State Advisory Council on Hereditary and Congenital Disorders

Minutes January 10, 2017

Members Present
Dr. Hilary Vernon, Chair
John McGing, Vice Chair
Sen. Ronald Young (phone)
Michelle Smith
Anne Eder
Erin Strovel, PhD
Dr. Richard Bruno
Del. Karen Young, (phone)

Staff
Linda Lammeree, RN, (Scribe)

Guests
Sarah Viall, CNMC Genetics (phone)
Christy Keppel, March of Dimes (phone)
Jamie Fraser,MD, CNMC Genetics (phone)
Carol Greene, MD UMMS Genetics
Ada Hamosh, MD JHH Genetics
Christine Peroutka, MD JHH Genetics

Members Absent
Monumental City--vacant
Rebecca Furman

Ex-Officio Present
Robert Myers, PhD (phone)
Lee Woods, M.D.
Fizza Gulamali Majid, PhD
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. Dr. Vernon summarized efforts to contact Monumental City Medical Group by phone, email or mailed letter over the last several months, has not yielded any response. She has concern that the organization may not be interested in continuing representation on the Advisory Council.
2. Dr. Vernon stated she has sent a letter to the organization this week, return receipt requested, advising that it is important to have full membership on the Advisory Council and with that in mind, the Advisory Council will take steps to change the executive order for the Council removing membership requirement for Monumental City Medical Group if they are unable to provide a candidate. The letter included a time frame for response in thirty days.
3. Dr. Greene volunteered to share names of several pediatricians with Johnna Watson and Dr. Vernon to see if they might be interested in membership on the Advisory Council.
4. Dr. Vernon reported that she has spoken with CNMC Genetics department. They are very interested in membership on the Advisory Council. They see many patients from the counties surrounding Washington DC area and the Eastern Shore. If Monumental City Medical Group is no longer interested in representation on the Advisory Council, CNMC Genetics department could be proposed to fill the vacancy..
5. Mr. Smith expressed interest in adding more consumer representation to the Council if the Council membership has to be restructured.

B Medical Foods letter
1. Dr. Bruno and Dr. Vernon continue to refine the letter urging support for medically necessary foods and formulas to be covered by insurances as an essential health benefit. Revised letter will be electronically sent to Council members for their review. Dr. Vernon requested suggestions for edits be sent as soon as possible. Letter will be forwarded to Health Secretary with cover letter once completed.
2. There was discussion about the role of REM (Rare and Expensive Case Management Program. Dr. Hamosh stated that many metabolic conditions are only covered until age 20, which seemed inappropriate since the medical condition continues lifelong and requires treatment from birth through death. REM participants need to meet diagnostic and financial requirements for coverage. Dr. Woods identified the new chief medical officer for Medicaid is Lisa Burgess. Dr. Hamosh offered to contact her to see how to amend REM to include medically necessary foods beyond age 20.

C. Discussion and voting for inclusion of Neimann Pick A/B

1. Dr. Vernon stated the power point presentation on Neimann-Pick disease was sent along with agenda for this meeting so the members could review prior to voting. Dr. Vernon offered to answer any questions re: Neimann Pick disease.
2. A motion was made and seconded to vote on recommending inclusion of Neimann Pick A/B Disorder to the Maryland Newborn Screening panel. A roll call vote was taken and vote tied with 4 approved, 4 opposed and Del Young, who joined call late did not vote:

   Dr. Bruno- yes
   Michelle Smith- no
   Anne Eder- no
   Erin Strovel – no
   Ben Smith – yes
   Senator Young- yes
   John McGing- yes
   Hilary Vernon- no
3. The scoring sheet was reviewed. Discussion revealed that lack of proven treatment is a concern for many. Treatment therapies are in clinical trials. Dr. Vernon stated she will contact the studies to see what the preliminary data is showing and update the Council at the next meeting.
4. Motion made and approved to table vote until next meeting

D. Member Updates

1. Dr. Myers stated that The Laboratories Administration had nothing new to report since last meeting.
2. There is a new Secretary for DHMH, Denis Schrader.
3. Dr. Woods reported that Donna Gugel is the new Director of Prevention and Health Promotion Adminstration. Recruitment efforts are underway to identify new director of Maternal Child Health Bureau.
4. Johnna Watson reported that the first case of Severe Combined Immunodeficiency was identified by newborn screening. Confirmatory testing is underway by immunologists.

III. New Business

A. Presentation of Krabbe Disease

1. Dr. Hilary Vernon, stated she will start the presentation of Krabbe Disese , last reviewed in 2014, until Dr. Kurtzberg calls in to provide treatment data.
2. There are more than 100 mutations identified in Krabbe Disease, however there are many of uncertain clinical significance. It also appears that enzyme activity nor mutation can predict severity of clinical presentation.
3. Dr. Kurtzberg presented data on her experiences at Duke University with treatment of Krabbe Disease showing 80% survival rate if treatment (bone marrow transplant) occurs prior to onset of symptoms. Her facility is no longer transplanting if infant is symptomatic although there are programs that will.
4. Dr. Vernon asked that Dr. Kurtzberg forward her slides so that they can be sent electronically to the Council members.
5. Dr. Kurtzberg's experience revealed that success of outcome is best for infants transplanted within 30 days of birth. Discussion identified that this tight time line may be difficult to meet. Questions –can lab perform sequencing or second tier testing in 36 hrs? Is the facility to perform transplant prepared to move very quickly including financial counseling for family and obtaining insurance approval?
6. Dr. Vernon will continue presentation and discussion at next meeting as this meeting was extending beyond scheduled time and some members had other commitments. Uncertain if members will feel comfortable voting on Krabbe at next meeting, scheduled for February 7, 2017.

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IV. Adjournment

Meeting adjourned at 7:20 PM.

State Advisory Council on Hereditary and Congenital Disorders

Minutes February 7, 2017

Members Present
Dr. Hilary Vernon, Chair
John McGing, Vice Chair
Sen. Ronald Young (phone)
Michelle Smith (phone)
Dr. Neil Porter (phone)
Erin Strovel, PhD
Dr. Richard Bruno
Del. Karen Young, (phone)
Ben Smith

Members Absent
Rebecca Furman

Ex-Officio Present
Dr. Robert Myers (phone)
Dr. Fizza Majid
Johnna Watson

Staff
Linda Lammeree, RN, (Scribe)

Guests
Kathleen Smith
Susan Sullivan
Jamie Fraser,MD, CNMC Genetics
Carol Greene, MD UMMS Genetics
Carolyn Jones
Mimi Blitzer, PhD (phone)
Adam Coleman, Laboratories Administration
Paul Vetter, PerkinElmer Lab (phone)
Kiley Morgart
Cassidy Stever
Heather Miller
Benjamin Miller
Melissa Shoemaker
Gene Shoemaker

Called to Order – 5:10 pm

I. Minutes of Meeting January 10, 2017
Minutes were reviewed and approved.

II. Old Business

A. Membership Update
   1. Dr. Vernon has had a response from Monumental City Medical Group. They plan to appoint a pediatrician to continue representation on the Advisory Council. In the meantime, Dr. Porter will attend meetings.

B. Medical Foods letter
   1. The original purpose of the letter to the Governor was to ask permission from the Governor for the State Advisory Council to support efforts at the federal level to provide insurance coverage for medical foods. Dr. Greene who is aware of the actions of the [Federal] Advisory Committee on Heritable Disorders in Newborns and Children work group on Medical foods will advise on the revision of the letter to the governor.
   2. While engaged in the drafting of the federal letter, it become apparent that there is a need to add language inclusive of medical foods to the insurance coverage for adults and children within the state of Maryland. Dr. Vernon will gather information on how to pursue changing DHMH regulations to address this separate but related issue of medical foods coverage.

C. Neimann Pick vote
   1. Dr. Vernon reported she has sent emails to the researchers conducting clinical trials to obtain further information re: treatment outcomes but has not had a reply. Therefore, the motion to table a vote until this information can be obtained, presented and reviewed by the Advisory Council continues.

D. Continuation of presentation on Krabbe Disease
   1. Dr. Vernon summarized the most recent study by Dr. Wasserstein on the treatment outcomes of children identified through newborn screening in New York State. The authors concluded that there are significant mortality and morbidity issues associated with the treatment and raises questions about the efficacy for infants identified through newborn screening. It further recognized that the identification of at risk children introduced unique ethical and legal issues.

2. Dr. Vernon reviewed potential new therapies/treatments on the horizon including chaperone therapy which stabilizes the enzyme, permitting it to work as it should, and also the mouse model therapy which is a combination of stem cell transplant, chaperone therapy and gene therapy.
3. Dr. Vernon reviewed criteria for inclusion on the newborn screening using several models including Wilson & Junger, Petros, and the scoring sheet developed by the Advisory Council.
4. Discussion points included recognizing validity of decreasing the diagnostic odyssey, need for published outcome data of current treatment, giving parents the choice/power to make decisions, weighing risk/benefits of monitoring identified at-risk children, role of Advisory Council per it's charter to protect from " improper treatment", role of Advisory Council making a public health decision.
5. Dr. Vernon will obtain clarifying information for the following points:
    a. contact Dr. Wasserstein to find out the outcome data of the at risk children
    b..obtain the Missouri data
    c. outcome data on the child from Kentucky that Dr. Kurtzberg transplanted
    d. psychosine data
    e. data on risks for anesthesia
    f. timing of sequencing
6. Dr. Fraser presented a brief summary of 5 children who received transplants at Children's National Medical Center. She also located publically available data through Health Resources and Services Administration (HRSA) on 21 children with Krabbe Disease who received transplants from 2010 through 2014 at 11 centers.
7. Dr. Myers and Mr. Majid discussed lab related concerns for setting up screening for Krabbe. It would likely require several hundred thousand dollars. There are two possible approaches: tandem mass spectrometry would require purchase of several more instruments or an alternate platform (Microfluidics) which may be more of a rental situation. It will also be necessary to obtain additional staff.
8. Motion to table further discussion and vote until new information available made, seconded and approved.

E. Member Updates

1. Laboratories Administration: Dr. Myers asked Dr. Majid to update the Council on the actions of the Laboratories Administration toward implementation of Fabry and Pompe screening. Quotes have been obtained and are being evaluated. Approval to hire personnel via MIPAR contracts has been obtained. Dr. Myers hopes that this will lay the groundwork for future personnel needs and reduce the very lengthy process.
2. MCHB: Johnna Watson provided brief update re: first infant confirmed to have SCID through NBS. Infant is schedule for bone marrow transplant in March.

F. Other:

1. Newborn Screening brochure: Johnna Watson reported that the newborn screening brochure continues in the revision and review process to include the language notifying parents that screening is available for genetic conditions not included on the Maryland newborn screen panel. Once finalized, the brochure will be available to families through hospitals, doctor's offices, and website.
2. Mr. Smith requested clarification of amount and use of special funding which was intended to be used for instrumentation and personnel for newborn screening. Dr. Vernon will reach legislative contacts for clarification.

G. Next meeting - scheduled for March 7.

1. It was suggested that perhaps someone from New York could present issues related to lab workflow and follow up concerns specific to Krabbe screening. Dr. Vernon will contact NY to issue an invitation.

IV. Adjournment

Meeting adjourned at 7:00 PM.

State Advisory Council on Hereditary and Congenital Disorders

Minutes March 7, 2017

Members Present
Dr. Hilary Vernon, Chair
John McGing, Vice Chair
Rebecca Furman (phone)
Michelle Smith
Dr. Neil Porter (phone)
Erin Strovel, PhD
Dr. Richard Bruno (phone)
Anne Eder (phone)
Ben Smith

Members Absent
Sen Ronald Young
Del. Karen Young

Ex-Officio Present
Dr. Robert Myers (phone)
Dr. Fizza Majid
Johnna Watson

Staff
Linda Lammeree, RN, (Scribe)

Guests
Sarah Viall, CNMC Genetics
Johns Gibson,
Jamie Fraser,MD, CNMC Genetics
Carol Greene, MD UMMS Genetics
Carolyn Jones
Mimi Blitzer, PhD (phone)
Adam Coleman, Laboratories Administration
Paul Vetter, PerkinElmer Lab (phone)
Kiley Morgart
Cassidy Stever
Arianna Francal
William Garrett
Benjamin Miller
Melissa Shoemaker

Called to Order – 5:04 pm

I. Welcome and Introductions
Mr. William Garrett was introduced as representative of Monumental City Medical Society to discuss continued interest in their Council seat.

II. Minutes of Meeting February 7, 2017
Minutes were reviewed and approved.

III. Old Business
A. Membership Update
1. Mr. Garrett spoke on behalf of Monumental City Medical Society to express their interest in continuing representation on the Advisory Committee. The representative has not yet been identified and Dr. Porter will continue to attend meetings until candidate is identified. Ms. Bernita Church will be point of contact and Mr. Garrett will provide Johnna Watson with her contact information.

B. Continuation of Discussion for Krabbe
1. Dr. Vernon addressed questions raised at previous Advisory Council meeting (02/07/2017)
    a. Status of Krabbe case in Kentucky—Dr. Vernon spoke with Chief of Pediatric genetics and metabolism at UK. Kentucky has screened approximately 58, 000 infants and has had one positive case of infantile onset Krabbe. Family self- referred to Duke University.
    b. Dr. Vernon obtained data on Krabbe screening in Missouri. She reported 2014 data available through Missouri's website stated approximately 91,000 infants screened with 2 positive cases of genotype of unknown significance. The infants have ongoing monitoring. A power point presentation by Missouri NBS program to NBSTRAN summarized 2015 data showing 8 positive cases, none of early onset. These infants also receive ongoing monitoring.
    c. Dr. Vernon looked at existing research re: effects of anesthesia since there are infants identified as at risk who require ongoing monitoring that includes the use of repeated MRI with anesthesia. There is an article in Pediatric Journal 2011 stating repeated exposure to anesthesia (such as for MRI) has cognitive consequences and appears to increase risk for learning disabilities. There was no operational definition of "repeated exposure".

d. Dr. Vernon also reported on use of psychosine as marker for identifying Krabbe disease. She spoke with Mayo Lab and with Dr. Orsini in NY, both experienced with psychosine used in conjunction with primary marker GAL C enzyme to try to reduce false positive results and decrease number of infants subjected to ongoing monitoring. There is no longitudinal data available yet as use of psychosine is too new. Each state is developing its own protocols consistent with mission. Pivotal question appears to be determining if goal of screening is to identify all cases of Krabbe or to focus on identifying early onset/infantile Krabbe. Once the program goal has been determined, issues of screening specificity and sensitivity will be addressed. It will likely be necessary to justify decisions to stakeholders including the public.

e. As requested, Dr. Vernon also enquired about the state funding of newborn screening. The special fund covers administrative costs, duties and facilities of NBS lab. Fiscal management is provided by Laboratories administration, DHMH Budget office, department of budget and Management and is subject to audits. It is understood that the revenues and expenditures are aligned. Annual budgets are prepared and reviewed on ongoing basis. Dr. Myers explained that adding conditions to the panel incurs additional expenses. Mr. Smith stated he understood there would be a surplus based on current fee. Dr. Myers explained surplus is not large enough to support new equipment and costs of adding personnel, etc. He explained NBS lab has to now pay a portion of the rental cost for the new lab which is not a state owned building. Those operating costs are still to be determined and subject to some variation.

f. After discussion, it was determined to table a vote on whether to recommend adding Krabbe to the list of disorders screened on Maryland newborn screening panel. Topic can be re-visited after additional published peer reviewed material is available for review, which may be 6 months or longer.

C. Medical Foods letter

Dr. Vernon stated she was advised to keep letter short and so it has been re-drafted to be succinct and to the point. It was suggested language be amended to represent view of Advisory Council as a whole. Final input must be received by close of business on 03/09/2017. If no further changes or edits, the council voted to send letter to the Secretary as soon as possible.

D. Newborn Screening Brochure

Johnna Watson presented edits of new brochure to include statement that testing is available outside of state laboratory for additional conditions not included on Maryland state screening. Language will include “additional conditions (supplemental screening)” to make it easier for families to search on the internet. The names of specific outside testing facilities were removed but will be available on the website.

E. Member Updates

Laboratories Administration

1. Dr. Myers reported that the lab continues the process of cost analysis for new conditions approved last summer- Pompe and Fabry. The cost analysis will then be submitted to the Budget Office. Dr. Myers stated the procurement process is long. Use of 2nd tier testing, such as molecular testing, has not been addressed at this time. The methodology has to be determined and approved first.
2. Discussion ensued re: creation of special fund for nbs generated by change in legislation that prevented newborn screening funds from being returned to general fund. Mr. Smith stated the intention was for the surplus to be available for new equipment, etc. Dr. Myers explained there is not a surplus large enough to cover costs of new testing and that increasing fees to have money available for future use/upgrades is not likely to be approved. Increasing newborn screening fee would be a political issue.
3. Mr. Smith was asked to compose list of questions geared towards how to improve use of the fund to help obtain new equipment, personnel, etc. These questions could then be discussed with Senator Young and Del Young, the Governor’s appointed members of the Advisory Council. Their expertise in legislative areas will likely be helpful.

IV Adjournment

Meeting adjourned at 6:15 PM. Next meeting is May 23, 2017. Dr. Regier of CNMC Genetics will present an update on MPS I.

State Advisory Council on Hereditary and Congenital Disorders

Minutes May 23, 2017

Members Present
Hilary Vernon, MD, Chair
John McGing, Vice Chair
Rebecca Furman (phone)
Michelle Smith
Sen Ronald Young (phone)
Erin Strovel, PhD (phone)
Del. Karen Young (phone)

Guests
Sarah Viall, CNMC Genetics
Debra Regier, MD, CNMC Genetics
Jamie Fraser,MD, CNMC Genetics (phone)
Paul Vetter, PerkinElmer Lab (phone)

Members Absent
Richard Bruno, MD
Neil Porter, MD
Ben Smith
Anne Eder

Ex-Officio Present
Robert Myers, PhD (phone)
Fizza Majid, PhD (phone)
Johnna Watson (scribe)
Lee Woods, MD

Called to Order – 5:00 pm

I. Welcome and Introductions
Roll call taken by Dr. Vernon since there were a large number of phone participants. Dr. Debra Regier was introduced to give an overview/updated presentation of Mucopolysaccharidosis (MPS-I).

II. Minutes of Meeting March 7, 2017
Minutes from March 7, 2017 were reviewed and approved.

III. New Business

- Presentation for MPS-I
  Dr. Regier presented background information regarding MPS-I, including clinical presentation and current treatment modalities. In summary, MPS-I causes accumulation of long chain sugars in the cells that results in cellular damage. Newborn screening for MPS-I was initiated to identify infants with Hurler's, the severe form of MPS-I with onset at less than 1 year of age and whose life expectancy is less than 10 years of age. Newborn screening can also identify attenuated (some residual enzyme present) MPS-I (Scheie) with onset at 3-4 years of age or in adulthood. Data for outcomes after treatment is a little limited at this time. Most treatment data is based on identification of siblings and treated the siblings earlier in life. Treatment for MPS-I is complicated and depends on age at diagnosis. If diagnosed early, hematopoietic stem cell transplant (HSCT) can be effective. HSCT performed within 3-9 months of age can slow the rate of cognitive decline. Enzyme replacement therapy (ERT) is most effective when started at an early age. ERT does not cross the blood brain barrier so no clear evidence that cognitive function will be improved. ERT can be effective in stabilization of symptoms in attenuated form. Data from states currently screening for MPS-I was also presented (slides attached). There have been pseudo deficiencies and carriers identified through newborn screening as well. Some states are using DNA sequencing as a 2nd tier test to reduce number of false positives. Environmental conditions can cause an increase in false positives if measuring enzyme levels alone. Sarah Viall reports there are a series of webinars currently being presented by APHL on MPS-I. Part III is scheduled for 5/24/2017. APHL webinars are archived and Dr. Vernon asked for links so she can send to the council members. There are multiple methods to screen for MPS-I. Dr. Vernon states MPS-I is now on RUSP as well so it has undergone a rigorous review process.
- Report from Advisory Committee on Hereditary Disorders in Newborns & Children meeting May11-12, 2017
  - Sarah Viall gave an update from the Federal ACHDNC meeting.
  - The members voted to move Spinal Muscular Atrophy (SMA) from nomination stage to review stage. Review will be performed over the next 9 months and then findings will be presented to the full committee for consideration for addition of SMA to the RUSP.
  - The committee is working on a white paper in support of insurance coverage for medical foods. Sarah also reports that she was on Capitol Hill with National PKU Alliance on

5/22/2017. There is bipartisan support for HB 2587 and SB 1194, Medical Food and Formula Equity Act introduced by Senator Grassley from Iowa. Bill requires insurances to cover medical foods for life time for individuals with identified disorders. Sarah recommends contacting senators to recommend support for these bills.

- Dr. Vernon reports medical foods letter was sent to Governor's office in March. No response at this time. She suggests sending a copy of the letter to Senators and Representatives and referencing HB 2587 and SB 1194 to garner support for these bills.

IV. Old Business

- Update from Labs Administration
  - Dr. Myers reports there has been some progress in screening for lysosomal storage disorders Pompe and Fabry. Identification of method has been determined and procurement has been started. Validation of instrumentation and testing method is expected over summer months with implementation possibly as early as late Fall of 2017 or early next year. Dr. Myers also reports MIPAR contracts are being renegotiated at this time so unable to start hiring process for staff needed for implementation.
  - Dr. Vernon asked about chosen methodology for lysosomal disorders, and Dr. Majid reports digital microfluidics has been selected.
- Update from MCHB
  - Dr. Woods reports there is a new MCHB Director. Courtney Lewis will be starting on June 7th. She has been with DHMH previously and comes back after a period of time with Washington DC Health Department.
- Update on Krabbe
  - Dr. Vernon reports there has been no new information to date on the use of psychosine as a testing method for Krabbe. She will continue to monitor for this information.

V. Next Meeting Dates

June 20, 2017 was selected as the next meeting date.
No meetings will be planned for summer.
Council members were also asked to hold September 19, 2017 open as a next meeting date.

VI. Adjournment

Meeting adjourned at 5:45 PM.

State Advisory Council on Hereditary and Congenital Disorders

Minutes June 20, 2017

Members Present
Hilary Vernon, MD, Chair
John McGing, Vice Chair
Rebecca Furman (phone)
Michelle Smith
Ben Smith
Sen Ronald Young (phone)
Erin Strovel, PhD (phone)
Del. Karen Young (phone)

Staff
Linda Lammeree, (scribe)

Guests

Sarah Viall, CNMC Genetics (phone)
Debra Regier, MD, CNMC Genetics (phone)
Jamie Fraser,MD, CNMC Genetics (phone)
Christine Keppell, March of Dimes (phone)

Members Absent
Richard Bruno, MD
Neil Porter, MD
Anne Eder

Ex-Officio Present
Robert Myers, PhD (phone)
Fizza Majid, PhD
Johnna Watson

Called to Order – 5:08 pm

I. Welcome and Introductions
Dr. Vernon suggested changing order of agenda to provide ample time for Council members to arrive or join by phone prior to considering voting. She will provide summary of use of Psychosine as a marker for Krabbe disease first and then open discussion and/or vote for inclusion of MPS 1.

II. Minutes of Meeting May 23, 2017
Minutes from May 23, 2017 were reviewed and approved.

III. Old Business

- Discussion of paper on Psychosine and Krabbe disease.
    1. Dr. Vernon reviewed paper currently available on line from Molecular Genetics and Metabolism but not yet in print. Dr. Vernon will make copies of power point presentation available to Council for their review.
    2. Psychosine is an additional biomarker for Krabbe Disease. The level of psychosine accumulates in patients with Krabbe Disease, however there is no correlation with the GALC enzyme level.
    3. Goals of the study:
        A. to determine if the psychosine concentration in the dried blood spot at birth is predictive of disease onset and clinical phenotype
        B. Assess relationship between disease progression and longitudinal changes in dbs psychosine concentration in patient with Krabbe Disease
        C. Evaluate the effects of treatment with HSCT on DBS psychosine concentration.
    4. Findings/conclusions:
        A. Likely psychosine levels > 3 nmol/L correlate to high probability of developing infantile KD.
        B. Determining outcome/clinical significance of mildly elevated psychosine (0.7-3 nmol/L) levels is uncertain and requires more longitudinal studies.
        C. Psychosine may increase with age in untreated patients and then, at a certain point decline in advanced disease state.
        D. There is an approximate 8.6 % standard error- an indication of some variability in psychosine measurements from the same DBS. May need to use multiple punches per patient when analyzing psychosine concentrations.
        E. More research is needed to determine whether psychosine levels a baseline or after HSCT correlate with clinical outcomes.
    5. Dr. Vernon suggested Council review the report and email her any questions. Report can be discussed at next meeting in September.
    6. Mr. Smith mentioned that Dr. Kurtzberg is planning to release information on same topic soon. Dr. Vernon stated the Council will review published data when it is available.
- Discussion/voting for inclusion of MPS 1 on NBS panel

1. Dr. Vernon asked if there were any questions re: MPS 1 presentation 05/23/2017 by Dr. Rieger. Dr. Rieger is present via phone to answer questions if needed.
2. Motion made and seconded to vote on adding MPS1 to Maryland NBS panel.
3. Quorum was met since 8 voting members were present to vote. Vote results: 8 votes approving inclusion of MPS 1 on Maryland NBS panel. No dissenting votes.
4. Dr. Vernon stated she will draft the letter to the Secretary of Health recommending inclusion of MPS 1 to Maryland NBS panel.

- Medical Foods letter
  1. Dr. Vernon stated she has not received any response to the letter sent to the Governor's office.
  2. Sarah Viall, CNMC Genetics, mentioned there has not been any action on the Federal level either and suspects it will be a long process.

IV. Member Updates

- Update from Labs Administration
  1. Dr. Myers reported the Laboratories Administration continues to actively pursue resolution of MIPAR contracts so that staff for the approved lysosomal storage disorders may be hired. Laboratory equipment and reagents in procurement stage.
  2. Dr. Myers will need to confirm with Dr. Majid if MPS 1 testing can be performed on same microfluidics platform.

- Update from MCHB
  1. Johnna Watson reported that Donna Harris, Director for Office for Genetics and People with Special Healthcare Needs has retired. Dr. Jed Miller is acting interim director.

V. Next Meeting Dates

- Confirmed next meeting will be September 19, 2017.
- Future agenda items include:
  1. Presentation on Spinal Muscular Atrophy. Dr. Vernon will ask Dr. Crawford to present.
  2. Guanidinoacetate Methyltransferase presentation to be considered for future agenda topic.
  3. Need to re-address Krabbe disease this fall/winter and will re-address tabled Neiman Pick disease in the spring, 2018.

VI. Adjournment

Meeting adjourned at 5:35 PM.

State Advisory Council on Hereditary and Congenital Disorders

Minutes October 24, 2017

Members Present
Hilary Vernon, MD, Chair
John McGing, Vice Chair
Rebecca Furman (phone)
Michelle Smith
David Myles, MD (phone)
Sen Ronald Young (phone)
Erin Strovel, PhD
Del. Karen Lewis-Young (phone)

Ex-Officio Present
Robert Myers, PhD (phone)
Fizza Majid, Ph D
Johnna Watson

Members Absent
Ben Smith
Neil Porter, MD
Anne Eder

Staff
Linda Lammeree, (scribe)

Guests
Adam Coleman, Lab Admin (phone)
Sarah Viall, CNMC Genetics
Ada Hamosh, MD, JHH Genetics (phone)
Carol Greene, MD, UM Genetics (phone)
Christine Keppell, March of Dimes
Kim Heinrich
Kelly Eakin
Brandi Akins
Mana Spencer, Cure SMA
John Gibson, Biogen
Paul Vetter, PerkinElmer Genetics
Carolyn Jones, Biogen

Called to Order – 5:07 pm

I. Welcome and Introductions

- Dr. Vernon introduced Dr. David Myles, newly appointed MedChi representative.

II. Minutes of Meeting May 23, 2017

- Minutes from June 20, 2017 were reviewed and approved.

III. New Business

- Presentation on Spinal Muscular Atrophy (SMA).
  1. Dr. Thomas Crawford, professor and neurologist at JHH provided power point presentation on SMA including description of disorder, diagnosis, testing and treatment. The slides will be available electronically.
  2. Additional speakers: Kim Heinreich, Kelly Eakin, and Brandi Aikens, parents of children diagnosed with SMA, each spoke about their experiences to urge Council to include SMA on Maryland newborn screening panel.
  3. Dr. Vernon outlined the process for the Council when considering new conditions for inclusion. Following a presentation for the condition under consideration, there is a discussion and opportunity to ask questions at the next Council meeting. Following discussion, a vote may be considered. That recommendation is then forwarded via letter to the Maryland Health Secretary.
  4. Dr. Majid, Laboratories Administration, Director of NBS Lab, confirmed that a pilot program is not required. The testing for SMA will be multiplexed with TREC testing (same platform used for SCID screening).
  5. Maryland NBS does follow the Recommended Uniform Screening Panel (RUSP) but can add conditions to the Maryland panel independent of the RUSP. Ms Spencer, Cure SMA, stated an application has been made to the Federal Advisory Committee on Heritable Disorders in Newborns and Children to include SMA but a decision has not yet been determined. Ms. Spencer will provide a copy of the application to the Advisory Council and will make someone available for the next meeting in the event there are questions generated by the application material.
  6. Dr. Vernon asked that any questions re: SMA presentation be emailed to her as soon as possible. She will consolidate and forward questions to Dr. Crawford.

IV. Member Updates

- Update from Labs Administration

  Dr. Myers reported there has been some progress in obtaining equipment and reagents for the lysosomal storage disorders, especially Fabry and Pompe. There has been a major issue obtaining personnel as recently lost approval to contract personnel through UMBC. Dr. Myers stated this decision is being appealed.

- Update from MCHB

  Johnna Watson reported that letter to the Secretary for inclusion of MPS 1 has been drafted and is awaiting decision about letterhead. Once that is determined, the letter will be forwarded to the Health Secretary.

V. Next Meeting Dates

- Next meeting will be November 28, 2017.
- Dr. Vernon states voting on Krabbe disorder will proceed after reviewing Krabbe materials as a group and to provide the new Council member, Dr. Myles, an opportunity to digest the information and to ask questions prior to voting.

VI. Adjournment

Meeting adjourned at 6:07 PM.

State Advisory Council on Hereditary and Congenital Disorders

Minutes November 28, 2017

Members Present
Hilary Vernon, MD, Chair
John McGing, Vice Chair
Anne Eder (phone)
Del. Karen Lewis-Young (phone)
Sen Ronald Young (phone)
Erin Strovel, PhD

Ex-Officio Present
Robert Myers, PhD (phone)
Fizza Majid, Ph D
Johnna Watson

Staff
Linda Lammeree, (scribe)

Guests
Lisa Kratz, KKI
Sarah Viall, CNMC Genetics (phone)
Dr. Jamie Fraser, CNMC Genetics (phone)
Carol Greene, MD, UM Genetics (phone)
Maria Spencer, Cure SMA
Jackie Glascock, Cure SMA
John Gibson, Biogen
Paul Vetter, PerkinElmer Genetics
Carolyn Jones, Biogen

Members Absent
Ben Smith
Neil Porter, MD
Rebecca Furman
David Myles, MD
Michelle Smith

Called to Order – 5:05 pm

I. Welcome and Introductions
- All attendees introduced themselves.
- Discussion to determine if a quorum was present to vote on meeting minutes and inclusion of SMA (Spinal Muscular Atrophy) was resolved with delayed arrival of a Council member.

II. Minutes of Meeting
- Minutes from October 24, 2017 were reviewed and approved.

III. Old Business
- Discussion of Spinal Muscular Atrophy (SMA) presentation.
  1. Dr. Vernon provided an opportunity for questions and discussion as follow up to the presentation on SMA by Thomas Crawford at last Council meeting 10-24-2017.
  2. There was discussion re: use of the Council created scoring tool. Dr. Vernon agreed the scoring tool may be of help with the thought process when considering conditions for inclusion on the newborn screening panel. She will review and provide copies of the tool for future use.
  3. A motion was made and seconded to vote on recommending inclusion of SMA on Maryland newborn screening panel.
  4. A roll call vote was taken and showed unanimous decision to recommend addition of SMA to the Maryland newborn screening panel.
  5. Dr. Vernon stated she will submit letter from the Advisory Council to the Maryland Secretary of Health recommending inclusion of SMA to Maryland newborn screening panel.

IV. New Business
- Presentation on Guadinacetate N-Methyltransferase (GAMT) and creatine deficiencies
  1. Dr. Lisa Kratz, Director of Biochemical Genetics Lab at Kennedy Krieger Institute provided a power point presentation on GAMT, including description of disorder, diagnosis, testing, treatment and review of literature: re use in newborn screening. The slides will be made available electronically.

IV. Member Updates

- Update from Labs Administration
  1. Dr. Myers reported he has received approval to hire two contractual laboratory staff for the screening of the lysosomal storage disorders. The Laboratories Administration continues to work through the procurement process for equipment and reagents.

V. Next Meeting Dates

- Dr. Vernon stated voting on Krabbe disorder will proceed after reviewing Krabbe materials as a group and to provide the new Council member, Dr. Myles, an opportunity to digest the information and to ask questions prior to voting. Two consecutive meetings will be devoted to review and vote on Krabbe Disease.
- A doodle poll will be sent out to help determine date of next meeting with tentative date of January 4, 2018. Del and Sen Young reminded Council that the Maryland Legislative session begins in January 10, 2018 and continues for 90 days, making it difficult for them to commit to Council meetings even by phone. Dr. Vernon stated all efforts will be made to encourage Council members to attend these next two meetings to address and resolve consideration of inclusion of Krabbe Disease on the Maryland newborn screening panel.

VI. Adjournment

Meeting adjourned at 5:48 PM.