

# State Advisory Council on Hereditary and Congenital Disorders

Minutes January 12, 2016

## **Members Present**

Anne Eder, Chair  
John McGing  
Ben Smith (phone)  
Neil Porter, MD  
Erin Strovel, PhD  
Hilary Vernon, MD  
Delegate Karen Lewis Young (phone)

## **Members Absent**

Ronald N Young  
Aaron Kauffman

## **Ex-Officio Present**

Deborah Badawi, MD  
Robert Myers, PhD  
Fizza Majid, PhD  
Dr. Lee Woods

## **Staff**

Linda Lammeree, RN, (Scribe)  
Tina Wiegand  
Johnna Watson, RN

## **Guests**

Elizabeth Hall (phone)  
Donae Smith (phone)  
Sarah Viall, NP  
Dr. Ada Hamosh  
Dr. Carol Greene

**Called to Order** – 5:00 pm

## **I. Welcome and Introductions**

- Anne Eder suggested the opening of the meeting be delayed for a few minutes giving others affected by the traffic conditions a little more time to arrive.
- Meeting commenced at 5:10 pm
- Members and guests introduced themselves, including two candidates for membership.
  - Elizabeth Hall introduced herself as a social worker and survivor of congenital heart disease. She is very interested in supporting activities related to improving systems of care. She has submitted an application to the Governor's office to be considered for a seat on the Advisory Council as Health unrelated member.
  - Donae Allen introduced herself as a program coordinator for a non-profit agency that provides services to people with developmental disabilities. Her son has dwarfism (capitalized?) and she is interested in supporting programs and activities that promote early discovery of medical conditions. She will submit her application to the Governor's office as soon as it is completed in order to be considered for a seat on the Advisory Council as Health unrelated member.

## **II. Approval of December 1, 2015 Minutes**

Minutes reviewed and approved.

## **III. Old Business**

- Anne Eder, chair, stated that she is re-arranging the order of the agenda in order to continue addressing the topic of membership started during introductions. The vote on X-ALD will take place after membership update.
- **Membership update**
  - Anne Eder reported that Dr. Robert Bruno has submitted an application to the Governor's office to be appointed as the representative for Med Chi (the Medical and Chirurgical Faculty of the State of Maryland), seat formerly held by Dr. Takai.
  - Anne also stated that with the applications of Ms. Hall and Ms. Allen there are multiple candidates for the Health unrelated seat.
  - With the selection of the Health unrelated representative, the Advisory Council membership will be complete.

- **Vote on X-ALD**
  - Anne Eder stated that there is a quorum present at this meeting to permit a vote on whether to add x-ALD to the Maryland newborn screening panel.
  - Prior to the vote, Anne Eder wanted to initiate discussion on the first time use of the screening tool developed by the Advisory Council.
    - Dr. Badawi stated a review of score totals and per item revealed mostly consistent scoring ranging from 1275-1650 out of possible 2100. The absence of any discrepancies or wide variations, indicated the presentation by Dr. Barananos in December, 2015 sufficiently covered all the points necessary for Council members to vote.
    - Anne Eder stated she would like to see the screening tool re-designed to make it more user friendly for the non-clinical, non-medical members.
    - Anne also remarked that she would like to see the presentations follow the screening tool's lead and be more easily understood by the less medically trained members of the Council.
  - Dr. Vernon made a motion to add X-ALD to the Maryland newborn screening panel and it was seconded by John McGing.
  - Motion passed with 6 votes and 1 abstention. Anne Eder abstained from voting as the position of the March of Dimes organization, whom she represents, has not yet been determined.

#### **IV. New Business**

- **Federal Update**

- Newborn Screening Contingency Plan
  - Dr. Badawi reported there is a plan to review and update the contingency plans developed in 2010 to help ensure continuation of operations of newborn screening labs and programs in the event of an emergency such as hurricane, etc.
  - A survey to determine the level of awareness of the contingency plan is being developed by the Maternal and Child Health program and will be sent to clinical geneticists, state laboratories and follow up programs, primary care providers, hospital administrators and nurseries and advocacy groups.
  - The goal is for feedback to be presented to the Federal level and revisions made by May, 2016.
  - In response to questioning about Maryland's contingency planning, Dr. Myers stated there are informal agreements in place to ensure the continuation of needs of newborns screening program in the event of an emergency such as hurricanes, etc. Formal mutual aid agreements between neighboring states are under discussion by Attorney General's office.

- **Council Handbook**

- Anne Eder stated is not necessary for the Council to vote on adopting the handbook.
- Changes to the handbook can be made as necessary and appropriate to reflect the work of the Council.
  - The timeline of relevant developments related to Maryland's newborn screening panel and program has not yet been added but is planned. Dr. Badawi thought this timeline might be formulated by working backwards using changes in regulations, proposed legislation, and correspondence.
  - Anne would like to finalize the handbook after the first of the year and requested that any further comments and suggestions be forwarded to her and Dr. Badawi.

- **Commercial Genetic Testing**

- Anne Eder introduced a discussion about commercially available kits for genetic testing that are marketed to the general public
  - While Maryland legislation currently prohibits genetic testing kits such as "23 and Me" because there is no physician involvement in the reporting of the results, there may be legislation proposed this session to change this.
  - Dr. Myers, as laboratory administrator, voiced the following concerns:
    - whether the test has been approved by FDA as valid,
    - whether test is being performed in a CLIA (not sure what this stands for – can you spell it out?) approved lab conditions
    - historically, the legislature was concerned that people could be using unreliable and invalid test results to make health care decisions.

- some kit companies were selling DNA to researchers without disclosing this to the consumers.
  - Dr. Badawi stated in 2013 she conducted a literature review on the topic
    - Approximately 27 states permit direct to consumer genetic test kits.
    - One study found people who used the test kits are more likely to participate in health screenings.
    - However there are legitimate issues of quality control and what is the test designed for—identification of disease state vs genealogy, that have not been resolved and vary from product to product.
  - Dr. Greene stated she is aware of the explosion of interest in this topic. She mentioned that a study reported same samples sent to several different laboratories resulted in conflicting results and too much ambiguity. She expressed the personal and professional concern that people may be making irreversible decisions such as mastectomy, sterilization, pregnancy termination and suicide based on these results.
  - Council members raised the following points during discussion
    - Personal decision making vs government making decisions for individuals.
    - Role of physicians in educating and counseling their patients. What if the physician does not know what the results mean?
    - Quality control and test validity standards.
    - What disclosures are included in these test kits?
    - What is the role of Advisory Council in this matter?
  - Anne Eder stated it is obvious there are many points of discussion for this topic. It is not necessary to make any decisions right now, but that Council members should be aware of the topic and related issues.
- **Legislative Issues for 2016 Maryland General Assembly**

This current legislative session is expected to consider a record number of bills. However, only those pre-filed (prior to December 2015) are visible at this time. Dr. Woods is not aware of anything relevant at this time.
- **Member Updates**
  - **SCID testing**
    - Dr. Majid stated the lab is working towards April 2016 implementation
    - CDC training has been completed.
    - Validation is expected to begin very soon.
    - A committee of immunologists is working with lab and Follow up program to develop algorithm.
    - Anne Eder asked how the providers and parents are informed about the addition to the panel.
    - Dr. Majid stated simultaneous educational efforts through letters to providers and through the website and in conjunction with Maternal Child Health Bureau.
  - **Status of conditions approved in 2015**
    - Pompe and Fabre were recommended in 2015 to be added to the Maryland Newborn Screening panel. A letter was sent to the Secretary of Health but a response has not yet been received.
    - Anne Eder will draft and send a reminder to Secretary of Health asking when Council might expect a response.
- **Meeting Schedule:**
  - Anne Eder acknowledged that it may be necessary to meet more frequently as issues related to proposed legislature become known.
  - Next meeting is February 23, 2016.
  - Meeting for March 8 needs to be re-scheduled due to conflict with American College of Medical Genetics conference

## **VI. Adjournment**

- Meeting adjourned at 6:20 pm