**State Advisory Council on Hereditary and Congenital Disorders**

**Minutes April 23, 2019**

**Members Present** **MDH Staff**

John McGing, Chair Stacy Taylor

Hilary Vernon (phone) Adam Coleman

Rebecca Furman (phone)

Erin Strovel (phone) **Ex-Officio Present**

Ben Smith (phone) Fizza Majid

Anne Eder (phone) Johnna Watson (scribe)

Michelle Smith (phone) Robert Myers

Sarah Viall (phone)

 **Guests**

**Members Absent** Paul Vetter, (phone)

Delegate Karen Young Ann Moser

Senator Ronald Young Shantia Fitzgerald

David Myles 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 Approval to hire contractual employee for SMA was approved and then rescinded.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.