State Advisory Council on Hereditary and Congenital Disorders

Minutes February 12, 2014

Members Present
Miriam Blitzer, PhD, Acting Chair
Erin Strovel, PhD, (nominee to Council)
Hilary Vernon, MD, (nominee to Council)
Caryl Siems
Neal Porter, MD
Anne Eder
Sandra Takai, MD

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

Members Absent
Delegate Shirley Nathan-Pulliam
Coleen Giofredda
Anika Wilkerson

Staff
Johnna Watson, RN (scribe)
Linda Lammeree, RN
Angela Sitler
Tina Wiegand

Guests
Carol Greene, MD
Katie Bisordi
Douglas Mogul, MD
Tiffany McNair, MD
Julie Hoover-Fong, MD

Called to Order – 6:00 pm

I. Welcome and Introductions
Members and attendees introduced themselves.

II. Approval of October 2013 Minutes
Minutes reviewed and Dr. Greene requested addition to the minutes to include comments made by herself and Dr. Hamosh regarding the quality of the currently published data regarding treatment for lysosomal storage disorders. Notes were given to Dr. Badawi regarding these comments.

III. Special Discussion

- **Newborn Screening for Biliary Atresia**
  - Presentation given by Dr. Mogul, pediatric gastroenterologist at Johns Hopkins.
  - Biliary atresia is most common cause of liver transplants in US.
    - ✓ Occurs in 1:8,000-18,000 births (higher prevalence seen in Asian population
    - ✓ In US, average age to diagnosis is 70 days. Outcomes would be improved with shorter time to diagnosis
    - ✓ Approximately 5-8 cases/year is predicted rate in Maryland
  - In 2004, Taiwan adopted stool color card screening
    - ✓ Pale stool is indicative of biliary atresia
    - ✓ Sensitivity is 97% and specificity is 99.5%
    - ✓ 5 year survival increased from 56 to 89%
    - ✓ Taiwan has a registry to track screening (80% response rate)

- Potential Screens are in development
  - ✓ Measurement of direct bili. In biliary atresia, direct bili increases in first 24 hours.
  - ✓ Measurement of bile acids in dried blood spots. Taurocholate (a bile acid) performed best with sensitivity of 79%
  - ✓ Mobile Health application for smartphones – plan is to use interactive color matching software to identify acholic (lack of bile) stools. The app would remind parents to check the color of their baby's stool at 2, 4, 6 and 8 weeks of age. If the stool is too pale, there would be an automatic reminder to call the baby's primary care provider.
- There is a current small pilot study in Hopkins nursery.
  - ✓ The baseline knowledge regarding stool color is being assessed, along with anxiety of the mother. Education given and then knowledge and anxiety are reassessed.
- Dr. Mogul states none of the states has done a large group study at this time. He wants to explore possibility of conducting one in Maryland.
  - ✓ Dr. Greene commented that by calling it a NBS DHMH would have to be involved, and Dr. Badawi indicated it should be a standard of care through AAP. Dr. Mogul said he is trying to get the screen placed on the neonatal periodicity table but wants to conduct a large scale study to prove effectiveness.
  - ✓ Dr. Badawi explained that all hospital births are currently in a database and DHMH might be able to support a compliance rate study.
  - ✓ Dr. Blitzer suggested starting with a few of the large birth hospitals in the State.
  - ✓ Dr. Mogul is exploring funding opportunities at this time and is in discussion with Proctor and Gamble currently to possibly add something to their diaper boxes.
  - ✓ Dr. Greene also offered that Joint Commission is trying to get total bili done on every baby at 24 hours of age so may be able to add direct bili as well.
- Dr. Mogul will continue to talk with Dr. Badawi regarding this issue.

IV. Old Business

- **Newborn Screening for Lysosomal Storage Disorders**
  - Dr. Blitzer reported that SB 433/HB891 also known as Lily's Law was introduced in this legislative session.
    - ✓ Hearing held by Finance Committee last week.
    - ✓ Laboratories Administration put in a fiscal note.
    - ✓ DHMH opposed bill secondary to process of review – The addition of disorders to the newborn screening panel is not legislated, but done through review process by the Council. Dr. Badawi indicated it is difficult to guess if the bill will go out of committee although some legislators seemed to be swayed by the family's testimony.
    - ✓ Dr. Greene stated that Krabbe disease was rejected for addition to the RUSP by the Federal Advisory Council prior to formal review.
    - ✓ NY currently screens for Krabbe disease. Missouri is performing a statewide pilot screening study. A few other states have passed legislation but are not screening yet..
    - ✓ Dr. Majid stated there is no FDA approved test for Krabbe disease screening at this time.
    - ✓ Hearing is not scheduled currently in the House.
    - ✓ Dr. Greene indicated that if screening is done using DNA will also identify carrier status and this will need to be addressed as well.

- **Update from NBS Lab/Laboratories Administration**
  - Dr. Myers reported that he has asked for supplemental funding for SCID NBS in the Governor's budget. No response at this time.
    - ✓ Anne Eder stated that March of Dimes has an Advocacy Day in the legislature and will include funding for SCID NBS in their plans. Will look into how to expand into meeting with the Governor's Office. Dr. Badawi stated that legislators do have some pull in adding to the supplemental budget.
    - ✓ Payments of the NBS fee, which is currently $100, goes into general fund and then may be reallocated to the Lab, but the funds do not go directly to support the NBS lab.
  - Move to new lab will be started in late May or early June.

- Dr. Majid reported that they have been working with submitters to increase timeliness of specimen submission. Goal is to have 90% of specimens received with 72 hours after collection. Measuring from 10/1-12/31/2013, 19 hospitals are above 90% now. Graphs are given to the hospitals for comparison. Dr. Greene suggested the graphs also go to Risk Management and Caryl Siems also suggested Patient Safety Committee.

- **Update from OGPSHCN**
  - Dr. Badawi reported that Maternal and Child Health Grant has been level funded which will result in some cuts that have been delayed from previous years.
- **Update on NCAA and sickle trait pilot project**
  - Dr. Blitzer reported that University System of MD is proposing to develop an educational program for the different campuses about sickle trait screening in athletes. Dr. Greene said that Alexia Thomas with HRSA is head of a program there discussing the NCAA screening policy and impact on NBS labs. Presentation on this program will be given at the next meeting.

V. New Business

- **Membership Changes and Election of New Chair**
  - There are 11 voting representatives on the Council
    - ✓ There is 1 non-health related vacancy
    - ✓ There is also a vacancy from the Senate
  - Dr. Blitzer moved that Anne Eder be elected Chair of the Advisory Council
    - ✓ The nomination was seconded and voting members elected Anne Eder as Chair
- **Current Legislation in General Assembly**
  - Dr. Badawi updated the Council on the Senate hearing on SB 433 (adding lysosomal storage disorders to newborn screening panel)
    - ✓ Mr. and Mrs. Smith (Lily's parents) gave testimony, along with her grandmother.
    - ✓ Difficult to determine how committee members will vote
    - ✓ DHMH does not support the bill on the basis that addition of disorders to the newborn screening panel should be determined by scientific review and not by legislation.
    - ✓ Subcommittee of this Council appointed to review lysosomal disorders for possible future addition to the newborn screening panel.
  - HB 906 – Direct to consumer genetic testing
    - ✓ Laura Herrera submitted a report which indicated DHMH thinks this type of testing may not be a good idea. DHMH support is not known at this time
    - ✓ Dr. Greene suggested there should be a disclaimer that an individual has to talk to a healthcare provider prior to testing.
  - HB 761 – Specialty Drug Coverage
    - ✓ limits copays for specialty drug coverage
    - ✓ No position from PHPA on this bill
  - SB 654 – provision of information to families of children diagnosed with Down's Syndrome
    - ✓ Information needs to be given to families of Down syndrome children diagnosed both prenatally and postnatally.
    - ✓ Dr. Hoover-Fong indicated this should be a standard of care.
    - ✓ DHMH did not take a position since this is an operational impact only.
  - It was recommended that the Council members review the remaining bills for possible comments.

- **Parking for Council meeting**
  - Dr. Badawi stated that parking is no longer available in the garage for evening meetings. Department of General Services feels it is a security issue since there is not enough security in the building after 6 PM. A member of security is supposed to escort the visitor to the room from the garage. It was reported from members of the Council that they are not escorted from the front desk to the conference rooms when they come in from the outside parking lots either. The Council Chair (Anne Eder) will write a letter to the Secretary of Health to see if he can get garage parking approved.

VI. Next Meeting

- Planned for April 1, 2014

VII. Adjournment– 8:00 PM

- Dr. Blitzer announced this is her last day on the Council. Appreciation for service expressed. Meeting adjourned.

State Advisory Council on Hereditary and Congenital Disorders

Minutes April 1, 2014

Members Present
Anne Eder, Chair
Erin Strovel, PhD
Hilary Vernon, MD, (nominee to Council)
Neal Porter, MD
Sandra Takai, MD

Staff
Johnna Watson, RN (scribe)
Linda Lammeree, RN
Angela Sittler
Tina Wiegand
Roya Alborz

Members Absent
Delegate Shirley Nathan-Pulliam
Coleen Giofredda
Caryl Siems
Anika Wilkerson

Guests
Carol Greene, MD
Ada Hamosh, MD
Ben & Kathleen Smith
Melissa Crandall
Ilise Marrazzo
Debbie Romanoski,
Senator Dyson's office
Barbara Burton, MD (phone)
Maria Escolar, MD (phone)
Mimi Blitzer, PhD (phone)
Delegate O'Donnell (phone)

Ex-Officio Present
Lee Woods, MD
Fizza Majid, PhD
Deborah Badawi, MD

Call to Order – 6:00 pm

I. Welcome and Introductions
Members and attendees introduced themselves.

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

III. Old Business

- **Update on SCID supplemental funding**
  - In Dr. Meyers absence, Dr. Majid reported that there is no new information to report on the effort to obtain this funding.

- **Review of SB433/HB891 and actions to date**
  - Dr. Badawi reported that two bills were introduced in the legislature to include screening for five lysosomal disorders, including Krabbe Leukodystrophy, to Maryland's Newborn Screening Panel.
  - Hearings have been held in both the House and the Senate but there has been no cross-over of the bills at this time.
  - Dr. Badawi reported that, as a result of a meeting that took place with Delegate Hammen, the Council will address each of the five disorders, discuss current research and literature to date, and vote on whether the disorder should be added.
  - Plan is to address one disorder at each meeting. Krabbe will be addressed this evening.

- **Newborn Screening for Krabbe Leukodystrophy**
  - Slide presentation given by Dr. Hilary Vernon (copy of slides attached)
  - Slides included current article review, telephone interview with Dr. Escolar (Pittsburgh) regarding data that is currently being prepared for publication, and telephone interview with Dr. Kurtzberg (Duke).

- **Discussion**
  - Dr. Porter pointed out that the animal studies included in the presentation show that drop off of function occurred. It is not known if and/or when human decline will occur since the population in the published studies are too young at this time.

  - Dr. Greene stated that some siblings with the same gene have different outcomes. The Federal Advisory Council determined there is not enough clinical data at this time to determine outcome of disease process at birth.

  - Outcomes of the patients seem to be better in infants diagnosed secondary to an older sibling's diagnosis or in utero because the transplant occurs earlier in life.

  - Dr. Escolar indicated that no patient is 100% normal but there are better outcomes with the new protocol that is pending publication.

  - Mrs. Smith stated that she had difficulty getting Lily in to see subspecialists after developmental changes occurred. Lily's transplantation process was completed in the interim between requesting an appointment and obtaining the appointment. She further stated that if Lily was screened at birth, her outcome would be better.

  - Dr. Badawi emphasized the importance of identifying a baby without an older sibling in a timely manner. The issue with newborn screening is turn-around time to confirm a result and complete confirmatory testing. Babies identified through newborn screening began treatment ranging from 2 weeks to 2.5 months of age.

  - Dr. Greene indicated that the major issues involve mobilization of resources. A prenatal diagnosis saves time, but babies identified by newborn screening could be transplanted by 2 weeks of age. The difficulty is determination of the onset of symptoms. There are well documented studies that show that onset of symptoms varies in families.

  - Dr. Escolar responded that, in her experience, there was one family that had cousins with variation of onset, but siblings have presented the same.

  - Dr. Greene asked Dr. Escolar if transplantation is still a clinical trial. If so, this causes a problem with insurance coverage. Dr. Escolar answered that the first transplantation protocol that was started 10 years ago is now a standard of care. The reduced toxicity regimen, that is currently being introduced, has an increased survival rate but it is currently a clinical trial.

  - Dr. Burton, a geneticist from Hunter's Hope, and a strong advocate for newborn screening for lysosomal storage disorders, gave an update on screening in Illinois. A trial will begin in May and full implementation will begin in July. Testing is to be done by MS/MS, and DNA is being sent to New York. Dr. Burton reported that Missouri's trial is 1.5 years old. This state has transplanted two children, one is a sibling of an affected child, a 2nd grader who walks with a walker. The first transplanted child did not survive.

  - Dr. Greene mentioned the possible effect of false positives on families. There should be a study on the 15-18 moderate and high-risk individuals who do not meet the criteria for transplantation.

  - Dr. Hamosh indicated that there is a 50% mortality rate for children identified through newborn screening, and treatment is not totally successful. Families with an affected child always want to know, but not all families want to know, particularly if they are at moderate risk. She described how a family brings an infant home, is notified of an abnormal screen, has all the tests done, and then is told that their baby may or may not have symptoms and is not a candidate for transplantation.

  - Mr. Smith stated this is the same situation if the family has an amniocentesis. They have to determine whether or not to take the risk.

- Dr. Escolar stated that a 50% mortality rate is not accurate because there are a smaller number of patients.

- Dr. Hamosh added that it was thought that 90% of identified Krabbe are early infantile but the 2006 data indicated that 90% are actually late onset. The Federal Advisory Council did not think it was time to implement screening for Krabbe. She stated that Maryland is not ready for implementation at this time. Screens have to be tailored to the population. For example, when screening for cystic fibrosis was started, MD had to tailor the screening to IRT/IRT because of the large African-American population in the state.

- Dr. Greene stated that those patients identified with moderate to high risk for Krabbe are "a ticking time bomb". Patients with late onset have symptoms that are more severe and rapid in onset.

- Dr. Escolar indicated that late onset can respond well to transplantation. Families are doing well watching for symptoms, and they should be able to have the choice whether or not to treat.

- Mr. Smith indicated that treatment is currently in the infantile state and will only improve. Maryland should step up and lead in the area of newborn screening.

- Delegate O'Donnell asked when the most recent clinical study will be published. Dr. Escolar responded that it is under review now and should be published in two months. Data was included in today's slide presentation.

- Anne Eder requested that the slides be distributed to the council members.

- Dr. Greene proposed testing for Krabbe only for the families who give consent. Dr. Majid indicated Maryland has never had this type of model before. Dr. Greene indicated Massachusetts had this type of model when mass spectrometry was being introduced. There would need to be a robust educational campaign for hospital staff and a separate blood spot collected if parents consented. Ilise Marrazo, of DHMH, asked if there was any way to look at the fiscal impact of the segregated testing and how the model was started in New England. Dr. Majid will obtain this information for review.

- Following a lengthy discussion, Anne Eder stated that the Council did not appear ready to vote. Additionally, there were only four of the nine voting members present, so the Council was absent a quorum. Further information is needed. Anne noted that the next meeting will be in June, but the Council may need to meet before then to continue its discussion. The Council also needs to explore the possibility of testing only those patients who have consented for the testing.

- Dr. Porter stated that he is undecided on how to vote and not sure if any data will be available in two months.

- Dr. Burton indicated that newborn screening is conducted for lots of diseases that are not curable. Some patients still die or require a lot of medical interventions. Treatment for Krabbe changes the course of the disease. Dr. Escolar stated that there is no question that treatment works.

- Delegate O'Donnell requested to be kept in the loop if another meeting is scheduled prior to June.

- Anne Eder indicated that she will keep everyone informed regarding the next meeting date. It is likely that this meeting will be June 24th.

VI. Next Meeting

- Planned for June 24, 2014

VII. Adjournment– 8:00 PM

- Meeting adjourned.

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

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)

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 Gioffreda, 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.

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.

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.