

State Advisory Council on Hereditary and Congenital Disorders

Minutes June 24, 2014

Members Present

Anne Eder, Chair
Erin Strovel, PhD
Coleen Giofredda
Ben Smith (phone)
Neil Porter, MD
Sandra Takai, MD (phone)
Caryl Siems

Members Absent

Delegate Shirley Nathan-Pulliam
Hilary Vernon, MD
Alan Kauffman

Ex-Officio Present

Deborah Badawi, MD
Robert Myers, PhD

Called to Order – 6:00 pm

I. Welcome and Introductions

Anne Eder welcomed Ben Smith and Alan Kauffman as newly appointed members of the Council, both in category of Health Unrelated. Mr. Kauffman was unable to join the meeting tonight. All members and attendees introduced themselves.

II. Approval of April 2014 Minutes

Minutes reviewed and approved.

III. Old Business

- **Update on SCID supplemental funding**
 - Dr. Myers reported there is an implementation plan ready to go. The laboratory is currently in negotiations with the budget office. The Secretary of Health has been involved in trying to obtain funding. They are working to obtain a loan to purchase equipment. There may also be a small increase to the newborn screening fee to cover costs. Funding is still under active consideration, and Dr. Myers will share the implementation plan once funding is secured.

- **Newborn Screening for Krabbe Leukodystrophy**
 - Anne Eder reviewed current progress in the discussion regarding adding screening for Krabbe Leukodystrophy. Dr. Hilary Vernon gave a slide presentation at the meeting in April and has forwarded updates to all Council members in the interim.
 - Dr. Badawi explained the process of adding a condition to the MD newborn screening panel. The recommendation to add a condition is forwarded via letter from the Council President to Secretary Sharfstein. The Secretary can accept or decline the recommendation.
 - Ben Smith stated that Tennessee has just added lysosomal disorders to their panel, joining 8 other states that are currently adding them to their panels.
 - Anne Eder asked if there were any questions or discussion. No questions or discussion was initiated. Dr. Porter moved to have a vote on the condition.
 - Voting followed with 4 votes against screening (Neil Porter, MD, Erin Strovel, PhD, Sandra Takai, MD and Anne Eder) and 3 votes for screening (Colleen Giofredda, Caryl Siems and Ben Smith)
 - Recommendation to not include screening for Krabbe Leukodystrophy in the Maryland Newborn Screening Panel will be forwarded in a letter to Dr. Sharfstein. Anne Eder indicated this condition can be reconsidered and the plan is to revisit screening for Krabbe on an annual basis.

Staff

Johnna Watson, RN (scribe)
Linda Lammeree, RN

Guests

Debbie Romanoski,
Senator Dyson's office
Mimi Blitzer, PhD
Ada Hamosh, MD
Ilise Marrazzo
Jessica Albert
Sarah Viall (phone)

IV. New Business

- **Council Membership**
 - Anne Eder reported that both vacant Health Unrelated positions have been filled. The Senate position is currently open.
- **Letter from Deborah Haupt regarding carrier status**
 - Dr. Badawi stated that a letter was received by the Secretary from Deborah Haupt who worked with New York NBS Program to add language in their parent brochure indicating that an infant may be determined to be a carrier for cystic fibrosis (CF) through newborn screening. Dr. Badawi shared copies of MD brochure to discuss whether carrier status should be added into the brochure.
 - Dr. Hamosh reported that since MD does not perform DNA at this time, carrier status is determined at the specialist level and is not identified during screening process, other than sickle cell trait.
 - Dr. Takai agreed the NBS brochure should not address whether or not carriers are identified.
 - Dr. Badawi suggested information regarding determination of carrier status should be more available for birth providers, via a website.
 - Caryl Siems also agreed this information should be given preconceptionally.
 - Ilise Marrazzo indicated there is no information currently on carrier status on a website in Maternal Child Health Bureau. She agreed this information should be addressed as a partner with birth and pre-conception providers. A Reproductive Life Plan is being developed currently and this information could be incorporated into it.
 - Dr. Myers reported that the Secretary is looking for input from the Council on how to answer Ms. Haupt's letter. Dr. Badawi stated she would provide him input based on the Council's discussion.
- **Member Updates**
 - Dr. Myers reported the lab is scheduled to move in the fall. The punch list for the 1st lab is currently in progress. The last 5% of construction is taking longer than expected.
 - Caryl Siems reported the clinical trial combining two CF drugs has had good results. This combination is formulated for individuals with 2 delta F508 mutations.
 - Since screening for Krabbe was voted down today, Ben Smith would like the Council to readdress offering supplemental screening to parents in Maryland. Recommended developing a brochure informing parents that additional screening is available through outside laboratories. Hunters Hope website has information on laboratories who perform screening for lysosomal storage disorders. Dr. Takai stated the MD Newborn Screening Program should not be advocating for screening for disorders that are not on their panel. Anne Eder indicated the possibility of developing a brochure could be placed in the letter to Secretary Sharfstein.
 - Dr. Badawi reported the Council will continue to discuss screening for lysosomal storage disorders. The next meeting will include a discussion of the next LSD, which is Pompe. Information on this disorder and outcomes will be gathered and disseminated. Dr. Blitzer suggested that information reviewed by the Discretionary Advisory Committee on Hereditary Disorders in Newborns and Children in making their decision be shared with the Council.

V. Next Meetings

- September 9, 2014
- November 18, 2014

VI. Adjournment

- Meeting adjourned at 7 PM.