

# State Advisory Council on Hereditary and Congenital Disorders

Minutes June 7, 2016

## Members Present

Anne Eder, Chair  
John McGing  
Ben Smith ( on phone)  
Neil Porter, MD  
Erin Strovel, PhD (on phone)  
Dr. Hilary Vernon  
Dr. Richard Bruno

## Staff

Linda Lammeree, RN, (Scribe)

## Guests

Carol Greene  
Sarah Viall  
Christy Keppel, March of Dimes

## Members Absent

Senator Ronald Young  
Delegate Karen Lewis

## Ex-Officio Present

Robert Myers, PhD  
Johnna Watson, RN  
Dr. Lee Woods

Called to Order – 5:10 pm

## I. Welcome and Introductions

Members and guests introduced themselves.

## II. Minutes of Meeting April 5, 2016

Minutes were reviewed and approved.

## III. Old Business

### A. Membership Update

1. Anne Eder stated that the terms of two Health unrelated members, Ben Smith and John McGing, are expiring. Both are eligible to and have agreed to serve a second term. Their applications for re-appointment will be reviewed by the Governor's office.
2. Anne Eder also stated that there are three applications that will be presented to the Governor's office for the remaining Health Unrelated vacancy. The appointment coordinator has stated that the Governor is interested in adding diversity – geographic, ethnic, and condition related - to the Advisory Council. It may be several months before decisions are made.
3. Dr. Neil Porter was invited to continue to represent the Monumental City Medical group until another representative has been identified.
4. Anne Eder stated her term as Chair of the Advisory Council is expiring although her term as a Council member continues. There is a quorum present and Anne nominated Dr. Hilary Vernon to serve as Chair effective 07/01/2016. There were no other nominees. The motion was seconded and Dr. Vernon was elected as Chair with no opposing votes.
5. Anne Eder also discussed that a Vice Chair is needed and nominated John McGing. No other nominees were offered. The motion was seconded. John McGing was elected as Vice-Chair with no opposing votes.

### B. Update on status of Pompe, Fabry and X-ALD

1. The Advisory Council sent a letter to the Secretary of Health in January 2016 notifying the Secretary that the Advisory Council has voted to recommend inclusion of these conditions to the state newborn screening panel. To date there has been no response from the Secretary's office. Johnna Watson, Chief of Newborn Screening Follow Up Program has asked Donna Harris, Director OGPSHCN, to follow up.

2. The Council asked Dr. Myers, Laboratory Director, when testing for these conditions could commence. Dr. Myers explained that until he receives the approval from the Health Secretary, he cannot pursue a cost analysis. The cost analysis will determine if new technology and new instrumentation is needed as well as if there are issues related to lab capacity. Once the cost analysis has been completed, a time frame to implementation can be formulated.
- C. Annual Review of Lysosomal Storage Disorders
1. Anne Eder stated that it is time to re-consider several conditions previously presented to the Council for inclusion in the Newborn Screening panel. Mucopolysaccharidosis Type 1 (MPS 1), Neimann Pick, and Krabbe were not recommended at the time of initial presentation and should be reviewed to identify current research and treatment updates. After the presentations, the conditions will again be put to vote.
  2. Dr. Vernon initially presented Neimann Pick to the council on 12/09/2014 and Krabbe on 04/01/2014. She agreed to provide an update on these conditions for the September, 2016 meeting.
  3. MPS 1 was presented by Dr. Regier, CNMC Genetics in November, 2014. Sarah Viall, representing CNMC genetics, offered that Dr. Regier would be happy to provide Council with an update on MPS 1.

#### **IV: New Business**

##### **A. Federal Updates**

1. Johnna Watson stated that the Federal Advisory Committee on Heritable Disorders in Newborns and Children is planning to review two disorders.
  - a. At the May 2016 Federal Advisory Committee, the work subgroup presented a summary of GAMT (Guanidinacetate methyltransferase deficiency), a creatine disorder. The Committee did not advise forwarding the condition to evidence review because a case has not yet been identified by newborn screening. Dr. Greene stated the workgroup also identified a lack of consistency re: treatment protocol for this disorder.
  - b. Dr. Vernon suggested that Dr. Lisa Kratz, Kennedy Krieger Institute Laboratory, present GAMT to the Advisory Council. The KKI lab is one of the few in the country that tests for this condition.
  - c. Spinal Muscle Atrophy (SMA) is also being reviewed by the Federal Advisory Committee for a second time. It was suggested that the Advisory Council may have enough to consider this year with the reviews of LSD and GAMT. A presentation on SMA can be considered for 2017.
2. The Federal Advisory Committee has also discussed the issues of medical food and will be reviewing the topic in subcommittee.
  - a. In follow up to Jennifer Payne's appeal for assistance in coverage for formula, the Council discussed whether there is a role for the Advisory Council on the issue that Ms Payne presented April 5, 2016 regarding medical foods, specifically addressing the labelling and classifying of medical foods so that they are covered by insurance. Following discussion, a motion was made and seconded recommending that the Advisory Council send a letter of support to the Federal Advisory Committee in favor of supporting inclusion of medical foods at the federal level.
  - b. Prior to drafting the letter, it was suggested that the Council confirm the support of the Maryland Secretary of Health. Dr. Bruno and Mr. Smith will draft a letter to the Governor's office and present it to the Advisory Council for comments and review. Dr. Greene mentioned that it may be helpful to review the language of letters sent to HRSA on this topic.
  - c. The motion was put to a vote and passed without any opposition.

##### **B. Legislative Update**

1. There was an amendment to language of the newborn screening legislation. As enacted, it requires DHMH's Newborn Screening Program to notify parents/guardians of newborn infants that laboratories other than DHMH's public health laboratory are authorized to perform post-screening confirmatory or diagnostic tests on newborn infants for hereditary and congenital disorders. The brochures that hospitals give to parents and the Laboratories administration website are being updated. Johnna

- Watson will present the brochure draft to Council members electronically for their review and comments.
2. Legislation related to Direct to Consumer genetic testing products did not advance beyond the House committee.
  3. Aiden's Law which requires states to test for X-ALD remains in federal committee, which recesses in August.

**C. SCID**

Dr. Myers reported screening for SCID (Severe Combined Immunodeficiency) commenced April 1, 2016. No confirmed cases have been identified. There is a problem with higher than anticipated volume of abnormal results on repeat specimens. The laboratory is reviewing the 2<sup>nd</sup> month's data and will revise the protocol if appropriate. The laboratory is interviewing candidates for the two contractual scientist positions recently approved for the SCID lab.

**D. Member Updates**

Laboratory Administration had no additional updates.  
Maternal Child Health Bureau - Dr. Woods stated that there are 19 travel related cases of Zika virus in Maryland. No locally acquired cases have been identified.

**E. Other Issues**

Mr. Smith recommended that Dr. Escolar be invited to speak on Krabbe and requested that this condition be reviewed at a meeting separate from the others. Mr. Smith will aid in contacting the speaker and arranging the date with the Council Chair.  
Dr. Bruno requested copies of the presentations of the previously presented LSD conditions so that he can review the content prior to the September meeting. Anne Eder also offered to send him the scoring tool electronically.

**V. Next meeting**

1. Next meeting date –September 6, 2016.

**VI. Adjournment**

1. Meeting was adjourned at 6 pm.