State Advisory Council on Hereditary and Congenital Disorders

Minutes September 9, 2014

Members Present
Anne Eder, Chair
Erin Strovel, PhD
Hilary Vernon, MD
Ben Smith (phone)
Neil Porter, MD

Members Absent
Delegate Shirley Nathan-Pulliam
Coleen Giofredda
Sandra Takai, MD
Alan Kauffman
Caryl Siems

Ex-Officio Present
Deborah Badawi, MD
Robert Myers, PhD
Fizza Majid, PhD

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

Guests
Carol Greene, MD
Debra Regier, MD
Mimi Blitzer, PhD
Ada Hamosh, MD
Pranoot Tanpaboon, MD phone)
Sarah Viall (phone)

Called to Order – 6:00 pm

I. Welcome and Introductions
All members and attendees introduced themselves.

II. Approval of June 2014 Minutes
Minutes reviewed and approved.

III. Old Business

- **Newborn Screening for Pompe Disease** (Dr. Pranoot Tanpaiboon)
  - Dr. Tanpaiboon was introduced by Anne Eder as a clinical geneticist at Children's National Medical Center who oversees their Lysosomal Storage Disorders Clinic for 6-7 years.
  - Dr. Tanpaiboon gave a slide presentation outlining Pompe Disease (slides attached)
  - Treatment is supportive and symptomatic. Enzyme replacement therapy (ERT) was approved by FDA in 2014 for infantile onset Pompe Disease. Without ERT, individuals with classic infantile Pompe usually die within the 1st two years of life. Success of ERT treatment depends on cross reactive immunologic material (CRIM) status. Individuals with CRIM negative status must undergo chemotherapy prior to ERT.
  - Taiwan has been screening for Pompe in dried bloodspots since 2005. Outcome for babies identified in Taiwan has only been followed for 4 years. It has been noted that ERT does not help all muscles.
  - Dr. Vernon asked if Dr. Tanpaiboon had any information on how NY experience is working. Dr. Tanpaiboon does not have this information.
  - Dr. Badawi indicated the Discretionary Advisory Committee on Hereditary Disorders in Newborns and Children has recommended Pompe be added to Federal RUSP but the Secretary has not responded.
  - Dr. Blitzer asked how many states are currently testing for Pompe. Dr. Majid responded that Missouri and NY are testing. Other states have not implemented testing at this time.
  - Dr. Majid stated there is not currently a good test for Pompe. Enzyme activity measurement with MS/MS is not reliable. Dr. Tanpaiboon stated there was a high false positive rate in Taiwan, and the criteria was changed, and since 2013 there is a 92.9% sensitivity with 2nd tier testing.
  - Dr. Vernon asked how to determine from newborn screening if baby has infantile form or late onset. Dr. Tanpaiboon indicated this is determined by symptoms. Dr. Hamosh indicated that all individuals with infantile form have cardiomyopathy so ECHO is used to differentiate between the two forms. Late onset Pompe is not treated until symptoms appear. Dr. Hamosh also indicated that there is an effective treatment in ERT.

- Dr. Greene stated there is a full evidence review by the Discretionary Advisory Committee on Hereditary Disorders in Newborns and Children. Dr. Badawi will circulate link to their findings.
- A quorum was not available at this meeting so a vote was not conducted tonight. Benjamin Smith announced his presence on phone, but an additional member was needed for a quorum.
- Dr. Blitzer asked if there was anything in the legislation that states MD has to screen for Pompe if it is added to Federal RUSP. Dr. Badawi replied the legislation does not have this stipulation.

- **Plan for completing voting on Lysosomal Storage Disorders** (Anne Eder)
  - Anne Eder reviewed plan for voting. A presentation was given on Krabbe by Dr. Vernon, and in a subsequent meeting, the Council voted not to add Krabbe to the state's newborn screening panel. The other conditions need to be reviewed and reported back to the House Government and Operations Committee by 12/2015.
  - Anne Eder indicated the Council needs to work on getting members to attend the meetings so voting can occur. Ben Smith stated there should be a condition that if a member does not attend 2 meetings then they should be removed from the Council.
  - Dr. Blitzer suggested that the vote on a condition should not take place on the same day as the presentation to allow members time to review the material in more detail. Anne Eder agreed with this plan.
  - Discussion occurred on how disorders should be grouped. It was discussed whether Gaucher and Neiman Pick will be reviewed together and Fabrey and Hurlers will be reviewed together. Ben Smith interjected that all of the diseases should be tested together and not considered separate. Dr. Badawi stated the disorders need to be reviewed separately because there is too much information to discuss all of the disorders at once. Ben Smith stated review process is taking too much time and is taking time away from screening for ALD. Dr. Hamosh and Dr. Strovel both indicated disorders should be reviewed separately. Plan is to hold 1 meeting a month or every other month to speed up the process.
  - Dr. Greene asked if Bylaws permit electronic voting. Anne Eder also suggested voting by phone. Dr. Badawi will check the Bylaws and will also send an APB out to members to obtain a quorum by the next meeting so voting can take place on Pompe.

- **Update on SCID supplemental funding**
  - Dr. Myers reported he has received an email from the Secretary that the laboratory can move forward with buying capital equipment necessary to start SCID screening. Based on move date to the new laboratory, plan is for validation in Spring 2015 and live in Summer 2015. There will be a modest increase in fees to cover costs. Ben Smith stated the NBS supplemental funding bill will be reintroduced in the next legislative session.
  - Dr. Majid reported 17 states are currently screening for SCID and 4 are in pilot stages. Dr. Badawi reported she has immunology contacts with each of the 3 centers. All 3 centers have reported ability to do transplants.

IV. New Business

- **Member Updates**
  - Office of Genetics and Children with Special Health Care Needs
    - ✓ Dr. Badawi reported CCHD data is being pulled through 1st year of screening. One of the graduate students is also working on discharge data to help see if there are any CCHD conditions that have been missed through screening.
    - ✓ Progress on HSRA long term follow-up grant reported: patient recruitment has been slow and all data is being entered by paper currently.
  - No additional member updates noted

- **Council Handbook**
  - Anne Eder distributed Member Handbook for the Maryland State Council on Cancer Control. She asked for the Council's input on the development of a similar resource for Hereditary Council members. If this is of interest, volunteers will be needed to assist with the development of the handbook.

- **Request for interview**
  - Anne Eder reported that March of Dimes is in need of geneticist for interview on September 25th. Focus on genetic testing and its importance. Members were asked to email recommendations to Anne.

- **Reimbursement for genetic testing**
  - Dr. Blitzer reported that the State of Colorado is working on regs to have Medicaid pay for genetic testing. Other states should look into this idea as well. Dr. Badawi will send the information that she has received by email.

V. Next Meetings

- October 7, 2014 (tentative)
- November 18, 2014

VI. Adjournment

- Meeting adjourned at 8 PM.