eroh	Quality Assurance Specialist: NBS Follow up
P	Rosalind Finney	Division Director
P	Victoria Floriani	Research Scientist 3: NBS Lab
P	Jing Shi	Research Scientist 1 Birth Defects and Autism Registry
P	Pamela Aasen	Research Scientist 3, EHDI Program
P	Thomas Kirn, MD, PhD	Medical Director, NJ Public Health Laboratories
P	Jessica Redeker, BSN, RN	Quality Assurance Specialist
P	Caitlin Russo	Research Scientist 1
P	Rachel Hammond, JD	Counsel, NJ DoH
P	Kara Unal	Legal Specialist, NJ DoH
P	Lisa Stout	Nursing Consultant, NJ DoH
	Public Attendees	**Affiliation**
P	Emilia Wilburn	Orchard Therapeutics
P	Jennifer Tourjée	Family Representative
P	Kristen Ondy	Mirum Pharmaceuticals
P	Rose St. Fleur	Jersey Shore University Medical Center
P	Emily Glogowski	Sanofi US Rare Disease Medical Affairs
P	Katie Long	Sanofi US Rare Disease Medical Affairs
P	Jeannine Paccioretti	Sanofi US Rare Disease Medical Affairs
P	Nicole Long	Sanofi US Rare Disease Medical Affairs
P	Kelly Boyle	Sanofi US Rare Disease Medical Affairs
P	Paul Vetter	Revvity

Dr. Post called the meeting to order at 9:10 am, Michelle Seminara announced the meeting was being recorded.

Approval of Tuesday, November 19th, 2024, Minutes: Approved without objection.

Introduction of Open Public Meetings Act	**Actions/Resolved**
- Suzanne explained the Open Public Meeting Act (OPMA) in detail to the group and how the Newborn Screening Advisory Review Committee (NSARC) is subject to follow those guidelines.	
Introductions of Members and Guests: Attendees introduced themselves and their affiliations.	

III. <u>OPMA Guidelines regarding Public Comments:</u>

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 Chair. 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. <u>If the meeting is attended virtually</u>, participants must place their name and municipality only within the chat section of the virtual platform (Microsoft Teams Meeting). No written questions will be read from this section. Participants must have audio and video capability to present comments. Participants will be called upon to present in the order their names are received. Although NSARC encourages public comment, the Chair of the NSARC may 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.

Introductions	
Overview of the Special State Officer requirements • Kara shared her screen and presented to the committee as a reminder of their ethics requirements as special state officer members to the NSARC committee. The paperwork and requirements are due and to be sent to Michelle for documentation purposes by the end of the year 2025.	

Speakers

1. Mohammed Harris, PhD

- Expected public attendee was not in attendance.

2. Jennifer Tourjée

- Jennifer addressed the group to advocate for the addition of MLD to the NBS panel. She shared her family's personal journey, including the challenges they faced following the diagnosis of their child and their caregiving experience. Jennifer emphasized the importance of including MLD in the NBS panel and provided detailed information to support her position.

3. Kristen Ondy

- Kristen shared her screen to advocate for the addition of Cerebrotendinous Xanthomatosis, CTX, to the NBS panel. Kristen’s presentation provided a detailed background on CTX that included data to support the importance of its inclusion.

4. Rose St. Fleur, MD

- From Jersey Shore Medical Center, addressed the Committee to ask a question regarding NBS testing. As per Rose, for patients who have requested to leave before a 24-hour period how does the hospital go about screening for that baby without having a missed sample? As per Miriam, this is an important question which is addressed in the New Jersey department of Health regulations for all birthing hospitals within New Jersey. Therefore, if a baby is discharged prior to 24 hours, a NBS sample is to be taken before the baby is discharged. Since the specimen is taken before 24 hours of life it will flag as UNSAT specimen at the lab even if the screen was normal and a repeat would be recommended. If the screening comes back as abnormal a repeat sample will be recommended. Testing kits are also sent to pediatricians’ offices for sampling to be done in office if requested. For moms who deliver at a midwifery center they are mandated to inform the parents that they must bring the baby to the hospital the very next day to receive the following testing, blood spot screening, hearing screening and CCHD screening. Rose was provided with Miriam's contact information and to reach out via email with any additional questions.

Announcements

1. **Lab Updates**

- According to Miriam, the lab has been actively involved in outreach by visiting birthing hospitals to maintain and strengthen relationships. Each month, the lab sends key performance indicator reports to the hospitals. These reports highlight the number of unsatisfactory (UNSAT) samples submitted, samples missing demographic information, and any delays in sample transit to the lab. In an effort to resolve these issues, the lab regularly contacts the hospitals to schedule in-person or virtual meetings to review the data and offer support.
- The lab has created an "All Star" chart to recognize hospitals that perform exceptionally well specifically, those with less than 5% in each of the key performance categories. Each quarter, hospitals that meet this standard receive a star on their chart. This fun and interactive approach encourages engagement and allows hospitals to track their progress while being acknowledged for their efforts.
- In 2022, the laboratory added Spinal Muscular Atrophy (SMA) and Adrenal Leukodystrophy to the newborn screening panel. The lab is currently working to add the analyte used to detect ADA SCID. Once the validation of this analyte is complete, the lab plans to add this analyte over the next several months to the panel to begin screening.
- GAMT was included in the RUSP in January 2023, but it has not been legislatively mandated in New Jersey. Due to its addition to the RUSP, GAMT has been reviewed by NSARC, and there are ongoing efforts to add it to the screening panel. MSPII and cCMV

have legislation in New Jersey and have been reviewed by the NSARC, with ongoing efforts to add them to the screening panel. • Additionally, the lab has partnered with the Public Health Lab and the bioinformatics group to discuss ways they can screen for an increase of variants for cystic fibrosis (CF). The lab is working to establish next generation sequencing (NGS) technology to allow for genetic testing for CF.	
2. Follow up Updates • Suzanne addressed the group to discuss updates within the follow up program. The IRB determination for the proposed Long-Term Follow-up (LTFU) work has been received – it was determined that IRB oversite is not required. Since March, Caitlin and Jessica have been working diligently on long term follow up and working on the logistics. The LTFU component of NBS is focused on the family experience, following up with all confirmed cases, and plans to follow children through 6 years of age. A short presentation will be presented to the committee in the Fall to give an overview of their progress so far. LTFU includes children who have been identified as having a blood spot screening disorder and who reside within the state of New Jersey.	

IV.

	Actions/Resolved
Subcommittee Reports	
Endocrinology As per Dr. Post, the Subcommittee spent their time discussing ways of trying to reduce the number of false positive tests and to reduce the number of tests the lab receives overall. By reducing the workload, it will help make other screens possible. Currently the department of health is updating the recommended regulations, and this was discussed as well.	

Hematology

As per Dr. Rifkin-Zenenberg the Subcommittee met on April 2nd and discussed agreed upon language that gets printed on the mailer for non-definitive results. Parents letters were also discussed for patients who screen abnormal for BARTS and traits. Next, the committee spent time to go over disorders they felt necessary to register for the Birth Defects Registry. The committee decided and agreed upon the newly revised NICU protocol which will eliminate the 7-day screening. They discussed the molecular techniques currently in use, along with insights into how the Newborn Screening (NBS) program is being implemented across the country. The conversation also covered sickle cell grants, their impact on patients, and ongoing efforts to improve diagnosis. Additionally, they highlighted updates to the family and school handbook, as well as the development of new resources for families.

Immunology (SCID)

The Immunology committee met and spoke about ADA SCID and how it should be reported out, regarding comments on the mailer and cutoffs were discussed.

Metabolic and Genetic

The Metabolic and Genetic Subcommittee met to discuss several topics related to cutoff changes, including considerations for POMPE disease, prematurity in infants, and the treatment of newborns receiving total parenteral nutrition (TPN). The subcommittee also reviewed conditions with critical screening levels and discussed the NICU protocol which will be revised to eliminate the 7-day screening for infants.

Additionally, the subcommittee explored the possibility of establishing a metabolic laboratory in New Jersey, in which they went over the challenges involved in setting up such a facility and the difficulties physicians encounter when working with external laboratories.

Finally, there was a recommendation to investigate other states that have transitioned to molecular testing in order to understand how they have successfully implemented these changes.

Cystic Fibrosis Dr. Zanni addressed the group to go over the three main topics of interest during the CF meeting. 1) Moving the floating IRT cutoff forward, and the lab is currently working on the validation process. 2) Expanding the genetic testing panel to the CFTR2 database. The lab is working on this, and the CF foundation just opened up an implementation grant that Mary has decided to apply for. 3) Developing a process for female patients that are pregnant and receiving modulators and how to relay this information to the lab so they can be aware and preform molecular testing for the blood spot specimens. The Cystic Fibrosis Foundation has established a Learning collaborative opportunity for all providers to discuss various treatment protocols and practices. The committee decided to attend the CFTR sequencing and improving communication meetings which will allow for pediatricians and families to meet every other month to discuss how to improve these measures. Regarding moms who are carriers, and pregnant with a baby identified as having CF, Dr. Westover inquired if the CF committee addressed this topic. As per Dr. Zanni, this is still an ongoing issue within the CF community and would require more discussion between the subcommittee at the next meeting in the Fall. **EHDI** The EHDI Committee met on April 28th, with four new council members in attendance. Key topics included the EHDI infrastructure plan, supported by a HRSA grant, and the development of system maps to improve program efficiency. Best practices for congenital CMV (cCMV) screening were reviewed, with updates on targeted hearing screening protocols led by Kathy Aveni, which were distributed to all hospitals. Part of the best practice protocol includes any baby who fails a hearing screen also receives a CMV test.	

The committee also addressed challenges, including federal funding cuts from CDC and HRSA, and the impact on state-level EHDI programs. Long-term follow-up and outreach efforts for CMV-affected families were noted as ongoing. Dr. Westover proposed coordinating OB department chairs and postpartum teams across the state to combine efforts with pediatric departments in all 45 birthing hospitals. Additionally, a CDC-funded grant will evaluate screening timeliness and symptom tracking for all infants. The group also discussed CDC oversight and the potential creation of an ad hoc group to support next steps. **CCHD** As per Suzanne currently the American Academy of Pediatrics (AAP) has developed a new algorithm for pulse ox screening for Critical Congenital Heart Defects (CCHD). The Department of Health, Family Health Services program, will be reviewing to determine if NJ will adopt the new algorithm. Updates will be shared with NSARC at the Fall meeting.	
Old Business	
There were no Old Business topics to discuss at this time.	
New Business	
1. **Removing 7-day specimen for NICU protocol** • The newborn screening regulations are being revised, with a specific focus on the NICU sampling protocol. Current law requires newborns in the NICU to have samples collected at 24 hours of life, and on days 7, 14, 28, and 42, or at discharge, whichever comes first. All samples are tested for congenital adrenal hyperplasia (CAH), congenital hypothyroidism (CH), and any other abnormal result the babies initial sample tested abnormal for. • After discussions with the endocrinology group, it was agreed that the day 7 sample is unnecessary. The delayed rise in TSH is the primary concern and the committee agrees is being properly monitored while	

the infant remains in the NICU. The new recommendation is to collect samples at 24 hours of life, day 14, and day 28 or discharge, whichever is sooner. If the infant is discharged between 5 and 14 days, a sample should be collected at discharge. If the infant remains hospitalized beyond 28 days, a serum sample should be taken at discharge to check for a delayed rise in TSH. • This recommendation was reviewed by other committees, all of which supported eliminating the day 7 sample. Implementing this change will require education for NICU staff and the lab is preparing an educational campaign to launch once the regulatory revision is finalized.	
2. Federal changes affecting NBS • According to Dr. Post, the NBS program is primarily funded through the sale of blood spot screening kits distributed to hospitals and physicians for testing. However, other ongoing programs may be impacted by federal budget cuts. Currently, HRSA has no updates regarding any changes to funding. According to Maryrose, the EHDI program is at risk, and some outreach initiatives have already been suspended due to federal funding concerns. Both the CDC and HRSA grants are currently on hold.	
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.	

I. Adjournment

Meeting Adjourned By: Dr. Post **Time:** 12:10pm
Minutes submitted by Program Support Specialist 1