

State Advisory Council on Hereditary and Congenital Disorders

Minutes September 1, 2015

Members Present

John McGing
Ben Smith
Neil Porter, MD
Erin Strovel, PhD
Hilary Vernon, MD (phone after 7 pm)
Delegate Karen Lewis Young

Members Absent

Anne Eder, Chair
Coleen Giofredda
Aaron Kauffman
Caryl Siems

Staff

Linda Lammeree, RN, (scribe)
Hilda Castillo, PhD

Guests

Mimi Blitzer, PhD
Sarah Viall, NP, (phone)
Lauren Catterton
Ada Hamosh, MD, PhD

Ex-Officio Present

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

Called to Order – 6:00 pm

I. Welcome and Introductions

Dr. Badawi stated that Anne Eder, chair, was unable to attend and that she would lead the meeting in her absence. Members and guests introduced themselves.

II. Approval of June 2, 2015 Minutes

Minutes reviewed and approved.

III. Old Business

• Newborn Screening Criteria

- Dr. Badawi presented results of the small work group that met since previous meeting with the goal to develop a more structured process to aid in decision making when reviewing disorders for newborn screening panel.
- Work group members included Anne Eder, Dr. Strovel, Dr. Majid, Dr. Vernon, Sarah Viall, Mr. Smith and Dr. Badawi.
- The work group reviewed the charge of the Advisory Council to make recommendations to the Secretary re: new conditions to be added to the newborn screening panel; the ACMG criteria; and articles by Botkin and Petros.
- The current review process recently used by the Advisory Council when considering the various lysosomal storage disorders included a presentation of the condition by a medical expert, followed by questions/answers and discussion. A vote on whether to recommend inclusion was then scheduled for the next meeting. The work group recommended continuing with this general format with the suggestion that the presentation be prepared using the criteria points in question format.
- Discussion points:
 - There was concern mentioned about reviewer bias. It is expected that the medical expert preparing the presentation of the condition being reviewed will use factual information based on available published studies. It is recognized that reviewer bias could exist and should be identified as a personal opinion if included in the presentation.
 - It is not the intention of the group that a scoring number be used to determine acceptance or rejection of a condition but instead use of a scoring sheet will

- Dr. Badawi reported that Anne Eder thought a Handbook for Advisory Council members may be helpful, both as an orientation for new members and also as a reference. Dr. Badawi reviewed other councils' handbooks and has started a rough draft, which was used for the first time to orient new members prior to this meeting. The executive order to the Advisory Council is approximately 30 years old. Mr. McGing commented that it may be necessary to update the language since things like genome and ELSI (ethical, legal, and social implications) are relevant today but not in existence at time executive order was written.
- Dr. Blitzer asked who needs to approve any changes in the language of the executive order. Dr. Badawi stated that an update requires approval of the Secretary, and changes to the statute authorizing original charge. The council can make recommendations to the Secretary. This is a lengthy process and not necessarily a primary focus of the Advisory Council at this time.
- It was decided that Dr. Badawi will take this discussion to Anne Eder, chair of council, asking if she would like to establish a workgroup to review the Executive Order section by section and see how document aligns with modern law and technology. If changes are recommended, will have to find out the process for this.
- **Member Updates**
 - Dr. Badawi explained that effective July 1, 2015, the Maternal Child Health Bureau has re-organized in order to reflect services to all ages across the population. Divisions within the Office of Family and Community Health Services will address perinatal health, children and adolescents, women, reproductive health (including men) and cross-cutting issues.
 - Dr. Woods is the medical consultant for the Office for Genetics and People with Special Health Care Needs.
 - Dr. Badawi is now Chief of the new Child and Adolescent Health Unit, of the Office of Family and Community Health Services, still in the Prevention and Health Promotion Administration within Maryland Department of Health and Mental Hygiene.
 - Newborn screening has moved to Family and Community Health Services and Dr. Badawi continues as the Medical Director.
- **Membership updates**
 - Caryl Siems—her term expired, but she is still a sitting member until a new Health unrelated member appointed.
 - Dr. Sandra Takai—is resigning. New member is needed as representative from MedChi.
 - Continue to need Senate representation—Delegate Young may be able to help.
- **Additional Discussion:**
 - Mr. Smith asked if the Advisory Council was aware of changes in the REM (rare and expensive medical conditions) program regulations effective 07/01/2015 changing eligibility requirements. Dr. Badawi will follow up on this issue.
- **Meeting Schedule:**
 - Next meeting is scheduled for December 1, 2015
 - Meeting dates for 2016 will be discussed at this next meeting, when Council Chair is present.
 - Agenda item: discussion of the roles and responsibilities of the advisory council members and council's accomplishments and projects for inclusion in the handbook.

VI. Adjournment

- Meeting adjourned at 8:00 PM.