State Advisory Council on Hereditary and Congenital Disorders

Minutes April 23, 2019

Members Present
John McGing, Chair
Hilary Vernon (phone)
Rebecca Furman (phone)
Erin Strovel (phone)
Ben Smith (phone)
Anne Eder (phone)
Michelle Smith (phone)
Sarah Viall (phone)

Members Absent
Delegate Karen Young
Senator Ronald Young
David Myles

MDH Staff
Stacy Taylor
Adam Coleman

Ex-Officio Present
Fizza Majid
Johnna Watson (scribe)
Robert Myers

Guests
Paul Vetter, (phone)
Ann Moser
Shantia Fitzgerald
Anna Torrey
John Gibson (phone)

Called to Order – 5:20 pm (delay due to IT technicality)

I. Welcome and Introductions

Members and guests introduced themselves. Shantia Fitzgerald is attending as a potential Council member. Anna Torrey is attending as a family member of an affected X-ALD patient.

II. Approval of Minutes

Minutes from meeting on October 9, 2018 were approved and will be posted on website.

III. New & Old Business

Status of addition of new disorders

- Dr. Myers reports there has been a delay in implementation of the lysosomal storage disorders and spinal muscular atrophy (SMA) secondary to staffing status. Staff has now been hired to perform lysosomal storage disorders. Approval to hire contractual employee for SMA was approved and then rescinded. A PIN position has been allocated and anticipate hiring by end of June. Implementation for Fabry, MPS-I, Pompe and SMA is now forecasted to begin June 30, 2019.

Presentation – SCID Screening

- Adam Coleman gave a presentation on the first two years of screening for severe combined immunodeficiency disorders (SCID).
- Implementation started in April 2016
- Background given of SCID, along with discussion of screening method and cut-off analysis
- Five infants in the first two years of screening have been identified as true cases. Four infants had bone marrow transplants or gene therapy and are currently thriving. The fifth infant was critically ill from another condition and did not survive to receive bone marrow transplant.

Presentation – CCHD and NBS Screening

- Johnna Watson gave a presentation on the first 6 years of screening for critical congenital heart disease (CCHD) and a summary of the conditions identified through newborn metabolic screening between 2010 and 2018.
- Review of newborn metabolic screening cases between 2010 and 2018 shows about 17,000 cases have been followed. Of these, about 2/3 of the cases have been resolved with a normal repeat newborn screening specimen.
- Disorders were broken down into the Core and Secondary conditions identified on the federal Recommended Universal Screening Panel (RUSP). In summary, 18 infants with time critical organic acid disorders, 43 infants with time critical fatty acid oxidation disorders and 15 infants with time critical amino acid disorders have been identified. The most common disorder is sickle cell, and the second most common disorder is congenital hypothyroidism.
- Review of CCHD Screening was provided, indicating how screening is performed and the target conditions.
- Screening rates throughout the Maryland birth hospitals has been fairly consistent averaging about 91% of babies having documentation of screening in the database.
- Screening began September 1, 2012 and 1 baby was identified as having a CCHD in the first 3 months of screening. In the subsequent 6 years of screening, an average of 6 babies per year are identified through pulse oximetry screening as having a CCHD. An average of 3 babies per year were either missed or had a negative screen that were later identified in the first few weeks of life as having a CCHD.
- Ben Smith asked if there are requirements for the pulse ox used in the screening process. Johnna states she will research this issue because Dr. Badawi was more involved with the implementation process. (Post-meeting addendum: Review of information surrounding implementation of CCHD screening reveals that the only stipulation regarding the equipment is the pulse oximeter has to be FDA approved for use in neonates. Specific equipment was not recommended by the State, other than the FDA approval.)

IV: Member Updates

- Laboratory Administration
  Additional update provided by Dr. Myers who indicates the lab has been testing multiple specimens for possible measles.

- MCHB
  Stacy Taylor from Office of Genetics and People with Special Health Care Needs reports that Michael Spencer has resigned as the Maternal and Child Health (MCH) Bureau Director. Maura Dwyer is currently Acting MCH Bureau Director. Tiereny Lloyd has been named the Deputy Director for MCH Bureau. Stacy also reports a bill to reinstate the Sickle Cell Steering Committee was passed this legislative session.

- Update regarding Federal Advisory Committee
  Sarah Viall reports that the Federal Advisory Committee, which is meeting today and tomorrow, is looking at developing an evidence review tool, changing how they will prioritize evidence. There are no new conditions being considered currently. The committee is also considering changing the screening process for homocysteinuria to homocysteine instead of solely methionine.

- Membership Update
  Johnna brought up the issue that Rebecca Furman is now working for MDH so may no longer be eligible to remain on the Council. Rebecca reports that she is working for DDA. Johnna will discuss this further with Rebecca directly after the meeting. Shantia Fitzgerald was invited as a possible Council member at the recommendation of Deputy Secretary Fran Phillips. Shantia

gave a brief description of herself, sharing she is a social worker who works with families and children services through Sickle Cell Disease Association.

- X-ALD Screening
  Ann Moser asked for projected start date for screening for X-linked adrenoleukodystrophy (X-ALD). Dr. Myers responded that the lab has a rough idea of how much the testing will cost. However, the impact that starting the 4 new tests will have on the budget is unknown at this time. After implementation of the 4 other screening tests is underway, the lab will have to perform a cost analysis to determine if they will have to ask for an increase in the newborn screening fee in order to purchase an additional mass spectrometer which will be needed to implement X-ALD screening.

- Krabbe Screening
  Ben Smith reports that 3 additional states have implemented screening for Krabbe Leukodystrophy. These states are Indiana, Michigan and South Carolina. He expressed concern that Maryland is falling behind the rest of the states. Johnna asked Hilary Vernon who has been the Council's researcher regarding screening and treatment for Krabbe if she knows if there is any new information currently available. Hilary stated she recently attended a meeting in which Krabbe was discussed, and there was nothing new presented in this area. John McGing stated that Ben's concern will be noted in the minutes.

V. Next Meeting Date:

- Next meeting is planned for October 22, 2019. This date was chosen since it will not conflict with any national genetics meetings. It should also be 4-6 months after implementation of the screening for the 3 lysosomal storage disorders and spinal muscular atrophy, which is the timeframe Fizza Majid stated she needs to be able to provide data regarding implementation.

VI. Adjournment

Meeting adjourned at 6:20 PM.

State Advisory Council on Hereditary and Congenital Disorders

Minutes October 22, 2019

Members Present
John McGing, Chair
Michelle Smith
Hilary Vernon (phone)
Delegate Karen Lewis-Young (phone)
Erin Strovel
Sarah Viall (phone)

MDH Staff
Jed Miller (phone)
Jennifer Taylor
Linda Lammeree, (scribe)
Monique Veney

Ex-Officio Present
Robert Myers
Fizza Majid
Johnna Watson

Members Absent
Ben Smith
Senator Ronald Young
David Myles
Rebecca Furman

Guests
Paul Vetter, (phone)
Ann Moser
Hannah Baer
Sarah Hash
Dan Shattuck
Dr Carol Greene
Jasmine Kretzer Noronha
Francis Rossignoc

Called to Order – 5:18 pm

I. Welcome and Introductions

Members and guests introduced themselves. Guests Sarah Hash and Hannah Baer are representing Coalition for Access to Prenatal Screening and will be presenting at the meeting this evening.

II. Approval of Minutes

Minutes from meeting on April 23, 2019 were approved and will be posted on website.

III. New & Old Business

Presentation: Coalition for Access to Prenatal Screening

- Sarah Hash, certified genetic counselor, and Hannah Baer, research associate, representing the Coalition for Access to Prenatal Screening presented evidence supporting the adoption of noninvasive prenatal screening and how policy changes can ensure all women have equal access to prenatal screening. The power point presentation was sent to Johnna Watson, Chief, NBS Follow up. The presenters shared the following key points:
    - Cell free DNA based noninvasive prenatal screening is increasingly utilized across all pregnancy risk groups. Since 2011, it has offered improved detection of fetal chromosomal abnormalities.
    - Cell free DNA based noninvasive prenatal screening is extensively studied in the general population (15+ studies with 88,000+ patients) and shows a very high positive predictive value compared to traditional screening with equal or better negative predictive value for all major aneuploidies.
    - Noninvasive prenatal screening provides better detection of Trisomy 21, 18, and 13. Its lower false positive rate, compared to traditional screening, leads to fewer invasive follow up procedures and procedure-related losses.

    - All major professional societies endorse or recognize cfDNA-based noninvasive prenatal screening as a clinically valid screening option for all pregnancies.
    - There is a clear disparity in access to noninvasive prenatal screening for many women enrolled in Maryland Medicaid. There should be a single standard of high quality care of all pregnant women.
- The Coalition is asking for the Council's support in the form of a letter of support to the State Medicaid program advocating for access to noninvasive prenatal screening for all pregnancies, not just high risk pregnancies.
- General discussion following the presentation indicated a letter of support could be seen as consistent with scope of Advisory Council. However, further information would be needed before a letter could be considered further, since an effective letter of support would need to address why access is not currently included and address those points, including fiscal, social, and political ramifications. The Coalition was invited to re-address the Council in the future with this information.
- Dr Majid provided an update on the new screenings added to the Maryland NBS panel as was requested at last Advisory Council meeting:
    - SMA (Spinal Muscular Atrophy) screening commenced May 30, 2019. A total of 31,900 infants have been screened for SMA. Dr. Majid discussed the status of presumptive positive screens and confirmed cases.
    - Screening for Pompe disease, Fabry disease and Mucopolysaccharidosis Type 1 (MPS 1) started June 17, 2019. Approximately 25,769 screenings for lysosomal storage disorders have been performed.
    - Dr Majid was pleased to announce that second tier testing for Cystic Fibrosis (CF) will begin in early 2020. Infants with elevations in the immunoreactive trypsinogen level on two screenings will then have DNA analysis performed for common CF mutations.
    - In addition, Dr Majid reported that second tier testing for Pompe disease (DNA sequencing) and MPS 1 (glycosaminoglycans assay) may also be available in 2020 as a send out to another lab. Second tier testing may help reduce the referrals to the genetics center.
    - As was discussed at last Advisory Council meeting, the lab has performed a cost analysis for X-linked Adrenoleukodystrophy (ALD) screening and is estimating it will cost between $10-15.00 per infant. This will cover the cost of additional instruments and staff for X-linked ALD screening. It will be necessary to request a fee increase for newborn screening which could add one year to the implementation time frame.
- There was discussion regarding confirmed cases of any of the lysosomal storage disorders.
    - Johnna Watson, Chief, NBS Follow up, reported approximately 24 infants referred to genetics centers for possible Fabry disease, 70 referrals for possible Pompe disease and 45 referrals for possible MPS 1.
    - So far, there are several cases of possible late onset Pompe disease and no confirmed cases of infantile onset Pompe disease. There are no confirmed cases of MPS 1 to date. Sarah Viall, CNMC Genetics reported that there has been one case of possible late onset Pompe disease in Washington, D.C., which started screening in 2017. Virginia started screening in Jan 2019 and has had one confirmed case of MPS1.

IV: Member Updates

- Membership Update
    - Currently there are two health unrelated vacancies in the Advisory Council membership. Additionally, it is likely that Senator Young may not be able to continue his position on the Advisory Council and so there will likely be a new legislative member vacancy.

- Johnna Watson reported that the statutory authority for the Federal Advisory Committee on Heritable Disorders in Newborns and Children expired as of September 30, 2019. Currently there is no standing committee or work group. There is more information on their website.

- Johnna Watson stated that an email has been received indicating the Attorney's General office has advised that members of the Advisory Council must be physically present at the meeting in order to vote. There was discussion about the impact this has on accomplishing even relatively uncomplicated tasks such as meeting minutes approval. Johnna also mentioned that a recent draft on attendance requirements at advisory committees permits attendance by phone. The Advisory Council would like more clarification on this issue.

V. Next Meeting Date:

Next meeting is planned for April 21, 2020 at 201 W Preston St.

VI. Adjournment

Meeting adjourned at 6:22 PM.