



**Newborn Screening Advisory Review Committee
Meeting Minutes**

Hybrid Meeting at Saint Peter’s University Hospital, *Sister Marie De Pazzi Conference Center MOB#5* 254 Easton Avenue, New Brunswick, NJ 08901 and via TEAMS

Tuesday, November 18th, 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 October 28th, 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
P	Sharon Anderson, DNP, NNP-BC, APNG	Neonatal Nurse: Rutgers School of Nursing Rutgers RWJ Medical School
P	Marylou Mickalauskas, RN, BSN, CEN, CGN	Nurse: Saint Peter’s University Hospital
P	Jennifer Barrett Sryfi, MHA	NJ Hospital Association (Resource Representative): Department of Health
P	Dalya Chefitz, MD	General Pediatrician: Rutgers, RWJ Medical School
A	(vacant)	Advocates for Children of NJ
P	Debra Day-Salvatore, MD, PhD	Geneticist: Saint Peter’s University Hospital Chair of Pediatric Metabolic and Genetic 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	Family Representative
P	Deborah Johnson-Rothe, MD	NJ Association of Health Plans
A	Geneve 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
P	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
P	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
P	Mary Ciccone	Director of Policy at Disability Rights New Jersey
P	Darius Adams, MD	Atlantic Health Medical Center
P	Rakesh Chhabra, MD, FAAP	Hackensack Meridian School of Medicine
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
P	Sandra Howell, PhD	Executive director for Special Child Health Services
P	Karyn Dynak	NBS Follow-up Program Coordinator: NBS Follow up
P	Hui Xing	Data/Research Scientist: NBS Follow Up
P	Kathy Aveni	Data/Research Scientist 1: Special Child Health Services
P	Michelle Marinilli	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
	Pamela Aasen	Research Scientist 3
P	Thomas Kirn, MD, PhD	Medical Director
P	Jessica Redeker, BSN, RN	Quality Assurance Specialist
P	Caitlin Russo	Research Scientist 1
P	Margaret Roberts	Regulatory Officer 2: Division of Public Health & Environmental Laboratories
	Public Attendees	Affiliation
A	Emilia Wilburn	Orchard Therapeutics
A	Terry Donahue	Public attendee
A	Aryan Doshi	Public attendee
P	Andrew Shenker, MD	Consultant for Orchard Therapeutics
A	Melanie Rumbel	Orchard Therapeutics
P	Mousumi Bose, Ph.D.	Montclair State University Professor and Parent advocate for MLD screening
P	Emily Pecot	Community Health Law Project
P	Paul Vetter	Revvity
P	Andy Betzer	Parent advocate for MLD screening
P	Susheela Jayaraman	Orchard Therapeutics

Dr. Post called the meeting to order at 9:08 am, Michelle Marinelli announced the meeting was being recorded.

Approval of Tuesday, May 20th, 2025, Minutes: Approved without objection.

Introduction of Open Public Meetings Act	Actions/Resolved
<ul style="list-style-type: none"> Suzanne explained the Open Public Meeting Act (OPMA) in detail to the group and how the Newborn Screening Advisory Review Committee (NSARC) follows those guidelines. 	

Introductions of Members and Guests: Attendees introduced themselves and their affiliations.	
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III. OPMA Guidelines regarding Public Comments:

Time will be allocated for public comment at this meeting. Members of the public wishing to address NSARC 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. Public comments should proceed without interruption. This is not a time for a question-and-answer exchange with the Board, Council, or Commission. If the meeting is attended virtually, 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	
Speakers	
<ul style="list-style-type: none"> • Mousumi Bose, Ph.D. – Dr. Bose addressed the committee to discuss Metachromatic Leukodystrophy (MLD) and advocated for its inclusion in the New Jersey Screening Panel. Dr. Bose highlighted the advantages of adding MLD to the screening panel, emphasizing that early detection would enable children affected by the condition to receive timely care and treatment, thereby improving their outcomes. Newborn screening for MLD has gone live in New York, and Pennsylvania is set to follow suit. Dr. Bose encouraged New Jersey be the next state to consider adding MLD to its screening panel to improve the outcomes for children in New Jersey. 	<p>Chair’s note: NY is doing universal MLD NBS but considers it to be a pilot. IL and Minnesota have added it to their panels.</p>
Announcements	
<p>1. Lab Updates – Miriam Schachter</p> <ul style="list-style-type: none"> • The lab has enhanced its collaboration with hospitals and launched the NBS All-Star program. This initiative includes all hospitals that submit samples to the laboratory. Each month, hospitals are evaluated based on three key performance indicators: <ol style="list-style-type: none"> 1) Rate of unsatisfactory specimens 2) Rate of missing demographic information 3) Any delay in receiving samples • Hospitals must achieve less than 5% in all three categories to receive a sticker and recognition of their performance. So far, seven hospitals have earned a sticker for each quarter. Additionally, hospitals can reach out to the lab for in-person or virtual in-services to address challenges, answer questions and improve training and education on newborn screening specimen collection. • The lab remained open on Memorial Day and Labor Day thanks to the new carrier, which has improved the timeliness of sample testing. 	

<ul style="list-style-type: none"> • The cut off for Cystic Fibrosis (CF) has changed from a fixed value of 70 to a floating cut off. The lab now tests the top 4% for molecular analysis, aligning with the CF Foundation guidelines. • The cut offs for Pompe Disease and Severe Combined Immunodeficiency (SCID) have also been adjusted to maintain accurate testing, a change prompted by the lab’s quality assurance measures. • The implementation of the EMR message system between hospitals and the lab is ongoing. The lab is in the process of purchasing the necessary software. Miriam will provide updates as more information becomes available. 	
<p>2. Follow up Updates – Suzanne Canuso</p> <ul style="list-style-type: none"> • The follow up program has been providing support to the lab by adjusting staffing schedules to ensure coverage on open holiday workdays. The long-term follow up initiative is now operational and will give a presentation later in the meeting. 	

Subcommittee Reports	Actions/Resolved
<p>Endo</p> <p>At the Endocrinology meeting they discussed that the lab is readjusting the congenital hypothyroidism algorithms to reduce screened positive babies without increasing the risk of missing potential CH cases. Crenessity is a new treatment medication approved for people ages 4 and up with classic CAH. It is designed to lower levels of androgens, which may reduce the need for high doses of glucocorticoids. This is a new significant change of treatment for CAH. Unusual cases of CAH were presented by Dr. Westover at the endo meeting and a congenital hypothyroidism case was also discussed.</p> <p>Hematology</p> <p>The hematology group discussed how to move towards Next-Generation Sequencing (NGS) as a confirmatory method for hemoglobinopathies. NGS will be particularly beneficial for patients requiring additional testing for conditions that are often difficult to diagnose due to insurance constraints or</p>	

accessibility challenges. Additionally, the group addressed the need for educational updates and the revision of Sickle Cell brochures to ensure that practitioners and families have access to the most current information.

SCID

The immunodeficiencies group discussed the positive impact of the TREC-based assay, which identifies most forms of severe combined immunodeficiencies (SCID) and significant T-cell defects. The lab is progressing with the addition of ADA screening to identify patients who may not be detected by the TREC-based newborn screen (NBS). This new screening will utilize tandem mass spectrometry, which is prevalent in certain communities due to a founder effect. The Department of Health (DOH) will notify referral centers conducting follow-up testing to inform them about these enhancements to the current process.

Metabolic/Genetic

The Combined Diseases subcommittee discussed genomic newborn screening and the various forms and opportunities it offers. The committee discussed their efforts to contract for second tier testing to screen for Krabbe and other conditions. Dr. Day-Salvatore received results from a Cross-Reactive Immunological Material (CRIM) assay that reported incorrect results from an outside laboratory and was able to correct the issue and get the patient on the correct care plan. It was determined that in order to properly diagnosis Pompe the CRIM assay should not be the only diagnostic test performed, and other evaluations of the patient need to be done to determine the correct diagnosis. During the meeting Dr. Adams presented on Metachromatic Leukodystrophy (MLD). Reflecting the decision of the Metabolic and Genetic subcommittee, Dr. Day-Salvatore wrote a letter to Dr. Post recommending MLD to be added to the New Jersey newborn screening panel. In support of this recommendation Dr. Day-Salvatore included an information packet created by the RUSP, pediatric article and publication of the New England Journal of

<p>Addition to Article VII - NSARC Pediatric Specialist Subcommittee:</p> <p>4. Currently, there are 6 subcommittees in existence. Dr. Post is proposing the addition of a 7th subcommittee, CCHD:</p> <ol style="list-style-type: none"> 1. Metabolic and Genetic 2. Endocrinology 3. Immunology (SCID) 4. Cystic Fibrosis 5. Hematology (Sickle Cell Disease) 6. Neurology 7. CCHD <p><i>Motion: Dr. Day-Salvatore moved to approve all the above changes. Seconded by Deborah Johnson-Rothe. The vote was taken by show of hands. The motion passed with 18 in favor.</i></p>	
<p>2. Long term follow-up presentation</p> <ul style="list-style-type: none"> • The long-term follow-up (LTFU) program is designed to ensure that children and families identified with a condition on the New Jersey Newborn Screening (NBS) panel have access to appropriate healthcare and community resources. The program also seeks to identify strengths and gaps in existing services and to better understand the challenges and barriers that impact effective chronic disease management. • To support these goals, families will receive a brief annual survey—approximately 5–10 minutes to complete—each year from age 1 through age 6. The survey includes general questions relevant to all NBS conditions, covering topics such as access to primary and specialty care, the child’s overall health, health management practices, and available support services or organizations. • The LTFU program uses REDCap, a secure database platform, to distribute surveys and store responses. Families also receive a 	

<p>welcome letter introducing the program, explaining its purpose, and providing next steps along with helpful resources and links.</p> <ul style="list-style-type: none"> • To date, the LTFU program has sent 201 welcome letters, reached 104 families by phone, distributed 18 surveys, and received 7 completed responses. • The program continues to explore strategies to improve survey distribution, increase family engagement and participation, and expand survey accessibility through translations into additional languages. 	
<p><u>Ernie announced that the Open portion of the session was completed. Executive committee members were asked to log onto the executive session via TEAMS.</u></p>	
<p><u>Executive Session (not open to public) started at 11:00 am</u></p>	

I. Adjournment

Meeting Adjourned By: Dr. Post **Time:** 12:21pm
 Minutes submitted by Program Support Specialist 1