Newborn Screening Advisory Review Committee (NSARC)

Meeting Minutes

Virtual Meeting Conducted via TEAMS / Tuesday, November 14th, 2023
9:30AM-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*, May 5, 2023, and the *Star Ledger*, May 5, 2023. 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. Michelle Seminara made an announcement that for quality assurance of the minutes the meeting was being recorded.

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)	Neonatal Nurse
P	Jennifer Barrett Sryfi, MHA	NJ Hospital Association (Resource Representative): Department of Health
A	Dalya Chefitz, MD	General Pediatrician: Rutgers, RWJ Medical School
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 Complex Disorder Specialty Groups
	(vacant, pending CoH approval)	Pediatric Hematologist: Hackensack University Medical Center Chair of Hematology Subcommittee
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

	Nicole Pratt	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
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
P	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	Al Gillio, MD	Co-Chair of Immunology Advisory Committee: Director, Children's Cancer Institute
A	Robert Zanni, MD	Chair: Pulmonology Specialty Group; Pulmonologist, Monmouth Medical Center
Absent/ Excused/ Present	**Guests/Name**	**Affiliation**
A	Alena Siddiqui	ACNJ's Kids Count Coordinator
P	Mary Ciccone	Director of Policy
P	ShaluNarang, MD, MPH	Pediatric Hematology-Oncology Specialist: Children's Hospital of NJ, Newark Beth Israel Medical Center
P	Stacey Rifkin-Zenenberg, DO, FAAP	Hematology-Oncology Specialist: Joseph M. Sanzari Children's Hospital at Hackensack University Medical Center
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	Dawn Mergen	Program Specialist 4: Social/Human 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 Seminara	Meeting Coordinator/NBS Public Health Representative 3: NBS Follow up
P	Suzanne Canuso, MSN, RN	Program Manager: NBS Follow-Up
P	Caitlin Russo	Research Scientist 3: NBS Lab
P	Sarah Eroh	Quality Assurance Specialist: NBS Follow up
P	Victoria Floriani	Research Scientist 3: NBS Lab

P	Nancy Kearney	Communications Manager: Deputy Communications Director
P	Rosalind Finney	Division Director: Department of Health
P	Jing Shi	Research Scientist 1 of birth defects and autism registry
P	Dalya Ewais, PCM, CDMP	Director Office of Communications: Department of Health
P	Julia Bixler	Program Specialist 2 Social Human Services: Department of Health
P	Morgan Spanier	Public Health Representative 3: Department of Health
	Public Attendees	**Affiliation**
P	Yug Raj Yadava	senior at Rowan University; presenting regarding metachromatic leukodystrophy
P	Kathy Hosking	Case Manager for Warren County Special Child Health
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 Aronsohn	Ombudsman for Individuals with Intellectual or Developmental Disabilities and their families
P	Didar Mohammed Yahia	Student at William Peterson University
P	Dana DiFilippo	Reporter, NJ Monitor
P	Brian Morris	Attorney with the Institute for Justice

Dr. Post called the meeting to order at 9:05 am, Michelle Seminara announced the meeting was being recorded.
Approval of Tuesday, May 9th, 2023, Minutes: Approved without objection, motion to approve First, Sharon Anderson, Second, Deborah Johnson-Rothe.

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 held 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 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	
Yug Raj Yadava – Yug Raj is a senior at Rowan University and has joined this meeting to discuss the possibility of adding Metachromatic leukodystrophy (MLD) to the New Jersey Newborn Screening Panel. Yug Raj provided the group with detail explaining this disorder. It is estimated that within the United States MLD affects 1:4700 births. The state of NY is doing a pilot study and the state of Illinois is currently looking into implementing MLD into their program. Dorian Minond – A parent of a baby born with Pyruvate dehydrogenase complex deficiency (PDCD) and diagnosed by Dr. Day-Salvatore, addressed the committee. This parent shared with the group the family's experience and importance of adding PDCD to the NBS panel. Dr. Bedoyan of UPMC Children's Hospital of Pittsburgh has recently conducted a pilot study of newborn screening for PDCD in Ohio. Through his research, he estimated that roughly 1:40,000 newborns have primary PDCD. Although there isn't a treatment for this illness at the moment, research has shown that early ketogenic diet intervention and some supplements can significantly improve outcomes. Didar Mohammed Yahia – Didar, a student at William Paterson University, suggested to the committee that improving accessibility to different language handouts and brochures in different languages on the Department of Health website would be helpful to non-English speaking families. Currently, the website has a Google Translate function that can convert the language from English to any other language. However, some handouts within the website only come in English or Spanish versions. Didar suggested that the PDF handouts and brochures already on the website to be turned into website content so that the Google translate function could do most of the heavy lifting and convert the information from English to whichever language they choose.	

Announcements	
Lab Updates: • Dr. Schachter discussed the private courier transport project that has been taken place. The 46 different birthing hospitals have been split up into four different routes throughout New Jersey. Each route has their own driver, and each driver visits each hospital every-day Sunday through Friday. The private courier, 24/7 Enterprises has two of the four routes currently running, the South route will go live 11/15/23 and the Northwest route will go live mid-December. The overall, benefit to this project is to make sure specimens arrive to the lab within a timely fashion and without damage. • The lab currently was awarded the HRSA Propel grant to help focus on implementing electronic messaging between the hospitals and the lab. A software developed by Revvity formerly known as Perkin-Elmer has implemented the HL7 messaging system in other states. The system will allow the hospital to send babies' demographic information to the lab electronically and allow the lab to send the NBS results to the hospitals electronically for inclusion in their Electronic Medical Records systems. This eliminates handwritten demographics and human error. This will be a drastic improvement in communication. • The overall implementation of this new electronic messaging system will be a multi-year project for the lab.	
Follow up Updates: • As for the follow up, the program will use the Propel grant to focus on establishing Long Term Follow Up utilizing the current Case Management infrastructure to evaluate program quality metrics such as equitable access to care for families, identifying barriers and gaps in services. • As per Ms. Canuso, the follow up program has had a few staffing changes. A long-time rep for the program has since retired, and due to this another staff member has been trained efficiently to take her place.	

IV.

	Actions/Resolved
Subcommittee Reports	
Cystic Fibrosis: • Co-chairs were not present therefore, no update was given. Endocrinology: • The incidence of Congenital Hypothyroidism has essentially doubled over the last decade or two. The numbers have risen to	

1:1000 babies as of 2022, which is extraordinarily high. Before starting NBS it was thought to be 1:4000 births. This rise in numbers did not happen suddenly; however, steadily increasing over the years. The reason behind this is unknown and further investigation is needed.

- The group at the endo meeting discussed X-linked adrenoleukodystrophy, XALD and so far, there are no confirmed cases.
- TSH lab cutoff was changed for the first time in several years due to a reagent modification by the vendor. A new set of reagents was not performing as it should, and the lab was able to catch and address this issue by changing the cutoff.
- Lastly, the group discussed how to reduce the burden of false positive results for CH, notifying the NICUs and those babies to try and simplify that process. The subcommittee and Department of health staff are also revisiting the current policy in the Regulations on retesting NICU babies at 7 days, 14 days, and 42 days of life.

Metabolic and Genetic:

- Dr. Day Salvatore spoke to the committee on three areas of business. At the recent meeting, Dr. Pedro discussed the addition of guanidinoacetate N-methyltransferase (GAMT) to the Recommended Uniform Screening Panel (RUSP). Revvity is looking to add the analytes for the FDA to approve testing for this disorder. This still holds true for Hunters Syndrome, also known as MPS2. While the subcommittee is in support of adding GAMT to the NBS panel, they will hold off until the lab has FDA approved kits.
- The second matter of business discussed that all positive NBS for LSD should be referred to an approved center for further evaluation and confirmatory testing. Repeat screens should not be sent to the NBS lab.
- Lastly, it was decided to create a borderline category for SUAC. Levels between one and two would be considered borderline and request for a repeat filter would be recommended. Therefore, levels greater than two would be considered presumptive and referred to a specialist for diagnostic testing. If there is a borderline for SUAC and tyrosine, it would be treated as a presumptive irrespective of the tyrosine and referred for further evaluation.

CCHD:

- No group has yet been established for this subcommittee. Dr. Howell spoke about heart defects stating that the Department of Health receives reports of all the failed pulse oximetry screens and explained to the committee how they are followed. Data on their findings can be provided at the next meeting in the spring.

EHDI: • Dr. McInerney informed the committee that the EHDI hearing evaluation council will be meeting in December. Currently, they have put into place a better communication system to share results with the pediatricians and have made updates to their website. Also shared was the cCMV CSTE statement that Dr. McInerney will send to Dr. Post and Ms. Seminara to later distribute as a handout for the Spring NSARC meeting. Immunology Meeting: • Co-chairs were not present therefore, no update was given. Hematology: • Due to Dr. Diamond's retirement, two new co-chairs, Dr. Shalu Narang and Dr. Stacey Rifkin-Zenenberg, were announced to the group. • Dr. Rifkin-Zenenberg then presented the exciting work involving new treatments for Sickle Cell Disease. She gave a presentation entitled "No Donor, No Problem", which is the concept of the newly found treatment where clinicians no longer need a stem cell donor to treat the patient. • Gene therapy treatments has been submitted for FDA approval and expected approval for this will be by the end of December. • More detail about gene therapy was discussed at length within the committee.	
Old Business	
Newborn Screening Awareness month (September): • September is designated as NBS Awareness Month in New Jersey. The New Jersey Newborn Screening Program will commemorate its 60th anniversary in September 2024. As a result, the lab and follow-up have been trying to come up with some ideas for how to make the celebration run smoothly. All parties involved in the occasion are invited, including families, DOH employees, and organizers of the NBS hospital. Dr. Schachter asked the clinicians to start thinking about potential families to be invited to the celebration. As more planning develops the lab will provide valuable information within the next couple of months.	
NSARC ad hoc Enhancing Education Follow Up: • Dr. Post shared his screen and presented the NSARC the ad hoc Enhancing Education report to the group. The report goes over the target audience, issues, communication methods, resources, accomplishments, ideas, and next steps. Implementing new ways of enhancing education will continue to be an ongoing topic as new advances and developments take place within the program.	

New Business	
Case Management: • Morgan and Julia shared their screen to go over the specifics of the New Jersey, Case Management Program. Case managers are registered nurses or social workers. They reach out to families to help develop a health care plan for children and their families. These services are available to families and their children from birth to 22 years old. Special Child Health Services have existed for more than thirty years. The case managers provide resources, referrals, and emotional support for families in need of these service. • Every newborn or child diagnosed with a disorder in the NJ NBS panel, gets reported into the Birth Defects registry and becomes eligible for case management services. Dr. Post asked "How quickly are these infants registered and then referred to case management?" As per Dawn, the registrations are processed quickly and once the BDAR gets the diagnosis, a case gets released to case management. Following the release of the child's record, case management has 14 days to assign the child to a case manager. After this assignment, the case manager must reach out to the family within 7 days to initiate contact. As per Dawn, in 2021, there were 17,000 plus children that were referred to case management for services. Dr. Post asked for all Chairs to reach out to Ms. Seminara if they would like the Case management program to present at their meetings for next Spring. • As Per Ms. Canuso, physicians on the call should also feel free to utilize the Case Management services. This includes informing parents about the case management program and providing any feedback they might have.	
Patient examples at NSARC, subspeciality meetings: • Dr. Post asked the panel if it would be worthwhile to request one or two case presentations at each NSARC meeting? Several opinions within the group agreed that it would be beneficial.	
International Consortia Newborn Genomic Sequencing: • The International Consortia Newborn Genomic Sequencing is an international effort across multiple countries across the world. They are large research funded consortiums within the US, with locations in San Diego, Boston, and North Carolina. Internationally, England, Australia, and several European countries have created a website and consortium. Dr. Westover attended the first two organizational meetings that have looked at the value of all ethical, legal, social, and economic aspects of performing genomic sequencing on all newborns.	

• There is data suggesting that genomic sequencing is valuable to newborns with potential genetic conditions. Genomic sequencing can look for diseases that are not present on the current RUSP. • As per Dr. Day-Salvatore, there are four factors to consider when talking about genomic sequencing. The first issue is informed consent and who will be responsible for obtaining that consent. The second factor to consider when doing sequencing like this is that full sequencing would provide many variants of unknown significance (VOUSs). Therefore, who will be evaluating these variants as pathogenic? Who will be available to treat and manage those patients? The third consideration is determining which disorders should be identified and reported. The fourth consideration is that biochemical testing will continue to be needed because there are always new genetic variants. Lastly, equity needs to be assured in this process. • Commercial companies may offer genomic sequencing as supplemental for NBS. As per Dr. Westover, there is a company already offering this testing which was published in JAMA this past summer. They were using it as a value add-on for families that have decided to do cord blood banking and offered it as an add-on cash only product. • Those interested in participating in an ad hoc Genetics committee to discuss implications of future genetic testing included but not limited to: Dr. Westover and Dr. Post as co-chairs – Sharon Anderson, Dr. Day-Salvatore, Dr. Weller, Dr. Schachter, and Dr. McCormack.	
Dr. Post announced that the Open portion of the session was completed. The guests were asked to disconnect from TEAMS and it was assured that they had done so.	

I. Adjournment

Meeting Adjourned By: Dr. Post **Time:** 12:03 pm

Minutes submitted by Program Support Specialist 3