Newborn Screening Advisory Review Committee

Meeting Minutes

Hybrid Meeting at Saint Peter's University Hospital and via TEAMS

Tuesday, May 7h, 2024 - 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* and the *Star Ledger*, on April 8th, 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
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
P	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
P	Shula Narang, 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
A	Nicole Pratt	Family representative
A	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
A	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**
P	Dr. Barbara Spitzer	Pediatric Hematology Oncology: Hackensack Meridian
P	Thomas Scanlin, MD	Rutgers RWJ Medical School
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 Seminara	Meeting Coordinator/NBS Support Program Specialist 1: NBS Follow up
P	Suzanne Canuso	Program Manager: NBS Follow-Up
P	Sarah Eroh	Quality Assurance Specialist: NBS Follow up
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
P	Kelly Anderson-Thomas	Deputy Commissioner
	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
A	Lauren Oplinger	
A	Didar Mohammed Yahia	Rutgers University, Full-time Genetic Counseling Student (2026)
P	Dana DiFilippo	Reporter, NJ Monitor

Dr. Post called the meeting to order at 9:04 am, Michelle Seminara announced the meeting was being recorded.

Approval of Tuesday, November 14th, 2023, 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	
1. **Dorian Minond** – Mr. Minond came as representation on behalf of families and patients with PDCD and addressed the group on the seriousness of the disorder. Mr. Minond gave background to the group explaining the complexities of the disease. PDCD known as pyruvate dehydrogenase complex deficiency is a genetic metabolic disorder affecting the breakdown of carbohydrates, which almost always results in increased lactic acid levels causing a wide range of symptoms. Patients with PDCD have symptoms that may include brain malformations, seizures, ataxia, hypotonia, neuropathy and serious developmental delays. Genetic testing is used as a definitive method of diagnosis, but families often cannot acquire genetic testing until far into their diagnostic journey. Research has shown that one of the best predictors of survival and cognitive outcomes is	

an early age diagnosis. A study out of Ohio has found that one in 42,000 live births are affected by PDCD. Mr. Minond also asked if there's any way for New Jersey to make existing deidentified data available for researchers to further validate those studies or carry that research forward. • Mr. Minond spoke about the 2024 study that addressed the specific amino acid ratio combinations that were found to be effective at discriminating PDCD from other metabolic conditions in newborn screening. • There is a parent advocacy group called the Hope for PDCD Foundation based out of California, which provides fundraisers and advocacy. They also fund research and gather data necessary to support the application at the federal level. Dr. Scanlin asked if PDCD has been added to California's newborn screening panel? As per Mr. Minond, this disorder has not been added to any state's newborn screening panel however, California is doing advocacy for children and their families. 2. **Lauren Oplinger** – Did not attend the meeting. 3. **Didar Mohammed Yahia –** Did not attend the meeting.	
Announcements	
1. Lab Updates • Miriam gave an overview on how the lab has been doing so far. There has been a huge improvement with timeliness of specimen transport, due to the new private courier system. The courier is called 24/7 Enterprises and travels along four different routes making pickups 6 days per week at all birthing hospitals within New Jersey. The couriers start their pickups around 8:00am in the morning, travel their routes, and deliver the specimens to the lab later in the evening. The hospitals enjoy that they have been able to build a relationship with the pick-up drivers and feel it has decreased the chances of lost packages. 24/7 Enterprises also picks-	

up specimens on weekends and holidays and delivers the specimens within a timely fashion to the laboratory. Since specimens arrive in the lab early in the morning the lab has been able to start working on the specimens sooner which helped to improve their time management.

- In January of 2023 before the private courier was implemented the lab found that 18% of specimens have been delivered to the lab more than three days from when the sample was collected. In April 2024, because of the private courier, the lab found that just below 3% of specimens have been delivered more than three days from when the sample was collected. The lab is pleased with how the courier has been benefiting everyone so far.
- This year NBS will be celebrating 60 years of newborn screening in New Jersey. New Jersey has gone from screening one disorder in 1964 to screening for 63 disorders in 2024.
- The lab will also be visiting all birthing hospitals in New Jersey in order to provide them with some additional educational resources.
- The lab has also been working on reporting out Congenital Adrenal Hyperplasia (CAH) on Saturdays. The lab is open Monday-Saturday and on Saturday's time critical disorders are reported out. The assay to screen for CAH is long to run, therefore, the lab has been working on ways to figure out the best way to report CAH on Saturdays. The lab can do the first review of results late Friday afternoon, and the timing on this is still ongoing. The lab is hopeful to get this implemented at some point later in the year.
- The lab will be meeting with the couriers later this week to discuss how they can also schedule pick-ups at birthing centers. They need to discuss how to fit the birthing centers and midwives into their current routes. Currently, birthing centers use FedEx to

send specimens to the lab, if the birthing centers do not have a FedEx account one can be set up for them to use.	
2. Follow up Updates • The follow up program has been awarded a HRSA Propel grant for newborn screening. The lab is working on improving interoperability and working on having their Lab Information Management System electronically linked with the hospitals using HL 7 messaging. This will increase efficiencies, timeliness, and decrease manual errors. The follow up program will be using this grant to establish a long-term follow up component. There is a pilot they hope to set up in the beginning of June. The follow up team will collaborate with Case Management services. Once a baby is registered in the New Jerseys birth defects registry, the baby is then referred to case management to make families aware of their services. • As per Suzanne, the pilot is set up to gather information within three counties, Gloucester, Mercer, and Hunterdon. The follow up program will be using REDCap to survey the families and collect data on their care. The follow-up program is still refining their workflow, and the group discussed different data collection techniques. • As per Dr. Day-Salvatore, if follow up is interested in doing longitudinal studies it will take them a long time to collect all the data. Therefore, Dr. Day-Salvatore suggested a retrospective approach where the follow up program may need to go back three years to see what has been happening within the three-year time period. Suzanne discussed consent and how to approach reaching back to those families from three years ago. Dr. Day-Salvatore suggested that since early intervention starts within the first three years of a child's life, this might be an ideal time to reach out to	

those families and understand how their experiences have been evolving. • According to Dr. Westover, the long-term follow-up pilot may have two advantages: a retrospective study provides a more thorough gap analysis, and a prospective cohort helps determine how best to proceed and what must be improved. • Follow up will also be looking into contacting families through emails and cell phones for an easier and less formal approach. Follow up is interested in reaching out to families annually, which will help build an understanding of their experience in care. REDCap and/or an automated system will be used to check in with families. In addition, follow-up is trying to improve the survey's usability and make it simpler for families to complete.	

IV.

	Actions/Resolved
Subcommittee Reports	
Cystic Fibrosis • Dr. Scanlin spoke about the cystic fibrosis subcommittee and what was discussed during that meeting. New Jersey state lab received a grant to help them screen for multi-mutations related to cystic fibrosis (CF). Due to the grant awarded, as of July 2022, the lab went from screening one mutation to screening for 139 mutations in CF. The group also discussed the possibility of applying for a new grant to help switch to a floating cut off as recommended by the Cystic Fibrosis Foundation. • Dr. Scanlin referred to the New England Journal and two articles based on using CRISPR-Cas9 gene editing in sickle cell and the impressive results on this form of treatment. The median survival rate in 1969 of someone with CF was ten years of age. Now with the	

new technology and modern medicine the survival of someone with CF in 2024 is now, 56-59 years of age. The availability of new medications and therapies has led to a rise in the number of pregnancies and births among women with CF.

Endocrinology

- Dr. Post addressed the committee to give his update on the Spring Endocrinology meeting. The group discussed the weekend holiday coverage issue. The group also addressed when to appropriately retake specimen samples while the baby is receiving care within the NICU, it was decided that the final specimen should be taken at 28 days instead of 42 days. As per Miriam this change has not gone into effect because they need to update it in the Regs. The Regs are due to expire February 2025 and the submission process has already been started. The group also discussed reducing the number of NICU notifications for the two endocrine disorders that have huge numbers of false positives and had a brief discussion about where things are going with adrenal leukodystrophy.

Metabolic and Genetic

- Dr. Day-Salvatore addressed the group and spoke about Guanidinoacetate Methyltransferase (GAMT) Deficiency, being added to the Recommended Uniformed Screening Panel (RUSP). The lab is waiting for GAMT and MPS2 to be added onto the NEO LSC kits and once that is completed the lab will need to do another mass spec to pursue screening.
- Adjusting cut offs for different disorders was also discussed. The lab will reevaluate the cutoffs for late onset Pompe disease.
- The group discussed the NBS follow up form and how to properly categorize variants and classics and some cases variants of uncertain significance verse pseudodeficiency and is still an ongoing topic they will continue to discuss in the fall.

• The group discussed other screening methodologies, Niemann Pick and the ad hoc subcommittee Universal Newborn Genomic screening and sequencing. • The subcommittee discussed reporting on state holidays and a list of anticipated disorders was reviewed. **CCHD** • Data will be presented at this meeting to provide further insight on CCHD. **EHDI** • Dr. McInerney is the chair of the Hearing Evaluation Council and introduced herself to the group. In April, the Early Hearing Detection and Intervention (EHDI) committee held a meeting. Audiologists follow the Joint Committee on Infant Hearing (JCIH) guidelines. Currently, the 1-3-6 guidelines are required which means that babies get screened by one month of age, identified with hearing loss by three months of age, and begin interventions by 6 months of age. Nationally there is a push for the algorithm 1-2-3 guidelines which would implement screening by one month of age, identifying by two months of age, and begin intervention by three months of age. The group also spoke about better ways to follow up with families, and how to improve monitoring children with risk factors. **Immunology (SCID)** • Dr. Spitzer spoke to the group to discuss what happened during the SCID meeting. The lab has been validating ADA SCID, which helps doctors to start treatment earlier by identifying a specific subtype. **Hematology** • Dr. Narang addressed the group to give an overview of the Spring meeting. Methods of screening for hemoglobinopathy were discussed, solubility versus hemoglobin electrophoresis. It was agreed by the committee to include a clear warning to the pediatricians in the results section that electrophoresis should be preformed and should not be verified by a solubility test.	

• A sickle cell data collection grant was awarded through the Centers for Disease Control and Prevention (CDC) and is projected to be a five-year endeavor. It was decided to include more links to websites for extra resources in the letters that the follow-up program sends to families. The hematology group is also interested in updating the school guide and the family guide to make them easier to use and more accessible to families.	
Old Business	
1. CSTE Council of State and Territorial Epidemiologist cCMV statement **a) NJ DOH update on cCMV work** • The Council of State CSTE has provided a standard definition of congenital cCMV and a position statement was approved and developed in September. • Kathy shared her slides to give an update on cCMV. There are two grants in which the department of health is being funded. One of the grants is going towards Surveillance for Emerging Threats to Pregnant People and Infant Network (SET-NET). New Jersey began work in August 2022 on the SET-NET pilot project to collect information on cCMV cases. The data collected is to understand the effects of emerging and reemerging threats on pregnant people and infants. • The second grant was used towards Pregnant People-Infant Linked Longitudinal Surveillance (PPILLS). PPILLS aims to gain longitudinal data to ensure timely reporting of key exposure and outcomes that impact pregnant people and their babies. PPILLS also	

tries to improve data quality, sharing data, to innovate clinical strategies and to build a strong collaborative network. • SCHS identifies possible cCMV cases from, the NJ Birth Defects registry, birth certificate system, and hospital billing. From 2018-2022 there were 132 pregnancies as potential cCMV cases. Research studies report 1 in 200 children are infected and 1 in 5 of those have permanent disabilities. Over 4 years of data collection only 45 cases have been diagnosed as having cCMV which is substantially fewer than what they expected. • PPILLS goal is to reach out to cCMV impacted families through age 6 to collect data on their experience and to also provide additional support. • In January the EHDI program met with the Office of Communication and the National CMV Foundation to exchange thoughts and ideas about public awareness. Some funding will be made available to the Office of Communication so they can continue to work on CMV awareness. To seek some additional guidance on how to do more public awareness the EHDI program will reach out to other state programs, such as Minnesota and Utah. • The EHDI program also surveyed the maternal child health directors at hospitals about their existing CMV programs and discovered that there are currently 7 hospitals that are doing CMV screening on babies that fail NBS hearing screening. Since then, the EHDI program will collaborate with those 7 hospitals to look into their policies to develop a best practice document. • Dr. Westover asked, "What percentage of the cases that were ascertained in the immediate post-natal period were also ascertained prenatally?" As per Kathy, there were a few cases that were confirmed with cCMV prenatally from amniocentesis, some infants were identified as a IUGR and later on testing for CMV was done.	

• Fetal death certificates were also investigated to see if CMV was the cause.	
2. Universal Newborn Genomic Sequencing Ad Hoc Subcommittee • Dr. Westover addressed the group to give an overview of what has been discussed at the ad hoc committee. Some factors have been identified as important considerations when deciding upon the implementation of whole genomic newborn sequencing. Those factors include, ethical, legal, and social implications, gene selection and analysis, validity and utility, and parental update and consent. A final report from the Universal Newborn Genomic Sequencing Ad Hoc Subcommittee will be presented to the committee before the next Fall NSARC meeting. Various topics of discussion will then be decided upon at the next NSARC meeting. • Whole genome sequencing (WGS) pilot studies are being done internationally. There are a lot of important issues we need to address before bringing WGS to New Jersey. • Currently, if families are considering supplemental screening for their child, they have the option to seek out commercially available labs that specialize in whole genomic sequencing.	
3. Review of updated Bylaws • As per Dr. Post, the process of approving edits to the Bylaws has changed. Legal staff at the department of health have now delegated the revision of the Bylaws to be reviewed and revised by NSARC. As per Dr. Post, as we see fit NSARC will propose amendments to the Bylaws.	
New Business	
1. NBS 60th Anniversary; in-service blitz and 9/19 event: • The lab and follow up plan to visit all 45 birthing hospitals within the state of New Jersey. So far, the they have visited 15 hospitals and plan to visit the other 45 by the end of August. The New Jersey newborn	

screening program will be celebrating 60 years of newborn screening and developed a new mantra they will be introducing to hospitals, called “No blank spot”. The “N” stands for Newborns are all tested, “B” refers to boxes are filled in legibly, and the “S” represents specimens that are satisfactory. For satisfactory testing the lab needs ten punches out of one dry blood spot, and this is important for accurate testing. The lab is also distributing a “no blank spot” badge buddy, that is the size of a business card. The front gives a step-by-step on how to properly take a sample. On the back a QR code is provided to give greater detail on how to take the sample and address any inquiries. The lab is tracking the hospitals process by implementing surveys to address the following questions, for example, “How many UNSAT specimens?”, “How many kits were missing key demographic information?”, “How many samples arrive at the lab three days or later after collection?”, etc. • September will be newborn screening awareness month, and the lab will be hosting an event. The event will be on September 19th, to include tours of the lab, and a panel of parents who may speak to express their experience with NBS overall. There will also be fun activities with food to follow. A save the date will be sent later and distributed to the NSARC committee.	
Dr. Post 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:** 11:52 pm
Minutes submitted by Program Support Specialist 1