Newborn Screening Advisory Review Committee

Meeting Minutes

Virtual Meeting Conducted via TEAMSc / Tuesday, May 17th, 2022

9:30AM-12:50PM

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 11, 2022 and the *Star Ledger*, May 11, 2022. 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.

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
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
E	Steven Diamond, MD	Pediatric Hematologist: Hackensack University Medical Center Chair of Hematology Subcommittee
A	Patrick Hill, PhD	Ethicist: Rutgers
	(vacant)	March of Dimes Representative
P	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
	(vacant)	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
E	Alan Weller, MD, PhD	Pediatrician: Rutgers, The State University of NJ
P	Thomas Westover, MD	Obstetrician-Gynecologist: 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	Chair of Immunology Advisory Committee: Allergist-Immunologist CHOP
A	Al Gillio, MD	Chair of Immunology Advisory Committee: Director, Children's Cancer Institute
A	Robert Zanni, MD	Pulmonologist, Monmouth Medical Center Chair: Pulmonology Specialty Group
Absent/ Excused/ Present	**Guests/Name**	**Affiliation**
P	Jill S. Menell, MD	Chief/Pediatric Hematology/Oncology: The Valerie Fund Children's Center for Pediatric Cancers and Blood Disorders
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	Donna McCourt	Microbiologist 4/Quality Assurance: NBS Lab
P	Karyn Dynak	NBS Follow-up Program Coordinator
P	Hui Xing	Research Scientist
A	Diane Driver	Nurse Consultant
P	Michelle Seminara	NBS Public Health Representative 3
A	Stephanie Agugliaro	NBS Public Health Representative 3
P	Suzanne Canuso	Public Health Consultant 1: Nursing, Interim Program Manager for NBS Follow-Up
A	Caitlin Russo	Research Scientist 3: NBS Lab

P	Sarah Eroh	Quality Assurance Specialist
A	Joy Rende, MSA, RNC-E-MNN, NE-BC	Nurse Consultant 1
P	Victoria Floriani	Research Scientist 3
P	Jen Hopkins, MPH, CHES	Program Specialist 1
	Public Attendees	**Affiliation**
P	Miranda McAuliffe	ALD Alliance: Parent
A	Elisa Seegar	Founder of ALD Alliance: Parent
P	Jesse Torrey	ALD Alliance: Parent
P	Taylor Kane	Patient Advocate and Carrier of ALD
A	Christen Jolicoeur	
A	Amanda Schneider	CAMcare
P	Mohamed A. Fouad	Parent
A	Michael E. Zwick, PhD	Human Geneticist: Rutgers Senior Vice President for Research
P	Diane Kane	President of Run for ALD, child is a carrier

Dr. Post called the meeting to order at 9:10 am

Approval of November 16, 2021, Minutes: Approved

Introduction of Open Public Meetings Act -	**Actions/Resolved**
• Suzanne explained that NSARC is now subject to follow the Open Public Meetings Act (OPMA). The purpose of OPMA is to give the public a forum to be able to attend and for their voices to be heard. Suzanne and the Chair, Ernie, welcomed the Public memebers to the meeting. The Chair also expressed his preference of the informality of using first names whenever possible.	
Introductions of Members and Guests: Attendees introduced themselves and their affiliations.	
Lab Update regarding X-ALD	

• Miriam shared that the Newborn Screening lab is activetly working towards the implementation of ALD screening. There are three major steps that need to be accomplished before screening can happen. Step 1, the laboratory needs to validate two tiers of laboratory tests. Step 2, the new disorder needs to be intergrated into the lab information management system (LIMS). Lastly, step 3 is for the follow up group to ensure babies detected by the testing will be followed up in the appropriate fashion. • The 2 tiered screening method is in line with what other states are doing. Validation of the 1st tier is completed, 2nd tier is in the process of being validated. Any baby that screens positive on that 1st tier will be screened on a 2nd tier. The lab needed to postpone the installation and validation of that instrument due to COVID. The lab has hired a dedicated staff member to work on this project. Plan to start screening Q4 of 2022. • As per Ernie a meeting is scheduled for June 8th which will include genetics, neurology, and endocrinology, to discuss the process of following up on positive ALD newborn screens.	

III. <u>Public Comment on Agenda Items Only:</u>

Motion: To open public comment on agenda items

Call to Order by Ernie Time: 9:55

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 presiding officer. Each comment shall be limited to three (3) minutes. No participant may speak more than once. No diaglogue 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 (MicrosoftTeamsMeeting). No written questions will be read from this section. Patricipants must have audio and video capability to present comments and questions during public comment. Patricipants will be

called upon to present in the order their names are received. Although NSARC encourages public comment, the Chair of the Newborn Screening Advisory Review Committee can 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 appropiate.

Speakers: 1) Jesse Torrey (Parent of ALD Alliance) – Jesse's testimonial included the story of her son who is diagnosed with ALD. She expressed the importantance of adding ALD to NBS by discussing how expensive it is for the State to care for children with a late diagnosis of ALD; the costs cross healthcare, education/busing, all areas of life. As per Jesse a rough estimate for all of her sons services is $130,000 a year. Since her son went to special schools for 13 years the cost to educate her son is over $1.6 million. Now her son receives services through DDD, which pays for his adult program and a caregiver who comes two days a week for a couple of hours. This budgets about $80,000 a year. So far the state has paid an additional $160,000. Jesse expects that her son's services for the next 50 years would cost around $4 million determined by costs remaining the same to where they are now. 2) Elisa Seeger (ALD Founder) – did not attend the meeting 3) Miranda McAuliffe (ALD Alliance) – Mother of son diagnosed with ALD that is so far asymptomatic. Other relatives at risk for ALD have also been identified. She is part of the ALD Alliance. Miranda discussed the importance of ALD being added to the RUSP. Miranda also provided the	

group with information about the ALD Alliance including a QR Code as well as a Care Package Program for ALD Newborn Screen Families. 4) Taylor Kane (Patient Advocate and Carrier of ALD) – Taylor is a carrier for ALD and discussed the importance to adding ALD to NBS. Her father died from ALD. She described her dismay at the long time it has taken from the passage of the ALD NBS law until its implementation. 5) Mohamed A. Fouad (Parent) – Is a parent of a child diagnosed with G6PD and spoke about the importance of adding G6PD to the NBS pannel and has asked NSARC to considering adding this disorder to the NJ panel.	

Dr. Post thanked the public speakers for their comments.

IV.

Old Business	Actions/Resolved
Subcommittee Reports	
Cystic Fibrosis Meeting – No one present to discuss; however, Ernie stated their current topic is refining the screen as is currently being evaluated with the laboratory. **Endocrinology Meeting –** Ernie informed the group that there were two main topics discussed at the last Endo meeting. 1) Whether Congenital Hypothyroidism should switch from a first tier T4 to a first tier TSH test? The reason for this change is due to the increase in the number of false positive tests. Perhaps changing the cut off might be a solution however, at this time there has been no definite answer. 2) The upcoming XALD Screen - at least two of the pediatric endos on the subcommittee are going to be involved in the discussion next month. Christiana presented a question, in which she was courious if the preemie population might be contributing to a majority of the false positives or if it was throughout all different gestational ages? Ernie responded that the largest number of false positives is the premature babies. The advantages of switching to TSH is false positives will be reduced. Disadvantages still occur but overall the TSH test will cut down on the NICU workload and help families overall with care. Christiana also brought up genomic newborn screening. As per Ernie in relation to insurance there is at least a state or federal law stating discrimination based on any genetic test results is illegal. Christiana also brought up genomic newborn screening. As per Ernie in relation to insurance, there is at least a state or federal law stating discrimination based on any genetic test results is illegal. • The Genetic Information Nondiscrimination Act (GINA) a. Passed in 2008, protects Americans from discrimination based on their genetic information in both health insurance (Title I) and employment (Title II)	

It is also in question whether or not to screen for disorders that are not treatable? Debra explained to the group that NBS is designed to screen for disorders that would be treated in infancy or childhood however, some diseases that could potentially be detected in NBS might not present until adulthood. NBS can sometimes detect the possibility of disease without knowing the expected age of onset of symptoms. If genetic testing goes forward, educating families would be the next hurdle to overcome. **Hematology Meeting –** Jill informed the group that after reviewing the data there has been an increase of patients who are lost to follow up in the last few years. The group has not determined the exact cause however, it might be related to families moving or the COVID pandemic. The Follow Up letter was updated to remind physicians to use hemoglobin electrophoresis to confirm diagnosis, due to two physicians using an incorrect test to clear infants of disease, but it is also recommended to consult a hematologist to help provide comprehensive care. Lastly, the hematology group spoke about the abnormal hemoglobins that are not clinically relevant, and discussed possibly discontinuing to report those because it does not have any significance for the child's furture and many times these are fetal hemoglobin abnormailities that go away by six month of age. **Metabolic and Genetic Subcommittee –** Debra, expressed that the last meeting was very lengthy. Follow up on SMA was discussed and also the need to review all of the positive lysosomal cases to date. They are trying to facilitate genotype/phenotype correlation in those cases. Also discussed was MPSII where Debra stated she submitted a letter on behalf of the subcommittee in support of the implementation of MPSII. Adjusting some cutoffs was discussed for some of the analytes, specifically CIT. There was concern regarding the responsiveness from the PCPs and timeliness of referrals for some disorders so there is a plan to work with the state follow	

up program to help ensure everything runs smoothly and in a timely fashion. Metabolic and complex disorders need to be cleared only by the specialist. **CCHD** –Critical Congenital Heart Disease – There is no subcommittee monitoring and supporting CCHD NBS yet. **EHDI** – Maryrose spoke to the group about the Early Hearing Detection Intervention program (EDHI). This program is also advised by the Hearing Evaluation Council, which meets 3 times a year; last meeting was in February 2022 and will meet again in June 2022. As per Maryrose they abide to a 1,3,6 algorithm that came out in 2007 and is guided by the Joint Committee of Infant Hearing (JCIH). There was an initiative for the New Jersey Head Start program and a pilot was created involving Montclair University, EDHI and Head Start, which went out to 5 different Head Start programs and screened those children. EDHI then sent out a needs assessment survey to physicians, pediatricians, pathologist, and early intervention individuals to look at the need for the EDHI program and suggested areas where education is needed. Lastly, the committee spoke about New Jersey regulations which are up for renewal. Overall, their goal is to revise the New Jersey regulations and follow the guidelines of JCIH 2019. JCIH 2019 came out in 2020 and recommend an algorithm of 1,2,3 but most states have not abided by those guidelines yet. The 1,2,3 guideline screening is done by one month, diagnosis completed by two months and treatment is to begin by three months of age. High risk babies such as CMV cases and the type of screening needed was also reviewed. **Immunology Meeting** – No one present to discuss	

New Business	
Overall Incidence and Program to Date Cases– Total cases were discussed dating back from 1964. Screened Positive by county from 2020-2021; Essex County has highest number of referrals in past two years, more than 200+ cases/year, which may be due to it being a high birth rate region. Hui presented program to date data and discussed with the group.	
Request to add to the NJ newborn screening panel –	
As per Suzanne NSARC was formally established by legislation a year and a half ago and the group is looking to amend the bylaws to include a formal review process for adding new disorders to the NJ NBS panel. Requests for a disorder to be added to the NBS panel can be submitted to the Commissioner of Health, the request would then be given to NSARC for review. A draft of proposed bylaws revisions was sent to the group and for their review and if approved, the draft will be sent to the DoH legal counsel and the Commissioner of Health for approval. Ernie states that as more treatments for diseases are becoming available mostly through gene therapy, NBS is on the verge of an explosion of adding new conditions. Debra mentioned considering genomic sequencing for the future, however, genomic sequencing presents challenges including requiring written informed consent. Tom W. discussed possibly correlating NBS with Prenatal Screening.	
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.	

IV. Adjournment

Meeting Adjourned: Time**: 12:39**

Minutes submitted by Michelle Seminara