

# State Advisory Council on Hereditary and Congenital Disorders

Minutes January 12, 2016

## **Members Present**

Anne Eder, Chair  
John McGing  
Ben Smith (phone)  
Neil Porter, MD  
Erin Strovel, PhD  
Hilary Vernon, MD  
Delegate Karen Lewis Young (phone)

## **Members Absent**

Ronald N Young  
Aaron Kauffman

## **Ex-Officio Present**

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

## **Staff**

Linda Lammeree, RN, (Scribe)  
Tina Wiegand  
Johnna Watson, RN

## **Guests**

Elizabeth Hall (phone)  
Donae Smith (phone)  
Sarah Viall, NP  
Dr. Ada Hamosh  
Dr. Carol Greene

**Called to Order** – 5:00 pm

## **I. Welcome and Introductions**

- Anne Eder suggested the opening of the meeting be delayed for a few minutes giving others affected by the traffic conditions a little more time to arrive.
- Meeting commenced at 5:10 pm
- Members and guests introduced themselves, including two candidates for membership.
  - Elizabeth Hall introduced herself as a social worker and survivor of congenital heart disease. She is very interested in supporting activities related to improving systems of care. She has submitted an application to the Governor's office to be considered for a seat on the Advisory Council as Health unrelated member.
  - Donae Allen introduced herself as a program coordinator for a non-profit agency that provides services to people with developmental disabilities. Her son has dwarfism (capitalized?) and she is interested in supporting programs and activities that promote early discovery of medical conditions. She will submit her application to the Governor's office as soon as it is completed in order to be considered for a seat on the Advisory Council as Health unrelated member.

## **II. Approval of December 1, 2015 Minutes**

Minutes reviewed and approved.

## **III. Old Business**

- Anne Eder, chair, stated that she is re-arranging the order of the agenda in order to continue addressing the topic of membership started during introductions. The vote on X-ALD will take place after membership update.
- **Membership update**
  - Anne Eder reported that Dr. Robert Bruno has submitted an application to the Governor's office to be appointed as the representative for Med Chi (the Medical and Chirurgical Faculty of the State of Maryland), seat formerly held by Dr. Takai.
  - Anne also stated that with the applications of Ms. Hall and Ms. Allen there are multiple candidates for the Health unrelated seat.
  - With the selection of the Health unrelated representative, the Advisory Council membership will be complete.

- **Vote on X-ALD**
  - Anne Eder stated that there is a quorum present at this meeting to permit a vote on whether to add x-ALD to the Maryland newborn screening panel.
  - Prior to the vote, Anne Eder wanted to initiate discussion on the first time use of the screening tool developed by the Advisory Council.
    - Dr. Badawi stated a review of score totals and per item revealed mostly consistent scoring ranging from 1275-1650 out of possible 2100. The absence of any discrepancies or wide variations, indicated the presentation by Dr. Barananos in December, 2015 sufficiently covered all the points necessary for Council members to vote.
    - Anne Eder stated she would like to see the screening tool re-designed to make it more user friendly for the non-clinical, non-medical members.
    - Anne also remarked that she would like to see the presentations follow the screening tool's lead and be more easily understood by the less medically trained members of the Council.
  - Dr. Vernon made a motion to add X-ALD to the Maryland newborn screening panel and it was seconded by John McGing.
  - Motion passed with 6 votes and 1 abstention. Anne Eder abstained from voting as the position of the March of Dimes organization, whom she represents, has not yet been determined.

#### **IV. New Business**

- **Federal Update**
  - Newborn Screening Contingency Plan
    - Dr. Badawi reported there is a plan to review and update the contingency plans developed in 2010 to help ensure continuation of operations of newborn screening labs and programs in the event of an emergency such as hurricane, etc.
    - A survey to determine the level of awareness of the contingency plan is being developed by the Maternal and Child Health program and will be sent to clinical geneticists, state laboratories and follow up programs, primary care providers, hospital administrators and nurseries and advocacy groups.
    - The goal is for feedback to be presented to the Federal level and revisions made by May, 2016.
    - In response to questioning about Maryland's contingency planning, Dr. Myers stated there are informal agreements in place to ensure the continuation of needs of newborns screening program in the event of an emergency such as hurricanes, etc. Formal mutual aid agreements between neighboring states are under discussion by Attorney General's office.
- **Council Handbook**
  - Anne Eder stated is not necessary for the Council to vote on adopting the handbook.
  - Changes to the handbook can be made as necessary and appropriate to reflect the work of the Council.
    - The timeline of relevant developments related to Maryland's newborn screening panel and program has not yet been added but is planned. Dr. Badawi thought this timeline might be formulated by working backwards using changes in regulations, proposed legislation, and correspondence.
    - Anne would like to finalize the handbook after the first of the year and requested that any further comments and suggestions be forwarded to her and Dr. Badawi.
- **Commercial Genetic Testing**
  - Anne Eder introduced a discussion about commercially available kits for genetic testing that are marketed to the general public
    - While Maryland legislation currently prohibits genetic testing kits such as "23 and Me" because there is no physician involvement in the reporting of the results, there may be legislation proposed this session to change this.
    - Dr. Myers, as laboratory administrator, voiced the following concerns:
      - whether the test has been approved by FDA as valid,
      - whether test is being performed in a CLIA (not sure what this stands for – can you spell it out?) approved lab conditions
      - historically, the legislature was concerned that people could be using unreliable and invalid test results to make health care decisions.

- some kit companies were selling DNA to researchers without disclosing this to the consumers.
  - Dr. Badawi stated in 2013 she conducted a literature review on the topic
    - Approximately 27 states permit direct to consumer genetic test kits.
    - One study found people who used the test kits are more likely to participate in health screenings.
    - However there are legitimate issues of quality control and what is the test designed for—identification of disease state vs genealogy, that have not been resolved and vary from product to product.
  - Dr. Greene stated she is aware of the explosion of interest in this topic. She mentioned that a study reported same samples sent to several different laboratories resulted in conflicting results and too much ambiguity. She expressed the personal and professional concern that people may be making irreversible decisions such as mastectomy, sterilization, pregnancy termination and suicide based on these results.
  - Council members raised the following points during discussion
    - Personal decision making vs government making decisions for individuals.
    - Role of physicians in educating and counseling their patients. What if the physician does not know what the results mean?
    - Quality control and test validity standards.
    - What disclosures are included in these test kits?
    - What is the role of Advisory Council in this matter?
  - Anne Eder stated it is obvious there are many points of discussion for this topic. It is not necessary to make any decisions right now, but that Council members should be aware of the topic and related issues.
- **Legislative Issues for 2016 Maryland General Assembly**

This current legislative session is expected to consider a record number of bills. However, only those pre-filed (prior to December 2015) are visible at this time. Dr. Woods is not aware of anything relevant at this time.
- **Member Updates**
  - **SCID testing**
    - Dr. Majid stated the lab is working towards April 2016 implementation
    - CDC training has been completed.
    - Validation is expected to begin very soon.
    - A committee of immunologists is working with lab and Follow up program to develop algorithm.
    - Anne Eder asked how the providers and parents are informed about the addition to the panel.
    - Dr. Majid stated simultaneous educational efforts through letters to providers and through the website and in conjunction with Maternal Child Health Bureau.
  - **Status of conditions approved in 2015**
    - Pompe and Fabre were recommended in 2015 to be added to the Maryland Newborn Screening panel. A letter was sent to the Secretary of Health but a response has not yet been received.
    - Anne Eder will draft and send a reminder to Secretary of Health asking when Council might expect a response.
- **Meeting Schedule:**
  - Anne Eder acknowledged that it may be necessary to meet more frequently as issues related to proposed legislature become known.
  - Next meeting is February 23, 2016.
  - Meeting for March 8 needs to be re-scheduled due to conflict with American College of Medical Genetics conference

## **VI. Adjournment**

- Meeting adjourned at 6:20 pm

# State Advisory Council on Hereditary and Congenital Disorders

Minutes April 5, 2016

## Members Present

Anne Eder, Chair  
John McGing  
Senator Ronald N Young (phone)  
Neil Porter, MD  
Erin Strovel, PhD  
Delegate Karen Lewis Young (phone)  
Dr. Richard Bruno

## Staff

Linda Lammeree, RN, (Scribe)  
Tina Wiegand

## Ex-Officio Present

Robert Myers, PhD  
Fizza Majid, PhD  
Johnna Watson, RN

## Members Absent

Ben Smith  
Hilary Vernon, MD

## Guests

Jennifer Payne  
Amanda Janson

## Called to Order – 5:10 pm

### I. Welcome and Introductions

- Members and guests introduced themselves.
- New member, Dr. Richard Bruno, introduced himself as a resident with interest in combined areas of primary care and preventive care. He fills the Med Chi vacancy.

### II. Meeting Minutes January 12, 2016

- Anne Eder, Chair, stated that voting on the minutes from the January 12th meeting would be delayed until quorum was present. Johnna Watson reported that a correction to the minutes is needed: Dr. Woods is an ex-officio member of the Advisory Council.

### III. New Business

#### A. Medical Foods

- Anne Eder introduced Jennifer Payne who detailed for the Council her experiences and difficulties obtaining insurance coverage for the medically necessary foods required to treat the genetic disorder of Phenylketonuria (PKU). These difficulties are due in part to changes in the labeling and classification of these products. Phenylketonuria, which is a condition on the newborn screening, requires a special diet.
- Several members of the Advisory Council acknowledged that they are not certain what, if any, role the Advisory Council can adopt in this issue. The members expressed appreciation to Ms. Payne for bringing the matter to their attention and enabling them to learn more about conditions identified by newborn screening that require special infant formulas and, later in life, medically necessary foods.

#### B. Membership Update

- Anne Eder stated that the terms of several Council members expire as of 06/30/2016 including:
  - ✓ Dr. Porter, representative of Monumental City Medical group who is not eligible for another term given he has served 2 terms. Johnna Watson reports a letter has been written to Monumental City Medical asking for candidates. Dr. Porter was also asked if he knew of any members of Monumental City Medical that may be interested in joining the Council. Although due to decreased membership in that group, it may be necessary to consider approaching other professional organizations dedicated to representing and advocating for the needs of underserved populations. Since this particular position is

specified in the make-up of the council, it may be necessary to change the council bylaws.

- ✓ Two health unrelated positions also expire- those currently held by John McGing and Ben Smith. Both members are eligible to apply for another term.
- ✓ Johnna Watson summarized a meeting with the Governor's appointment specialist. It was determined that the Council needs to explore ways to recruit members that will add more variety as well as geographical and ethnic diversity to the Advisory Council.
- ✓ Anne Eder stated that although her term continues, her position as Chair is expiring. This highlights the need for a Vice Chair who can move seamlessly into the position of Chair when the Chair's term expires. Currently, no one on the Council holds the Vice Chair position.
- ✓ Anne asked that members let her know if anyone is interested in assuming the roles of Chair and Vice-Chair.

#### **IV. Old Business**

##### **A. Federal Contingency Plan**

- Johnna Watson stated that the Federal contingency plan is being finalized. It is a very broad plan encouraging each state to assess barriers to practice and develop procedures to ensure continuation of work, i.e. how newborn screen specimens will be received, processed, screened, and reported in the event of significant emergency.
- Dr. Myers stated that the lab has handshake agreements with neighboring programs in the event an emergency prevents the lab from operating.

##### **B. Scoring Tool**

- Anne thanked Dr. Porter who configured the scoring tool to auto-calculate the score. This is a great time saver.
- Anne also stated that perhaps sometime in the coming year, a work group could help make the tool more "lay member" friendly.

##### **C. Legislative issues of 2016 Maryland General Assembly**

- House Bill 827- sponsored by Delegate O'Donnell- As originally introduced, would have enabled parents to request newborn screening from a laboratory other than the State laboratory. The bill was amended and passed the House and Senate. As enacted, it requires DHMH's Newborn Screening Program to notify parents/guardians of newborn infants that laboratories other than DHMH's public health laboratory are authorized to perform post-screening confirmatory or diagnostic tests on newborn infants for hereditary and congenital disorders.
- House Bill 4692- Aiden's Law- mandates state to screen for X-ALD with inclusion of time frame for implementation. This bill was introduced March 3<sup>rd</sup> in House and Senate. It was referred to House Energy and Commerce Committee and then referred to Subcommittee on Health.
- House Bill 1392 (*Medical Laboratories – Direct-to-Consumer Genetic Testing*) would have authorized laboratories to market direct-to-consumer genetic testing in Maryland. The bill was introduced several years ago and opposed by the physician community. The bill received an unfavorable vote by the House Health and Government Operations Committee.

##### **D. Member updates**

- Dr. Majid, Newborn Screen Lab Director, stated that SCID (severe combined immunodeficiency) screening went live on 04/01/2016 with three days of results obtained and reports in the process of being generated. Dr. Majid has received approval to approach UMBC for MIPAR (contractual) employees, since additional staff members are needed to complete SCID screening. Providers were notified by website and memo about addition of SCID screening to the panel.
- Maternal Child Health Bureau-NBS Follow up - Johnna Watson, Chief of NBS Follow-up Unit, reported that Dr. Badawi, who served as Medical Director for NBS Follow-up, has resigned effective February. NBS follow-up nurses are asked to contact the genetics centers should situations develop beyond scope of nursing practice. Protocols will be reviewed systematically to help reduce any ambiguities.

#### **V. 2016 Meeting Schedule**

- Next meeting dates are:  
June 7, 2016  
September 6, 2016  
December 6, 2016

#### **VI. Adjournment**

- Prior to adjournment, Anne stated that a quorum was present due to arrival of additional members after the start of the meeting. Therefore it was possible to vote on the minutes from the January meeting.
- John McGing made a motion to accept the minutes with correction. Motion seconded by Erin Strovel. Minutes accepted by vote of members present.
- Meeting adjourned at 6:30 pm.

# State Advisory Council on Hereditary and Congenital Disorders

Minutes June 7, 2016

## Members Present

Anne Eder, Chair  
John McGing  
Ben Smith ( on phone)  
Neil Porter, MD  
Erin Strovel, PhD (on phone)  
Dr. Hilary Vernon  
Dr. Richard Bruno

## Staff

Linda Lammeree, RN, (Scribe)

## Guests

Carol Greene  
Sarah Viall  
Christy Keppel, March of Dimes

## Members Absent

Senator Ronald Young  
Delegate Karen Lewis

## Ex-Officio Present

Robert Myers, PhD  
Johnna Watson, RN  
Dr. Lee Woods

Called to Order – 5:10 pm

## I. Welcome and Introductions

Members and guests introduced themselves.

## II. Minutes of Meeting April 5, 2016

Minutes were reviewed and approved.

## III. Old Business

### A. Membership Update

1. Anne Eder stated that the terms of two Health unrelated members, Ben Smith and John McGing, are expiring. Both are eligible to and have agreed to serve a second term. Their applications for re-appointment will be reviewed by the Governor's office.
2. Anne Eder also stated that there are three applications that will be presented to the Governor's office for the remaining Health Unrelated vacancy. The appointment coordinator has stated that the Governor is interested in adding diversity – geographic, ethnic, and condition related - to the Advisory Council. It may be several months before decisions are made.
3. Dr. Neil Porter was invited to continue to represent the Monumental City Medical group until another representative has been identified.
4. Anne Eder stated her term as Chair of the Advisory Council is expiring although her term as a Council member continues. There is a quorum present and Anne nominated Dr. Hilary Vernon to serve as Chair effective 07/01/2016. There were no other nominees. The motion was seconded and Dr. Vernon was elected as Chair with no opposing votes.
5. Anne Eder also discussed that a Vice Chair is needed and nominated John McGing. No other nominees were offered. The motion was seconded. John McGing was elected as Vice-Chair with no opposing votes.

### B. Update on status of Pompe, Fabry and X-ALD

1. The Advisory Council sent a letter to the Secretary of Health in January 2016 notifying the Secretary that the Advisory Council has voted to recommend inclusion of these conditions to the state newborn screening panel. To date there has been no response from the Secretary's office. Johnna Watson, Chief of Newborn Screening Follow Up Program has asked Donna Harris, Director OGPSHCN, to follow up.

2. The Council asked Dr. Myers, Laboratory Director, when testing for these conditions could commence. Dr. Myers explained that until he receives the approval from the Health Secretary, he cannot pursue a cost analysis. The cost analysis will determine if new technology and new instrumentation is needed as well as if there are issues related to lab capacity. Once the cost analysis has been completed, a time frame to implementation can be formulated.
- C. Annual Review of Lysosomal Storage Disorders
1. Anne Eder stated that it is time to re-consider several conditions previously presented to the Council for inclusion in the Newborn Screening panel. Mucopolysaccharidosis Type 1 (MPS 1), Neimann Pick, and Krabbe were not recommended at the time of initial presentation and should be reviewed to identify current research and treatment updates. After the presentations, the conditions will again be put to vote.
  2. Dr. Vernon initially presented Neimann Pick to the council on 12/09/2014 and Krabbe on 04/01/2014. She agreed to provide an update on these conditions for the September, 2016 meeting.
  3. MPS 1 was presented by Dr. Regier, CNMC Genetics in November, 2014. Sarah Viall, representing CNMC genetics, offered that Dr. Regier would be happy to provide Council with an update on MPS 1.

#### **IV: New Business**

##### **A. Federal Updates**

1. Johnna Watson stated that the Federal Advisory Committee on Heritable Disorders in Newborns and Children is planning to review two disorders.
  - a. At the May 2016 Federal Advisory Committee, the work subgroup presented a summary of GAMT (Guanidinacetate methyltransferase deficiency), a creatine disorder. The Committee did not advise forwarding the condition to evidence review because a case has not yet been identified by newborn screening. Dr. Greene stated the workgroup also identified a lack of consistency re: treatment protocol for this disorder.
  - b. Dr. Vernon suggested that Dr. Lisa Kratz, Kennedy Krieger Institute Laboratory, present GAMT to the Advisory Council. The KKI lab is one of the few in the country that tests for this condition.
  - c. Spinal Muscle Atrophy (SMA) is also being reviewed by the Federal Advisory Committee for a second time. It was suggested that the Advisory Council may have enough to consider this year with the reviews of LSD and GAMT. A presentation on SMA can be considered for 2017.
2. The Federal Advisory Committee has also discussed the issues of medical food and will be reviewing the topic in subcommittee.
  - a. In follow up to Jennifer Payne's appeal for assistance in coverage for formula, the Council discussed whether there is a role for the Advisory Council on the issue that Ms Payne presented April 5, 2016 regarding medical foods, specifically addressing the labelling and classifying of medical foods so that they are covered by insurance. Following discussion, a motion was made and seconded recommending that the Advisory Council send a letter of support to the Federal Advisory Committee in favor of supporting inclusion of medical foods at the federal level.
  - b. Prior to drafting the letter, it was suggested that the Council confirm the support of the Maryland Secretary of Health. Dr. Bruno and Mr. Smith will draft a letter to the Governor's office and present it to the Advisory Council for comments and review. Dr. Greene mentioned that it may be helpful to review the language of letters sent to HRSA on this topic.
  - c. The motion was put to a vote and passed without any opposition.

##### **B. Legislative Update**

1. There was an amendment to language of the newborn screening legislation. As enacted, it requires DHMH's Newborn Screening Program to notify parents/guardians of newborn infants that laboratories other than DHMH's public health laboratory are authorized to perform post-screening confirmatory or diagnostic tests on newborn infants for hereditary and congenital disorders. The brochures that hospitals give to parents and the Laboratories administration website are being updated. Johnna



- Watson will present the brochure draft to Council members electronically for their review and comments.
2. Legislation related to Direct to Consumer genetic testing products did not advance beyond the House committee.
  3. Aiden's Law which requires states to test for X-ALD remains in federal committee, which recesses in August.

**C. SCID**

Dr. Myers reported screening for SCID (Severe Combined Immunodeficiency) commenced April 1, 2016. No confirmed cases have been identified. There is a problem with higher than anticipated volume of abnormal results on repeat specimens. The laboratory is reviewing the 2<sup>nd</sup> month's data and will revise the protocol if appropriate. The laboratory is interviewing candidates for the two contractual scientist positions recently approved for the SCID lab.

**D. Member Updates**

Laboratory Administration had no additional updates.  
Maternal Child Health Bureau - Dr. Woods stated that there are 19 travel related cases of Zika virus in Maryland. No locally acquired cases have been identified.

**E. Other Issues**

Mr. Smith recommended that Dr. Escolar be invited to speak on Krabbe and requested that this condition be reviewed at a meeting separate from the others. Mr. Smith will aid in contacting the speaker and arranging the date with the Council Chair.  
Dr. Bruno requested copies of the presentations of the previously presented LSD conditions so that he can review the content prior to the September meeting. Anne Eder also offered to send him the scoring tool electronically.

**V. Next meeting**

1. Next meeting date –September 6, 2016.

**VI. Adjournment**

1. Meeting was adjourned at 6 pm.

# State Advisory Council on Hereditary and Congenital Disorders

Minutes October 20, 2016

## **Members Present**

Dr. Hilary Vernon, Chair  
John McGing, Vice Chair  
Rebecca Furman  
Ben Smith (phone)  
Anne Eder  
Erin Strovel, PhD (phone)  
Del Karen Lewis Young (phone)  
Dr. Richard Bruno  
Sen. Ronald Young ( phone)

## **Staff**

Linda Lammeree, RN, (Scribe)

## **Guests**

Sarah Viall, CNMC Genetics  
Adam Coleman, Ph.D, DHMH Lab  
Christy Keppel, March of Dimes

## **Members Absent**

Monumental City--vacant

## **Ex-Officio Present**

Robert Myers, PhD (phone)  
Dr. Fizza Gulamali Majid  
Johnna Watson, RN

**Called to Order** – 5:05 pm

## **I. Welcome and Introductions**

Members and guests introduced themselves.

## **II. Minutes of Meeting June 7, 2016**

Minutes were reviewed and approved.

## **III. Old Business**

### **A. Membership Update**

1. Hilary Vernon stated the applications for re-appointment of John McGing and Ben Smith are awaiting review and approval by Governor's office. Both fill Health unrelated positions on the council. Michelle Smith's application has also been forwarded to the Governor's office to fill the remaining vacant health-unrelated position.
2. It was noted that Dr. Porter, who represents Monumental City Medical Group had been invited to continue attending until a replacement representative was identified. It is not clear whether Dr. Porter is able or willing to do so. There was discussion about whether the committee can contact members of Monumental City Medical Group directly. A membership list was located for the group on line.

### **B. Update on status of Pompe, Fabry and X-ALD**

1. Secretary Mitchell sent a letter approving the addition of Pompe, Fabry and X- ALD conditions to the Maryland Newborn Screening panel.
2. Discussion ensued re: timeframe and process for implementation. Dr. Myers and Dr. Majid discussed that the next step is to perform a cost analysis and determine if an adjustment to the screening fee would be necessary. The cost analysis will determine if new technology and new instrumentation is needed as well as if there are issues related to lab capacity and personnel needs. If a lab fee increase is needed, the request would need to go through the approval process. For these reasons, it is not yet possible to determine a time frame to implementation of testing.

### **C. Review of Lysosomal Storage Disorders**

1. Dr. Vernon discussed the time table for updated presentations for Krabbe, Neimann Pick, and MPS 1 (Hurler's). She explained that Dr. Joanne Kurtzberg, a

nationally recognized expert on Krabbe Disease, was not available to present to the Council at the December 6, 2016 meeting. It is likely the presentation on Krabbe will occur in early 2017. Meeting dates need to be established for 2017 and Johnna Watson will work with Dr. Kurtzberg's scheduler to arrange the discussion on Krabbe.

2. Dr. Vernon offered to update her previous presentation on Neimann Pick disease for the next Council meeting 12/06/2016.
3. A date for a presentation on MPS 1 (Hurler's Disease) will be determined once the meeting schedule for the Advisory Council is determined.

D. Letter of Support for Medical Foods

1. Dr. Bruno reported that he and Mr. Smith are working on a draft for the letter of support to be sent to the Federal Advisory Committee encouraging inclusion of medical foods by insurance at the federal level. Once the draft is completed, it will be presented to the Advisory Council for review.
2. Christy Keppel (March of Dimes) stated that the Senate Armed Service National Appropriations Committee is reviewing a bill sponsored in part by Senator McCain including medically necessary foods for all ages for Tricare insurance, which covers military personnel. This legislation could set a precedence for private insurance companies as well.

**IV: New Business**

A. Newborn Screening regulations

1. Johnna Watson, chief, NBS Follow up Unit, presented to the Council the revised Newborn screening regulations. She summarized the following changes:
  - a. Changes in language of the regulations to permit inclusion of birth centers and other non- hospital based delivery facilities.
  - b. Severe Combined Immunodeficiency was added to the disorders list.
  - c. Removed hearing screening regulations since they are covered by another document.
  - d. Included the recent legislative amendment requiring DHMH to notify parents of newborn infants that laboratories other than DHMH state laboratory are authorized to perform post-screening confirmatory or diagnostic testing.
2. A motion was made by John McGing to accept the regulations as written and seconded by Anne Eder. The motion was accepted with no dissenting votes.

B. Birth Defects Fact Sheets

1. Johnna Watson provided the Council with several birth defects fact sheets for review. She explained that regulations require that fact sheets be sent to families of infants with certain birth defects/anomalies at six months of age. The time interval was designed to allow for death certificates to be entered into the Bureau of Vital Statistics because the Birth Defects Program is not allowed to send information to a family whose baby has died in the newborn period.
  - a. Down Syndrome (Trisomy 21) Fact Sheet
  - b. Anencephaly Fact Sheet
  - c. Holoprosencephaly Fact Sheet
  - d. Hydrocephaly Fact Sheet
  - e. Microcephaly Fact Sheet
  - f. Zika Fact Sheet
2. Dr. Vernon offered to review the sheets for accuracy from the medical point of view.
3. Several members of the Council discussed that the fact sheets should be easily understood by the lay (i.e. non medical) person. There was also discussion about the content of the sheet given that it is provided to families at six months of age. Content should be focused on information needed at 6 months of age. The Council members were asked to review these sheets and send any comments to Johnna Watson via email.
4. A brief update on the Zika virus was provided. There are 103 confirmed cases with 43 in Baltimore area. The birth registry is being set up to follow pregnant women who have tested positive for Zika. The hiring of a nurse to provide case finding and follow up has been approved. Follow up intervals have been defined to occur at 2, 6 and 12 months of age. The plan is to focus coordination activities in Prince Georges and Montgomery Counties since these counties have the largest immigration

population from countries/regions that are known to have Zika virus epidemics. Information from the birth defects surveillance will be forwarded to the CDC.

**C. Newborn Screening Brochure**

Johnna Watson reported that the updated brochure is still in internal review process and she is expecting it to be available to the Council at the next meeting. Changes include information directing families to the website, and inclusion of a statement directing families to discuss alternate testing sources for metabolic conditions not included on the current panel with their pediatrician.

**D. Member Updates**

1. Laboratory Administration –Dr. Myers stated that the lab has been engaged in preliminary cost estimates for adding screening for Pompe, Fabry and X-ALD to the panel. Now that the conditions have been approved for addition, the focus will be to refine costs by reviewing technical needs, instrumentation, staff, and supplies. The cost per test needs to be estimated and if an increase is needed, Dr. Myers will request approval. Dr. Myers does not have a time frame at this time but warned the Council that the current procurement process is protracted.
2. Maternal Child Health Bureau – Johnna Watson provided an update on the Zika grant and stated that they are currently in the process of hiring a registry coordinator and a nurse coordinator. She also stated the Oz database is currently being modified by the vendor to permit documentation of follow-up for Zika affected infants and other birth defects and to provide ease of reporting cases to the CDC.

**V. Next meeting**

Next meeting date –December 6, 2016

**VI. Adjournment**

Meeting was adjourned at 5:33 PM.

# State Advisory Council on Hereditary and Congenital Disorders

Minutes December 6, 2016

## **Members Present**

Dr. Hilary Vernon, Chair  
John McGing, Vice Chair  
Rebecca Furman (phone)  
Michelle Smith  
Anne Eder  
Erin Strovel, PhD

## **Members Absent**

Monumental City--vacant  
Ben Smith  
Dr. Richard Bruno  
Sen. Ronald Young  
Del Karen Lewis Young

## **Staff**

Linda Lammeree, RN, (Scribe)

## **Guests**

Sarah Viall, CNMC Genetics (phone)  
Christy Keppel, March of Dimes  
Dr. Carol Greene, UMMS Genetics

## **Ex-Officio Present**

Robert Myers, PhD (phone)  
Dr. Fizza Gulamali Majid (phone)  
Johnna Watson, RN

**Called to Order** – 5:10 pm

## **I. Minutes of Meeting October 20, 2016**

Minutes were reviewed and approved.

## **II. Old Business**

### **A. Membership Update**

1. Hilary Vernon welcomed new member Michelle Smith, filling Health un-related vacancy.
2. Hilary Vernon also reported that John McGing and Ben Smith were each re-approved for a second term.
3. In an effort to recruit a member from Monumental City Medical group, letters were sent to several pediatricians identified on a membership list from Monumental City Medical Group's website. To date, there have not been any replies. After discussion, it was suggested that a more personal approach might be helpful. Johnna Watson agreed to contact some of the pediatricians via phone to see if there is anyone interested in filling the vacancy on the Advisory Council.
4. In the event that no one from Monumental City Medical Group is interested in serving on the Advisory Council, it may be necessary to change the Advisory Council charter which would involve a legislative process.
5. Johnna Watson mentioned that CNMC sees many infants residing in Maryland and they have expressed interest in being on the Council if there is a health related vacancy, which would be created if Monumental City Medical Group does not have an interest. CNMC serves a population apart from the Baltimore area and so would add geographic diversity to the Council.

### **B. Medical Foods letter**

1. Dr. Bruno and Ben Smith drafted a letter urging support for medically necessary foods and formulas to be covered by insurances as an essential health benefit. Letter was submitted to Council members for review. Hilary urged edits be sent to her and she will make changes for the council to review at next meeting. Letter will be forwarded to Health Secretary with cover letter once completed. It was suggested that language in the letter be kept easy to understand since the target audience is not medically savvy.
2. Christy Kepler provided Council with update that Tricare, insurance for federal government/military, is now covering medical foods. She stated the language is broad and she will forward a copy just approved by Senate committee in case it is helpful with the medical foods letter currently under review by Council members.
3. It was suggested that the Maryland insurance commission may have a role to play in this issue. May need to be cautious about citing ACA because it may be repealed by new presidential administration.

4. Dr. Greene offered to provide additional clarification re: medical foods letter comments she submitted. She urged caution in use of certain language to avoid any suggestion that could be construed as requesting re-classification of medical foods. Reclassification of medical foods may result in removal of already limited products during what would likely be a lengthy review process.

### **III. New Business**

#### **A. Review of Lysosomal Storage Disorders**

1. Dr. Hilary Vernon, JHH, presented an updated review of Niemann Pick Disease types A/B, including recent research studies/clinical trials, newborn screening methods, and other states history with screening for this disease.
2. Discussion centered around the fact that currently there is not an available treatment although there is a promising therapy on the horizon. Question to be considered is impact of identifying a disease for which there is no treatment. It could be argued that providing patients and families with a shorter diagnostic odyssey is benefit enough for some but not all. Perhaps inclusion on the NBS panel insures continued research, leading to quicker development of a treatment.
3. Dr. Vernon suggested that council members review presentation using the council developed scoring tool and asked that they be forwarded to her. The use of the scoring tool is helpful to determine if there is consistency among members regarding the points to be considered prior to voting on inclusion in the nbs panel.
4. Dr. Majid and Dr. Myers provided general outline of factors to be considered by lab including need to select technology for testing, determine/obtain necessary equipment, obtain personnel, training. It will be necessary to determine if fee increase is needed and start that legislative process if necessary. It is likely time to testing is still years away.
5. Plan is to provide opportunity to discuss condition again at next meeting and then vote on whether to recommend inclusion in the Maryland nbs panel.

#### **B. Review of NBS Brochure**

1. Johnna Watson, chief of NBS Follow Up Unit, provided a copy of the NBS brochure to council members for their review. Please send recommendations for edits to her.
2. This brochure is intended to be presented to parents and could be disseminated by the hospital staff, OB offices, Childbirth classes, etc.
3. Suggestions generated by Council members included the need to include reference to federal resurces such as Baby's First Test. The Council questioned if there is a plan to include a QR scan code that parents could scan from phones, providing families with links to additional resources.

#### **C. Microcephaly Fact Sheet**

Dr. Greene brought to the Council's attention that there are different definitions in use for microcephaly by hospital nursery staff and provider office staff. The hospitals use head circumference below 10th percentile while post discharge providers use 5th percentile as cut off for determining microcephaly. Therefore some families receive the microcephaly fact sheet who should not be receiving the fact sheet because the fact sheets are mailed out using hospital nursery information.

### **IV. Member Updates**

#### **A. Laboratory Administration**

Dr. Myers stated that the lab has been engaged in comparison of existing technologies testing as part of determining cost estimates for adding screening for Pompe, Fabry and X-ALD to the panel. Now that the conditions have been approved for addition, the focus will be to refine costs by reviewing technical needs, instrumentation, staff, and supplies. The cost per test needs to be estimated and if an increase is needed, Dr. Myers will request approval. Dr. Myers does not have a time frame at this time but warned the Council that the current procurement process is protracted.

#### **B. Maternal Child Health Bureau**

Johnna Watson provided an update on the Zika grant and stated that the registry nurse and nurse coordinator positions have not yet been posted.

Michele Spencer has resigned as Director PHPA. Donna Gugel is the acting director PHPA and Dr. Moy (former director of Family Health Administration) was appointed acting deputy director PHPA.

#### **V. Next Meeting**

Dr. Vernon suggested meeting once a month in January, February and March, 2017 to complete the presentations of the conditions under review for inclusion to the newborn screening panel.

Council Meetings dates are as follows:

January 10, 2017,  
February 7, 2017,  
March 7, 2017 and  
May 23, 2017.

Since speakers for the remaining disorders of Krabbe, and Hurler's (MPS 1) have not yet been approached, some flexibility may be needed but all efforts will be made to accommodate these dates.

Dr. Greene mentioned she is aware of newly released data from New York's experience with Krabbe re: outcome. She will send the information to Dr. Vernon for dissemination to Council members.

#### **VI. Adjournment**

Meeting was adjourned at 6:51 PM.