

State Advisory Council on Hereditary and Congenital Disorders

Minutes February 3, 2015

Members Present

Anne Eder, Chair
Aaron Kauffman (phone)
Hilary Vernon, MD
Ben Smith (phone)
Neil Porter, MD
Sandra Takai, MD (phone)

Members Absent

Delegate Shirley Nathan-Pulliam
Coleen Giofredda
Erin Strovel, PhD
Caryl Siems

Ex-Officio Present

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

Staff

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

Guests

John McGing
Sarah Viall (phone)
Ada Hamosh, MD (phone)
Carol Greene, MD (phone)
S. Daniel Leydorf, medical student
Sarah Viall, NP (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 January 2015 Minutes

Minutes reviewed and approved with correction to Dr. Castillo's credentials.

III. New Business

- Anne Eder reports that some members will be late to the meeting so voting for inclusion of Fabry Disease in Maryland newborn screening panel will take place after discussion of new business
- **Legislative Updates**
 - Anne Eder reports that hearing was held in House subcommittee regarding HB 5, allowing money from newborn screening fees to return directly to newborn screening instead of the general fund. Anne testified in support of the bill. Aaron Kauffman was also present at the hearing in support of the bill. Department of Budget and Management testified against the bill. Anne and Aaron both relate positive feedback regarding passage of the bill. Ben Smith also relates speaking to Delegate O'Donnell who also feels bill will pass and there is bipartisan support for it. Anne reports bill is currently filed as an emergency bill for fast enactment but she is not sure if it will stay as an emergency or not. Implementation date may be delayed if does not stay as an emergency.
 - Dr. Badawi reports on SB 156 Sickle Cell Disease Outreach Program. Bill is very broad in scope requiring DHMH to coordinate with community to provide education, social support services and any other resources need to reduce acute care services for both adults and children. DHMH is opposing secondary to scope being unclear. DHMH is in the process of writing up a fiscal note because money was not included in the bill.
- **Member Updates**
 - Laboratories Administration
 - Dr. Myers reports the lab has ordered equipment for SCID testing but not received. Pilot testing will begin after the move to the new laboratory. Building has been turned over to the lab in January. Move to new facility will be in April and May.
 - OGPSHCN

- Dr. Badawi reports OGPShCN continues to work with the laboratory to formulate follow-up protocols for SCID.
- Member Updates
 - Anne Eder reports the application for John McGing to be a member of the Council has been submitted and is pending. She also reports the House of Delegates is working on proposing a Delegate to serve on the Council. At this time there is no movement in the Senate to provide a member.
- **Strategy for New Disorders**
 - Dr. Badawi reminded the Council that the lysosomal storage disorders will be reviewed annually to determine if new information is available to evaluate whether or not they should be included in newborn screening. Typical process of nomination of condition for consideration for inclusion in newborn screening is through one of the Council members or the laboratory itself.
 - Since multiple disorders have been reviewed in the past year, Dr. Badawi asked for feedback regarding process.
 - Dr. Vernon thinks process went well.
 - Mr. Smith feels voting was based on feelings and not information. He thinks the 2009 Assessing New Criteria for Newborn Screening by Botkin should have been used in review of the disorders. Dr. Vernon indicates that she included these criteria in her presentation.
 - Dr. Takai feels presentations were excellent and there was enough time to review information prior to voting. She feels voting was based on information presented.
 - Mr. Smith thinks lysosomal disorders should be looked at individually and not lumped together. Dr. Badawi indicates the presentation of information was separate and voting occurred separately for each condition.
 - Mr. Smith feels the grading scale from Botkin's article should have been used for each disorder.
 - Dr. Greene indicates the Botkin paper is now out of date. On the Federal level, the advisory committee has made changes to how disorders are looked at for consideration.
 - Dr. Badawi states the Council could assign a grade using a scoring tool in the future. Dr. Takai requests a copy of the most recent criteria for review to be disseminated to the Council members. Mr. Kauffman requests articles in plainer language and agrees to a grading process.
 - Anne Eder states the Council will look at how decisions are made. She feels presentations were outstanding and voting has been scientifically sound.

IV. Old Business

- **Voting for Inclusion of Fabry Disease**
 - Anne Eder asks if any further discussion regarding Fabry Disease is needed. No comment from council members. Move to vote on adding Fabry Disease to the Maryland RUSP was moved and seconded.
 - Voting initiated by roll call and results noted as follows:
 - Ben Smith – yea
 - Dr. Sandra Takai – yea
 - Dr. Hilary Vernon – yea
 - Anne Eder – yea
 - Dr. Neil Porter – yea
 - Aaron Kaufman – yea
 - Inclusion of Fabry Disease in the Maryland NBS RUSP is recommended at this time by a vote of 6 to 0 and no abstentions.
 - Dr. Badawi indicates next step is to send a letter to the Secretary regarding this recommendation.
 - Mr. Smith wants to review all of the disorders again. If screening for 2 of the disorders, then all of the disorders should be screened for.
 - Dr. Badawi stated that voting was based on scientific information and available treatments for each of the disorders. Anne Eder reiterates the discussion of lysosomal disorders will be revisited on an annual basis.

V. Next Meeting

- March 10, 2015 is being held for a possible date for the next meeting in case anything comes up in the legislative session. If not needed, next meeting will be June 2nd.
- June 2, 2015
- Sept 1, 2015
- Dec 1, 2015.

VI. Adjournment

- Meeting adjourned at 6:45 PM.

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.

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

help focus discussion, identify consensus or lack of consensus, and provide helpful information if a condition is being presented for re-consideration. It may provide a visual of why we did what we did at a specific point in time.

- There is a reluctance to use cost as a consideration. Mr. McGing reminds the committee that evaluation of cost is included in the wording of the executive charge so it is appropriate to be considered. However cost of testing should not determine the decision.
 - The wording was amended to include benefits and risks to individuals and benefits and risks to family and society including ethical, legal and social implications
 - Mr. Smith made a motion to adopt workgroup recommendations with the modifications proposed tonight. Motion seconded by Mr. McGing. Called to a vote by Erin Strovel. Motion passed with 6 votes in favor and no dissenting votes.
 - Dr. Badawi stated that the criteria list and procedure will be used with X-Linked ALD, the next disorder to be presented. Adjustments can be made as necessary.
 - Dr. Badawi thanked the members of the work group for a thorough and thoughtful review and their hard work.
- **SCID screening update**
 - Dr. Majid reported that all equipment has been purchased for SCID screening. She has visited other laboratories to review various set up and testing methods. The next step is validation. Plan to begin screening for SCID by end of 2015.

IV. New Business

- **NYMAC video**

- Dr. Blitzer reported that in April/May 2015, UMMS (University of Maryland Medical System) applied for and received a small grant from NYMAC (New York- Mid-Atlantic consortium for Genetics and Newborn Screening Services) to produce a short video focused on presenting newborn screening to midwives. The video is in both English and Spanish and was written to avoid any region specific references so that the target audience could be expanded to include childbirth educators, doulas, and other healthcare workers in contact with pregnant women. Video will be shown to the Advisory Council once editing is completed.

- **Federal Update**

- **ACHDNC meeting—(Advisory Committee on Heritable Disorders in Newborns and Children)**—Dr. Badawi reported on the most recent meeting held in Washington DC on August 27-28. She attended via webinar on 8/27.
 - X linked Adrenoleukodystrophy was approved to be recommended to the Secretary for addition to the recommended uniform screening panel
 - Secretary Burwell has approved the addition of Pompe Disease to the RUSP, and acknowledges the support needed by states to begin screening.
 - A letter was sent to Secretary Burwell reporting on the ACHDNC review of policies and practices related to timeliness of newborn screening in the US which included identifying barriers, strategies and recommended specific policies and timelines related to newborn screening. It is possible grant monies will be made available.
 - NewSTEPS (Newborn Screening Technical assistance and Evaluation Program). Website is www.newsteps.org. This organization is funded by the Association of Public Health Laboratories via Health Resources and Services Administration. At the ACHDNC meeting they presented their recently compiled national database identifying state by state what is screened, fees, program demographics. They are currently developing memoranda of understanding to begin data gathering of positive cases.
 - Dr. Badawi also mentioned that there was a round table discussion that may be of interest to the council, especially since the recent work in developing criteria for conditions being considered for inclusion in MD newborn screen panel. The topic: should a lack of available therapy constitute a barrier for inclusion of a condition in NBS panel? No conclusion reached but interesting that the idea is being examined.

- **Council Handbook**

- 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.