

State Advisory Council on Hereditary and Congenital Disorders

Minutes June 2, 2015

Members Present

Anne Eder, Chair
Aaron Kauffman (phone)
John McGing (nominated)
Ben Smith
Neil Porter, MD
Delegate Karen Lewis Young (phone)

Members Absent

Coleen Giofredda

Erin Strovel, PhD
Hilary Vernon, MD
Caryl Siems

Ex-Officio Present

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

Staff

Johnna Watson, RN (scribe)
Linda Lammeree, RN
Hilda Castillo, PhD
Tina Wiegand (phone)

Guests

Carol Greene, MD (phone)
Kendra Sullivan
Susan Sullivan
Katie Sheehan
Melissa Shoemaker
Heather Bryant
Claire Pierson
Jennifer Ehrlich
Mimi Blitzer, PhD
Scott Williams (phone)

Called to Order – 6:00 pm

I. Welcome and Introductions

Anne Eder welcomed members and guests. Members and guests introduced themselves.

II. Approval of February 2015 Minutes

Minutes reviewed and approved.

III. Old Business

• Newborn Screening Criteria

- Anne Eder recapped the process for voting on the addition of new screens to the state's newborn screening panel. The process includes a comprehensive scientific literature review, expert presentations, discussion, and voting.
- Dr. Badawi, prior to the meeting, distributed scoring criteria from Botkin and Petros. The most recent scoring criteria is Petros which was published in 2012. Anne inquired as to what criteria is used at the Federal level. Dr. Badawi stated that the federal committee evaluates the clinical utility of testing as well as feasibility.
- Dr. Blitzer stated that in hindsight, it would have been helpful if the council had defined the criteria to use prior to the voting process.
- Aaron Kaufman added that, as a non-medical professional, he felt like a fish out of water due to the highly-technical nature of the articles and presentations.
- Anne stated that she felt there was a nice mix of medical and non-medical people contributing to the discussion and the voting process.
- Ben Smith stated the need for criteria for selection of disorders was discussed at the last meeting as a means of adding transparency to the process. The use of specific criteria provides insight into why a particular disorder is not included.
- Dr. Badawi agreed that such a process would help make it clear why particular decisions are made. She noted that the original criteria used by the NBS community in years past was more simplistic. Part of criteria now being discussed include decreasing time to diagnosis and the ability to provide a diagnosis, even if treatment is still evolving. There should also be a facility to provide treatment.
- Dr. Blitzer stated there are different interpretations of treatment. The Council needs to develop a list of criteria to use when discussing each disorder. This should be consistent information that is used as a foundation when discussing each disorder. Criteria are subjective and will be based on what is important to each Council member. Dr. Blitzer added that the Council should also discuss what is happening at the Federal level.

- Ben Smith indicated that Federal review process is too long.
- Dr. Badawi indicated that Federal review process now must be completed within a 9-month period.
- Anne Eder suggested convening a workgroup of 2-3 people to draft a checklist for the Council to use when considering additions to the newborn screening panel. This group will meet by phone over the summer. Ben Smith, Anne Eder and Dr. Majid volunteered to serve on the workgroup. Drs. Hilary Vernon and Erin Strovel will be asked to serve as well.
- **Strategy for reviewing lysosomal storage disorders**
 - Dr. Badawi stated that the Council is committed to revisiting each of the disorders on an annual basis. Medical experts will be invited to present any new information. The disorders can also be reviewed using the checklist developed by the workgroup.
- **SCID screening update**
 - Dr. Myers and Dr. Majid reported that the lab is currently purchasing equipment by using residual funding from leftover capital funds. Some of the new equipment has arrived. The machine will not be delivered until July. The plan is to visit two states to observe techniques and training. There is funding available from the Association of Public Health Laboratories for startup costs. The goal is for screening to be underway in the fall or by end of the year.
 - Ben Smith asked for a tour of the new lab. Dr. Myers stated the meeting in September can be scheduled at the new lab and include a tour of the facility.

IV. Old Business

- **Legislative Updates**
 - Anne Eder reported that the NBS Fund Bill passed. The legislation establishes a fund to cover the administrative, laboratory, and follow-up costs associated with performing newborn screening tests. The fund will assist in the more timely implementation of new screening tests and will cover the purchase of new equipment. Delegate Lewis Young voted for the bill. Ben Smith reported that Delegate O'Donnell was primary sponsor of this bill.
 - Dr. Badawi reported on the status of other bills:
 - SB 156 – Bill creating educational outreach for adult sickle cell patients. Bill did not make it out of committee.
 - HB 9 – Bill providing licensure of direct entry of non-nursing midwives was passed. The midwives have to provide NBS screening. Babies must be seen by pediatrician within 72 hours of age.
 - HB 781 – Bill concerning ostomy supplies in MD was passed but vetoed because coverage was already addressed under different regulations.
 - HB 1122 – Bill stipulating that an individual cannot be denied an organ transplant due to mental or physical disability. Successfully passed.
- **Member Updates**
 - Dr. Blitzer reported that she and Johnna Watson worked with the University of MD and NYMAC to create a film to educate midwives about their responsibilities in conducting newborn screening. Filming was completed 2 weeks ago in Albany and is available in both English and Spanish. The film will be shown to Council members at the next meeting. It will be promoted via YouTube, state websites and the Nurse Midwifery Association.
 - Dr. Myers reported that the Newborn Screening Laboratory moved to the new lab building on 05/11/2015. Building is at the corner of Ashland and Rutland Avenues. Tour to be available during September Council meeting.
 - Dr. Badawi reported that Maternal and Child Health Bureau is undergoing reorganization, and Dr. Badawi will be moving out of OGPSHCN. At this time, Newborn Screening Follow-Up will be located administratively in the new Child and Adolescent Health Unit, but it is unclear if it will remain there or move to labs.
 - Ben Smith introduced Melissa Shoemaker who has a baby diagnosed 2 months ago at 6 months of age with Krabbe disease. New testing is also being trialed by Dr. Glove at University of Washington for MLD (metachromatic leukodystrophy).
 - Ben Smith asked about screening for X-ALD. Dr. Badawi stated Federal review is underway on X-ALD. Will plan to discuss at December meeting.
 - Anne Eder reported that a letter was sent to the Secretary of Health in April recommending screening for Pompe but no response at this time.
- **Membership Updates**

- There will be One Health Unrelated position open at the end of June when Caryl Siems term expires. Lauren Hicks has applied for this position.
- Delegate Lewis Young will be new representative from House of Delegates.
- There is an open Senate position at this time.
- Ben Smith asked about members who haven't been participating in the meetings. Anne Eder indicated that Mr. McGing is replacing one of the members who has not been attending.

V. Meeting Schedule

- September 1, 2015 (Agenda will include a tour of new state lab facility and viewing of Dr. Blitzer's nurse midwifery film)
- Dec 1, 2015

VI. Adjournment

- Meeting adjourned at 8:00 PM.