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, November 19th, 2024 - 9:00AM-12:00PM

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 22nd, 2024. 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
	(vacant)	Nurse
P	Sharon Anderson, DNP, NNP-BC, APNG	Neonatal Nurse: Rutgers School of Nursing Rutgers RWJ Medical School
P	Andrea Siering, MS, RD, CSP	Nutritionist: Saint Peter's University Hospital
A	Patrick Hill, PhD	Ethicist: Rutgers
A	Jeannette Mejias	Family Representative
A	Nicole Pratt	Family Representative
A	Genene Romond	Family Representative
P	Alan Weller, MD, PhD	Ad hoc member: Public Health Physician: Rutgers, RWJMS
P	Michael McCormack, PhD, FACMG	Ad hoc member: Genetics Professor: Cell Biology and Neuroscience, Rowan-SOM
A	Gwen Orlowski, JD	Disability Rights New Jersey
P	Thomas Westover, MD	American College of OB/Gyn: Capital Health

P	Christiana R. Farkouh-Karoleski, MD	American Academy of Pediatrics
P	Deborah Johnson-Rothe, MD	New Jersey Association of Health Plans
P	Konstantinos Petritis, PhD	Centers for Disease Control (CDC): Laboratory Chief, Biochemical Mass Spectrometry Laboratory, Newborn Screening and Molecular
P	Jennifer Barrett Sryfi, MHA	NJ Hospital Association: Department of Health
A	Mary Coogan, Esq.	Advocates for Children of New Jersey
P	Shakira Williams-Linzey, MPH	March of Dimes Representative
A	Thomas Lind, MD, FAAP	New Jersey Dept of Health Services: NJ Medicaid
P	Dalya Chefitz, MD	General Pediatrician: Rutgers, RWJ Medical School
A	Michael Katz, MD	Pediatric Neurologist: Hackensack Meridian
P	Debra Day-Salvatore, MD, PhD	Geneticist: Chair of Metabolic and Complex Disorder Specialty Subcommittee: Saint Peter's University Hospital
P	Stacey Rifkin-Zenenberg, MD	Hematologist: Chair of Hematology Specialty Subcommittee: Hackensack University Medical Center
P	Ernest Post, MD	Endocrinologist: Chair of Endocrinology Subcommittee and NSARC: Cooper University Hospital
P	Robert Zanni, MD	Pulmonologist: Chair of Cystic Fibrosis Specialty Subcommittee: Monmouth Medical Center
A	Jennifer Heimall, MD	Allergist-Immunologist: Chair of SCID Specialty Subcommittee: CHOP
P	Maryrose McInerney, PhD	Chair of Early Hearing Diagnosis and Intervention Advisory Committee
Absent/ Excused/ Present	**Guests/Name**	**Affiliation**
P	Mary Ciccone	Director of Policy at Disability Rights New Jersey
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
A	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 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 Scientist2: NBS Lab
P	Jing Shi	Research Scientist 1 Birth Defects and Autism Registry
P	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
	Public Attendees	**Affiliation**
P	Dorian Minond	Parent of a child with Pyruvate Dehydrogenase Complex Deficiency advocating that PDCD be added to the NBS panel in NJ
P	Paul Vetter	Revvity
A	Smrithi Venkatesh	Genetic Counseling Student

Dr. Post called the meeting to order at 9:02 am, Michelle Seminara announced the meeting was being recorded.

Approval of Tuesday, May 7h, 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. OPMA Guidelines regarding Public Comments:

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. 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.

Speakers	
Dorian Minond – Mr. Minond addressed the group to talk about his journey advocating for his daughter who has been diagnosed with Pyruvate Dehydrogenase Complex Deficiency (PDCD). Mr. Minond spoke about the issue of false positives and challenges with effective treatment for this disorder. Jiraria Krikor Bedoyan, MD is a clinical geneticist in Pittsburgh associated with Children's Hospital of Pittsburgh and has been involved in research and performed pilot studies enabling him to gather more information on PDCD. Mr. Minond believes that further research is necessary and is interested in exploring the possibility of New Jersey sharing their newborn screening data with researchers to help validate or refine approaches for analyzing amino acid levels.	
Announcements	
1. **Passing of Thomas Scanlin, MD** • The committee took time to address the passing of Dr. Scanlin. Mary spoke about Dr. Scanlin, his passing, and the incredible work and dedication he had to children and families dealing with the challenges of cystic fibrosis. Dr. Scanlin will be deeply missed.	

2. Lab Updates

- Miriam addressed the group to go over accomplishments and updates the lab has experienced over this past year. In 1964 newborn screening in New Jersey began with screening for PKU and now 60 years later screens for 61 disorders from the dried blood spots. To celebrate 60 years of newborn screening in NJ the lab collaborated with follow-up to host an event. Families, stakeholders, staff, doctors, and specialist were all in attendance for this event. The lab provided tours, and a family panel presented their stories on how NBS positively affected their lives.
- During the months of March through August the lab and follow-up staff visited all 45 birthing hospitals within NJ. This was an opportunity to educate the medical professionals in the hospitals about the importance of NBS as well as collecting updated contact information from each hospital. The lab had a presentation and distributed Badge Buddy’s which are used as a tool to remind staff on how to properly take a NBS sample. The mantra of the Badge Buddy is “No Blank Spot”, which is a play on the acronym for newborn screening. The “N” represents newborns are all tested, “B” boxes are filled in legibly, and the “S” is for satisfactory specimens. Overall, this initiative was highly successful with positive feedback from various hospitals involved.
- The new specimen retention policy went into effect on Nov 1st, 2024. The lab was previously storing samples for 23 years and now will store samples for two years. Parents can request early destruction of the specimen or to request to store the sample longer. Forms to make such requests can be found on the NJ state NBS lab website. Once a family sends the form to the lab, the lab will fulfill the request and once completed mail a letter out to the families informing them that their request has been completed. The retention policy can also be viewed on the state NBS laboratory website.

Currently, the lab is working on keeping the specimens in an environmentally controlled space and is close to achieving this goal.

- **Link to request forms:** https://www.nj.gov/health/phel/public-health-lab-testing/newborn-screening-lab/
- Lastly, the NBS program received a grant, and the lab is utilizing their funding portion to begin the implementation of HL7 electronic messaging between hospitals and the NBS laboratory. Revvity has a software system to help set up that communication and will be working with the lab and NJ hospitals. This will be an ongoing project for the lab, and they will begin to reach out to the hospitals to start this process.

3. **Follow-up Updates**

- As per Suzanne the follow-up program has hired two new employees Caitlin Russo and Jessica Redeker who will be working on long term follow-up tasks for the newborn screening program. Currently, follow-up is waiting for an IRB determination and hopefully by the end of this month they will be receiving feedback.
- Dr. Westover asked if the IRB application can be circulated to the committee. As per Suzanne if the IRB states that they need oversite, then the follow-up program will need to go with what was submitted. Once this process is done, it can be shared with the committee.
- Dr. Post asked if the retention forms have been translated into Spanish? Currently, the family brochures that are distributed to families have a Spanish and English version however, retention forms have not been translated into Spanish. Miriam and the lab are working on having more discussions to resolve this.

IV.

	Actions/Resolved
Subcommittee Reports	
Endocrinology • The endocrine committee talked about the protocols and retesting for NICU babies. It was decided by the committee to cut back on the amount of retesting for NICU babies as well as cut back on presumptive positive retesting. False negatives were also discussed and the urgency of reporting out presumptive positives. **Hematology** • The lab gave an update to the committee to discuss their upcoming changes. The committee spoke about the benefits of repeat testing for hemoglobinopathies at 4 weeks of age and how they would like to continue to be able to do this and ways in which the lab could accommodate this request. Dr. Carayannopoulos addressed various cases that were of interest to the committee which they discussed at length. **Immunology (SCID)** • Dr. Spitzer has joined the group and alongside Dr. Heimall will be taking on the responsibility of co-chair. • ADA SCID may be missed by the TREC screening that the lab does. Therefore, since there is an analyte in the current mass spec kit, the lab will be moving forward in validating that analyte so they can add screening for the ADA subset of SCID, in hopes to not miss it. Sharon Anderson asked, what is the incidence of SCID? As per Miriam there is a range of one in 60,000, resulting in approximately one or two cases per year in New Jersey. **Metabolic and Genetic** • Dr. Day- Salvatore, stated that the committee discussed an update on variant vs classic, cutoff for biotinidase, late onset for Pompe and reviewed updates on SUAC and tyrosine, the ability of second tier testing for LSDs, and possible biomarkers vs molecular testing. The	

NSARC decision regarding PDCD was also addressed. Finally, the committee spoke about the utility of not repeating screens after a prior presumptive positive and the lab reporting out on Memorial Day and Labor Day.

- The committee also addressed an issue regarding a hospital that sent out 2 repeat screens for one patient, one to the state and one to a commercial lab. This resulted in a disparity in recommendation and lead to confusion. It was agreed upon that a recommendation from NJ state is what physicians should be following and it is not appropriate for hospitals to rely on commercial lab recommendations. The commercial lab is an expansion of NBS which is separate and in addition to.

Cystic Fibrosis

- As per Dr. Zanni we are moving IRT from a fixed cutoff to a floating cutoff which is hoped to be implemented early 2025. The group discussed CFTR variants and moving to a second-tier assay that would detect all variants in the CFTR2 database (expanding from the 139 variants currently detected). In 2025 the meeting guidelines should be finalized and will be brought back into discussion at the next Spring meeting.
- Women with CF who utilize CFTR modulator therapy during pregnancy may expose their unborn children, which could cause a false negative NBS. The committee needs to discuss how they will move forward to alert the state of these unique cases.
- Dr. Day-Salvatore discussed a case with the group regarding a fetus that was diagnosed with CF and echogenic bowel with a likely meconium blockage. The parents asked whether the maternal use of a modulator might assist with the meconium ileus. As per Dr. Zanni moms that are carriers of CF and having issues with their unborn babies possibly having meconium ileus can benefit and potentially reverse this by taking a modulator. The problem is getting the

medication to mothers who are not diagnosed with CF and to have it covered by insurance. The modulators are expensive causing challenges for providers to help get coverage for these patients which can cause stress and financial burdens for families. This is an ongoing issue for the CF foundation; therefore, they are working together to figure out a plan on how to get this resolved. **EHDI** • At their most recent meeting, which took place on October 28th, they talked about the 1-3-6 algorithm nationwide and the percentage of New Jersey families who are successfully adhering to it. The baby is screened at one month of age, and NJ has a 98.4% rate; at three months, the baby is diagnosed, and NJ has a 32.2% rate; and at six months, the baby is followed up with programs, and NJ has a 42.9% rate. The committee wants to improve those results. Each state has been awarded a HRSA grant to help make improvements. • Dr. McInerney has participated in many focus groups and theme analysis nationally to understand why families may not be following up with care. This research identified geographic challenges, insurance, scheduling, and social economic determinants. As per Dr. McInerney, Black infants account for 30% of cases lost to follow-up, despite representing only 13% of births in the state. • Special Services met with the committee and talked about what they can do for children with suspected hearing loss and diagnosed hearing loss. They provided the committee with an infographic to share with families as an easier guide on how to properly manage and move through the appropriate next steps in care. • Pam Aasen added that to increase awareness, hearing consultants have prepared a PowerPoint presentation that includes the information on the infographic, and they are planning to share it with early intervention service providers over the next year.	

- **Link to infographic:** [Usher Syndrome Parental Support Needs Infographic]

CCHD

- No representatives were present, and it was not discussed during the meeting.

Old Business

1. **Update on NBS 60th Anniversary event**

- On September 19th the lab and follow-up hosted a 60th anniversary event. The lab provided tours of the laboratory allowing people to see how the lab operates, what tasks they perform, understand the instruments used, and timeliness of their process. A family panel of individuals with PKU, CF and Sickle Cell were in attendance and shared their stories and experience with NBS. The feedback from the event was very positive and an overall huge success.

New Business

1. **ICoNS meeting update**
    - Dr. Westover shared with the group notes from the International Consortium on Newborn Sequencing meeting. It was hosted in NYC and has been floating between the UK and the United States. This was a two-day event. Many research teams were represented and discussed if whole genomic sequencing belongs in NBS and how this would work. Nothing is definitive yet and since the newborn screening system is working very well, they do not want anything to interfere with that system.
2. **Association of Public Health Laboratories, APHL NBS Symposium Update**

    A) NJ round table/posters

- As per Suzanne the APHL NBS symposium update discussed how to target pediatricians, parents, and the community and educate them on NBS, which has been an ongoing endeavor and will continue to be so.

The RUSP was discussed and how they will begin to restructure the list, including ways a disorder can be reconsidered or removed. • In May the FDA came out with a new rule for state laboratories that they must follow. Labs that are implementing testing that is not approved by the FDA will need to start following a process to obtain FDA approval. Therefore, continuing to test the second-tier test for CF and looking into testing for variants included in the CFTR2 database, will need to be on hold as it is not FDA approved and the lab will need to go through the process of obtaining approval. • Victoria shared her screen to discuss the presentation at the APHL meeting. The lab had the opportunity to present a round table to discuss CH screening in older infants. There were break out discussions and the results on those discussions were presented at length. • Two of the labs supervising medical technologists shared poster presentations at APHL. Pallavi Patel presented a poster entitled, Continuity of Operations Plan: The Mass Spectrometry perspective. Laxmi Nayak presented a poster entitled, Lot-to-lot comparison of GSP Neonatal hTSH kit reveals a significant negative bias attributed to a manufacture change in reagent formulation. A summary of both poster presentations was discussed with the group.	
3. Reporting out CAH on Memorial Day and Labor Day • In the past, the lab has been open on most state holidays, receiving specimens and calling out time critical results, primarily metabolic and CAH disorders. Since the new courier transport system can deliver on all holidays, beginning in 2025, the lab will report time critical results and be open on Memorial Day and Labor Day. Since both Memorial Day and Labor Day fall on a Monday being open both holidays will alleviate back up of specimens.	

• Moving forward on the years where New Years, Fourth of July, or Christmas happen to fall on a Monday, the lab and follow-up program will work together to have the program operational on those days.	
4. Not ordering/repeating screens after prior presumptive positive • Currently, if the lab receives a repeat sample for a baby whose initial sample screened presumptive positive, the lab will run the repeat for whatever the infant tested positive for, plus CH and CAH. All repeat samples are tested for CH and CAH. For most disorders the lab does not recommend a repeat sample after a presumptive positive, however, the lab recommends they seek guidance from a specialist. Therefore, to eliminate the number of specimens tested by the lab and reduce confusion of a possible normal screen after an abnormal, they would like to stop repeat screens after prior presumptive positives are reported. The purpose of this change would be to avoid delay in diagnosis and to encourage more timely referrals to specialists. • The Hematologist, however, rely on the 4-week repeat screen and utilize this in a diagnostic algorithm therefore, the lab will continue to ask for a repeat hemoglobinopathy screen to not interrupt their flow of care. • It is important for us to try to educate the pediatricians ordering the repeat at the lab. • Paul Vetter will see if across the RUSP if there is any cutoff with age limit and testing. • The regulations expire every 7 years and will expire February 2025. A big overhaul was made and is in the process of going through approval, open public comment, and addressing those comments before getting approved.	

5. **CLSI updates** • Dr. Post addressed the group and shared his screen and guidelines that are currently available through NBS 10. Dr. Post asked the group that if anyone has suggestions to offer to reach out to himself or Michelle.	
6. **Mailer comments and review by pediatricians** • As per Miriam all mailer comments need to be revisited and reassessed for edits. Miriam asked if a group of pediatricians can come together to begin working on this initiative. Dr. Weller will reach out to NJAAP to see if he can get a group together and Miriam will work with Dr. Weller to make this happen.	
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:** 12:01pm
Minutes submitted by Program Support Specialist 1