Genetic Diseases Program Advisory Committee Meeting

February 4, 2022, 2:00 p.m. – 3:30 p.m.

Minutes

Attendees:

Name:	Title:	Organization
Adrian Savant, MD	Pediatric Pulmonologist	Children's Hospital/Tulane University
Rachelle Boudreaux	Boards and Commissions Coordinator	Bureau of Family Health
Pooja Rao, MD	Neurologist	Ochsner
Amanda Perry	Interim Clinical Systems Team Lead	Bureau of Family Health
Michelle Duplantier	Statewide Social Work Consultant	Bureau of Family Health
Sharonda Smith	Admin Coordinator w/CSPT	Bureau of Family Health
Tiffany Allemand	Parent Liaison	
Nora McCarstle	Pediatric Statewide Nurse Consultant	Bureau of Family Health
Ngoc Huynh	Epidemiologist	BFH Genetics Diseases Program
Maynard Freisz	Cure SMA	National Organization – Spinal Muscular
Jantz Malbrue	NBS Follow-up Coordinator	BFH Genetics Diseases Program
Rainbow	Filling in for Pediatric NBS Nurse (Bonnie)	Children's Hospital N.O.
Randy Leggett	Quality Improvement	Bureau of Family Health
Paul Roesch	Sr. Director of Newborn Screening	Baebies Inc.
Cynthia Suire, DNP	Consultant	Bureau of Family Health
Cara McCarthy	Reproductive Health Program	Bureau of Family Health
Meredith Allain, MD	Medical Geneticist	Ochsner
Mary Schroth, MD	Medical Officer	Cure SMA
Tyler Craddock	Government Affairs	Bluebird Bio
Charles Ugokwe, MD	Pediatric Neurologist	Practice in Alexandria, LA
Beverly Ogden, MD	Medical Director	OPH Lab
Joe Bocchini, MD	Pediatric Infectious Disease	Tulane University

I. Welcome & Introductions
Cheryl Harris welcomed attendees. Attendees introduced themselves and stated the organization they represented.

II. Review of the Committee Minutes from previous meeting
A copy of the minutes sent out prior to the meeting for review along with a copy of the agenda.

III. Status of Adding New Conditions and Rulemaking – Amanda Perry
Timeline in update with new conditions that are being screened; SMA, MPS1 Pompe implementation. Published back in December with comment period January 25th and no comments received. Rulemaking goes into effect March of this year (next month).

- SMA, MPS1 and Pompe: We are sending those samples collected in the lab in Louisiana and normal screening panel is done in Louisiana.
    - Shipped out daily

- OPHstate lab here in Louisiana is finalizing some of the calibrations and onboarding new scientists
- All of the retrospective cards for the Newborn Screening (NBS) as of January 1st have been screened or tested.
- Every child is being screened and we anticipate SMA will be tested in house in March (next month). We anticipate MPS1 and Pompe will be tested in house around May.

IV. Laboratory Updates – Meredith Ogden

- Need to keep up with Rulemaking.
- When we say, we need to do new testing, the lab has new needs; equipment, people, etc.
- SMA testing is screened parallel with Severe Combined Immunodefiency. All in house and it's a Perkin Elmer method used.
- For MPS1 and Pompe, we brought in new technology and hired new staff
- In the meantime, we made an arrangement with Florida State Lab to pick up our test.
- Everything will be in house as soon as we get our staff and up to date.
- As of January 1, 2022, any baby born January 1st moving forward is being tested for those three conditions.
- Questions/Responses:
  - Question: With the testing being done now, is it including both the SMA1 and SMA2 gene? For whenever planning to do in house, will that also be the case? Will that common point mutation be screened as well?
    - Response: Our SMA is actually being performed at Perkin Elmer at this time, so they are getting tested even though we don't have SMA in house. Already contracted with Perkin Elmer.
  - Question via chat: Will NBS show carrier status for SMA?
    - Response: We won't be able to tell you the count of the genes on the initial part, but we will be able to use some follow up testing and if abnormal, we will be able to tell you.
  - Question: How many babies tested positive since these three conditions were added?
    - Response: Since we've added these three conditions, we've had two that have been screened positively for MPS1. We had others from Perkin Elmer we've received and we had about seven babies through their laboratory received for follow up. As for SMA, we haven't received any up to this point, so far for those tested since January. MPS1 have been referred to the Tulane Metabolic Center for follow up.
  - Question: Previously talked/emailed about possibly NBS are positive for MPS1 that are Ochsner patients; is this something we need to coordinate with Dr. Anderrson to be done as outpatient? After their results initially called out? Or is this something we would do through the state?
    - Response: The program can set up patients that come from your system and they can refer them back. Would help if they would put pediatricians on the slip. They receive about two or three per week, because initially they were batched to send to Florida. We may see a little more in our first couple weeks, then we still anticipate for MPS1 maybe two to three a week and one or two a month for Pompe presumptive.
  - Question: Point mutation testing that is beyond the scope of current SMA NBS. Does suspicion of point mutation require gene sequencing?
    - Response: Typically not a reflex test. Usually is one SMA copy and typically NBS you wouldn't do gene sequencing unless the baby became

symptomatic and you changed your protocols for how you manage one copy of SMA1. We haven't had enough samples to really look at any issues with follow up or further testing, but as we progress I'm sure those issues will come along.

- Question: Curious what the follow up was for SMA babies with the positive screen? What happens after that?
    - Response: Two babies as Women's; get the baby in and draw blood then send off for sequencing.
- Rephrased question: If state gets a positive NBS for SMA, what do they do at that point?
    - Response: When we receive a positive infant for any presumptive positive condition, we contact the child's primary care provider or if baby is still in the NICU, we contact the NICU. Then we make recommendations based on the ACT sheet and particularly for SMA it is referred to a neurologist. Only had a few that were in the Baton Rouge area and referred to Dr. Holman. We are in the process of creating a regional list of primary care providers; neurologist across the state who have expressed interest in treating infants with SMA and will be referred to a local provider. Spoke with Dr. Holman and she anticipates only six cases a year in our state and she is happy to handle those cases. With any presumptive positive case, we'd notify the primary care provider and we'd ask them to refer the infant to a specialist with whom we've had some kind of contact with. Also let them know that a physician's office would be contacting them about a referral. That occurs with any infant with a positive screen.
- Question via chat: Are there specific child neuromuscular specialist that pediatricians can be provided with?
    - Response: Yes, currently working on a sheet; list of providers that we'll publish and fax to the primary care providers when we get a positive case.

Last year there were four conditions we were adding to the NBS panel along with MPS1, Pompe and SMA, there is also XALD. Tyler and his crew at Bluebird gave fantastic presentation on XALD.

Laboratory Updates – Alyson Saadi and Dr. Richard Tulley from our OPH Laboratory

- XALD is on agenda for next year, we have to get instrumentation and that will be a pretty large validation; has about 60-65 Nano lights in it. It's the next piece on the agenda. Per Dr. Tulley, the Association of Public Health Laboratories has fellowships they are offering and they are going to apply with primary emphases on XALD validation.

V. Other Business

- Rare Diseases Advisory Committee – Jantz Malbrue
    - The Rare Diseases Advisory Council has continued to meet and the council itself is charged with providing advice and giving a platform to rare disease and the community. Trying to raise awareness through diverse members, community support partners, and stakeholders.
    - Their charge is mainly identifying the areas and also looking at solutions.
- Newborn Screening Scorecard – Ngoc Huynh
    - This is the first time developed/produced product to send out to the hospital. Also for monitoring and tracking performance at the hospital.

    - The NBS Scorecard is great information for Newborn Hearing screening, new stick screening and breastfeeding.
    - Each hospital will see how they are doing and where they are with the state.
    - Also compare to national level.
    - This was done yearly, but some of the next steps is to try to ensure that we send this out at least quarterly or twice a year so that hospitals can have for their own internal purposes. They have been getting very positive feedback from the hospitals inquiring about better collection techniques.
- Question: Would the state consider a metric regarding time to treatment from diagnostic to time to treatment in the future?
    - Response: Something the program will definitely look at and if they do have metrics or information from the APHL website, we can share with this group.
- ACHDNC: Review of nominated conditions. Looking for conversations regarding the following:
    - MPS II
    - Shortages in newborn screening staff
- Two important things on the agenda are the ACHDNC, will be
    - Voting on whether or not to implement MPS1. If they vote to include it on the recommended uniform screening panel, that's another condition we'd have to assess to determine if that is something we want to start testing for in Louisiana.
    - Discussion around JMT and adding that to the NBS panel.

VI. Upcoming Meeting Dates for 2022 are listed.

- April 29, 2022
- July 29, 2022
- October 21, 2022

Note: Before announcements, please complete the post- meeting survey. Link added to the chat.

- Question via chat: Concerning Rare Disease Advisory Board - Whether we are looking at other conditions. Those mutations, but not actual conditions?
    - Response: Yes, we are looking at those. We are focusing not only on the state NBS panel and the universal NBS panel, but also looking at other conditions outside of that. We can have more of a broader rare disease council which is why we're reaching out to various patient organizations, other specialist around the state and make sure we are encompassing other types of diseases and mutations that may not necessarily be recognized or identified through NBS.

VII. Adjournment