State Advisory Council on Hereditary and Congenital Disorders

Minutes January 4, 2018

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
Michelle Smith (phone)
David Myles, MD (phone)

DHMH Staff
Adam Coleman, PhD
Jed Miller, MD

Guests
Mimi Blitzer, UM Genetics (phone)
Sarah Viall, CNMC Genetics (phone)
Christie Keppel, March of Dimes (phone)
Carol Greene, MD, UM Genetics (phone)
Dr. Jamie Fraser, CNMC Genetics (phone)

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

Members Absent
Ben Smith
Neil Porter, MD
Rebecca Furman

Called to Order – 5:05 pm

I. Welcome and Introductions

- All attendees introduced themselves.
- Dr. Jed Miller introduced himself as the newly appointed Director of the Office of Genetics and People with Special Health Care Needs.

II. Minutes of Meeting

- Minutes from November 28, 2017 were reviewed and approved.

III. Old Business

- Review of Krabbe Leukodystrphy
  Dr. Vernon provided a slide presentation on Krabbe Leukodystrophy, providing the latest published reports on newborn screening for Krabbe. Slide show is attached for review. Scoring sheets were also discussed to be used as a way to organize thoughts. A copy of the scoring sheet will be distributed to members as well
- Discussion regarding Krabbe Leukodystrophy
  1. John McGing expressed concern regarding the impact on families for babies who are labeled as high risk but are not identified as having the disease, particularly concerned over serial MRI and LPs that would be required to follow these babies long term. He asked if diagnostic options are moving away from such invasive procedures.
  2. Dr. Vernon stated that what she thinks is on the verge of improving is the screening process to help reduce the number of babies who screen positive, and screen out those infants who are less likely to develop the disease. If the screening process improves, then less babies may be identified that would have to go through the invasive procedures, but there is nothing published at this time.
  3. Dr. Jamie Fraser, CNMC Genetics, states she has attended two separate meetings this past year discussing this issue. She states the psychosine data is looking better as a secondary marker to help stratify risk for disease, but providers are not comfortable relying on quantitative psychosine at this time until more data is published. CNMC Genetics has recently hired a physician from New York who saw 5 patients that screened moderate to high risk and ultimately they all turned out to be carriers. However, each of these patients was followed with serial LPs

and MRIs while going through this diagnostic odyssey to find out they were carriers. Dr. Fraser further states the community of physicians providing care for these babies feels the risk of having a high false positive rate with screening is high at this time.

4. John McGing further expresses that the impact on the babies and families regarding the serial LPs and MRIs makes him want to slow down at this time. Dr. Fraser agreed that not enough progress has been made at this time to allay his concerns.

6. Dr. Vernon will make all of the slides available with resources for review.

7. Michelle Smith asked how many times has Krabbe come up for discussion. Dr. Vernon indicated this is the 3rd time she has presented Krabbe. It has been reviewed upon request.

8. John McGing brought up the discussion of either taking a vote tonight on Krabbe or plan to vote at next meeting which may not be until April after the legislative session is over to allow the two legislators to vote.

9. Dr. Fraser interjected to add the comment that when States add screening for conditions that are not on the RUSP available information to provide physicians is not standardized as it is for conditions on the RUSP. American College of Medical Geneticist ACT sheets are not available to be used as a resource to providers to help them discuss the condition with families or to help with determining next steps for diagnosis. She states this is another consideration that should be taken into account

10. John McGing asked again what the members felt about voting tonight or waiting to vote. Michelle Smith states she does not need to wait to vote, she can vote today. Anne Eder asked to have more time to review the presentation in greater detail and then vote. Dr. Vernon states the Council will follow the usual procedure and the vote will need to be held at an upcoming meeting.

11. John McGing stated the vote will be placed on the agenda at the next meeting. John asked the legislators when they will be available. Delegate Karen Lewis-Young states it is difficult to know when they might be available but may have more flexibility at the end of January or the beginning of February. John proposed meeting after April 9th. Dr. Vernon proposed a phone call only meeting on a Sunday night. Dr. Jed Miller states this is a public meeting so the meeting cannot be exclusively by phone. Senator Young states they could meet on a Monday at 5 PM because session does not start until 8 PM and committee meetings are rarely scheduled on a Monday. Mondays work for all members present except for Dr. David Myles who states the vote should not be held up on his account. Dr. Vernon states a Doodle poll will be sent out to see if a quorum can be obtained on a Monday in the next 3-5 weeks. Presentation to be distributed and any questions should be addressed to Dr. Vernon in preparation for the meeting.

IV. New Business

Delegate Lewis-Young raised a concern about a bill introduced last session requiring opiod testing on newborns. The bill was voted down. Maryland has a $700,000 Federal grant that is in danger of being lost if newborns are not followed up for testing positive tested for opiods and other substances. She has discussed this issue with DHMH and was told this issue was not appropriate for newborn screening since it is not an inherited condition. She would like to share the information with the Council to review and determine if this is an issue that falls under the realm of this Council. Christie Keppel states she would be glad to talk to Delegate Lewis-Young about what other states are doing on this issue. Dr. Vernon states that if this discussion does not help with the issue then the information related to the bill should be forwarded to the Council for review. If the Council is not appropriate to address the issue, the Council may be able to determine the appropriate avenue for this discussion

VI. Adjournment

Meeting adjourned at 6:30 PM.

State Advisory Council on Hereditary and Congenital Disorders

Minutes April 17, 2018

Members Present
Hilary Vernon, MD, Chair
John McGing, Vice Chair (phone)
Erin Strovel, PhD
Ben Smith
David Myles, MD (phone)
Michelle Smith
Rebecca Furman (phone)

Members Absent
Anne Eder
Neil Porter, MD
Delegate Karen Young
Senator Ronald Young

Ex-Officio Present
Robert Myers, PhD
Fizza Majid, PhD
Johnna Watson
Lee Woods, MD

DHMH Staff
Adam Coleman, PhD
Jed Miller, MD
Linda Lammeree (Scribe)

Guests
John Gibson, Biogen (phone)
Sarah Viall, CNMC Genetics (phone)
Kendra Sullivan (phone)
Carol Greene, MD, UM Genetics (phone)
Kathleen Smith
Jamie Fraser, MD, CNMC Genetics (phone)
Arianna Guillard
Ryan Kuehl
Doug Loesch
Shayad Kheradmand
Emma Perez
Brittany Allen
Paige Babb
Becky Sheedy
Ashlee Vargason
Hadley Bryan

Called to Order – 5:12 pm

I. Welcome and Introductions
- Quorum not present at beginning of meeting so meeting delayed several minutes to give members more time to arrive.
- Dr. Vernon reviewed old business items after calling meeting to order to give more time for members to arrive so that items requiring a vote might proceed.
- Agenda items will be covered but not necessarily in order.

II. Minutes of Meeting
- Initially, due to lack of quorum, minutes for meeting January 4, 2018 were not proposed for review and approval.
- Minutes of Advisory Council meeting January 4, 2018 were reviewed and approved once quorum present.

III. Old Business
- Discussion of Krabbe Leukodystrphy
    1. Mrs. Smith summarized the experience her family has had getting her daughter diagnosed and treated. She is advocating Krabbe be added to the Maryland newborn screening panel to ultimately improve the outcomes for others.
    2. Dr. Fraser expressed her concern as a provider of care that there continues to be a lack of specific guidelines and insufficient data re: treatment outcomes. Due to rate of false positives, at risk, and inconclusive results, many infants and children are subjected to invasive procedures for years. Current treatment available can be considered by some to be experimental as the vast majority of medical providers say treatment has not become standard of care as there is too little data.
    3. Dr. Greene clarified use of cure vs treatment language, stating that conditions on the newborn screening panel can be treated but not cured. Dr. Greene stated she recently attended professional conferences hosted by SIMD and ACMG. Her sense is that families are deeply appreciative of screening while providers are deeply

unhappy due to the false positives and at risk patients that require years of invasive monitoring.

4. Mrs. Smith stated that without newborn screening to help identify these infants early there will not be significant progress made in improving the screening methods or treatment because the number of those diagnosed with Krabbe is so few.
5. Sarah Viall expressed that the families' experiences and points of view are so very important to this process. As a clinician, clearer guidelines are needed.
6. John McGing stated his concern about the impact on the families of infants identified as at risk and the risks of monitoring protocols which are invasive and require sedation.
7. Dr. Frasier states the use of second tier testing such as pyschosine can help reduce these numbers infants identified as at risk but it is still far from perfect. The purpose of newborn screening is not to provide subjects for research.
8. Michelle Smith asked if treatment would be covered by insurance. In Maryland REM covers Krabbe for those families that meet the financial requirements. Dr Greene stated some of the molecular diagnostic testing would not be covered. This would impact infants with at risk results or inconclusive findings.
9. Ben Smith stated approximately 12 states have had legislation passed requiring newborn screening for Krabbe and approximately 8 states are testing. Mr. Smith stated if approved, Maryland would be the first state to start testing for Krabbe based on Advisory Council recommendation.

- Vote on Krabbe
    1. A motion was made to recommend adding Krabbe Leukodystrophy to the Maryland newborn screening panel and seconded.
    2. A roll call vote was taken.
        a. Ben Smith and Dr Myles voted in favor of recommending Krabbe be added to the Maryland newborn screening panel.
        b. Michelle Smith, Erin Strovel, John McGing, Dr Hilary Vernon voted against recommending Krabbe be added to the Maryland newborn screening panel.
    3. By a vote of 4 against and 2 in favor, Krabbe Leukodystrophy will not be recommended to be added to the Maryland newborn screening panel at this time.

IV. New Business

- Discussion of Maryland Medicaid coverage for pediatric genetic testing
    1. Dr. Vernon stated that Maryland Medicaid is not covering a standard first line test called SNP array that is important to help diagnose and treat children with developmental and physical concerns. In addition, some of the key codes for pediatric genetic counseling, prenatal genetic counseling and cancer genetic counseling are being denied.
    2. Dr. Greene stated it is a standard cytogenetic test and there really is not another option with same specificity. Medicaid simply provides statement that claim is denied, not a covered benefit. Dr. Greene stated she has discussed this with Dr. Hamosh, JHH geneticist, who concurs this testing and counseling is crucial. There is some concern that the denial could be viewed as discriminatory given that the ethnicities more likely to use Medicaid are minorities.
    3. Dr. Vernon proposed the Advisory Council send an opinion letter to Maryland Medicaid. Mr. Smith volunteered to help. Dr. Vernon planned to get outline to Council by May 1, 2018.

- Discussion re: opiod testing on newborns
    1. There is no update at this time. Christy Keppel (March of Dimes) was to provide more information to Delegate Karen Lewis-Young outside of the Council, but she is no longer employed with March of Dimes, and Delegate Lewis-Young was unable to attend this meeting.
    2. Dr. Vernon stated she will have to look into this topic and get back to the Council at next meeting.

V. Member Updates

- Laboratory Administration

1. Dr. Majid, Director of Newborn Screening Lab, reported that procurement and personnel efforts continue but that so far there is not a date for testing for the new conditions to begin.
2. Personnel requests are awaiting Budget office approval. Once approved and staffed, the lab needs approximately 4-5 months to start testing for the lysosomal storage disorders of Pompe and Fabry. Dr. Majid stated testing for X-linked ALD will take longer to implement.

- MCHB
    1. Dr. Jed Miller, Director of Office for Genetics and People with Special Healthcare Needs, provided update on SB105 that changes the membership of the Advisory Council and the appointment process. He explained the bill was passed, and at time of meeting, was awaiting Governor's signature. It is expected to take effect July 1, 2018.
        a. SB105 removed a member seat for an agency that is no longer in existence and also removed the seat formerly assigned to Monumental Medical Society.
        b. The bill created a seat for Children's National Medical Center (CNMC), recognizing their role and expertise in addressing the needs of children in the State of Maryland.
        c. All members of the Advisory Council, both voting and non-voting, will be appointed by the Maryland Secretary of Health.
        d. Current terms will not be affected although adding CNMC as a voting member of the Council will take effect 07/01/2018.
    2. Johnna Watson, Chief of Newborn Screening Follow up Unit, reported the (Federal) Advisory Committee on Heritable Disorders in Newborns and Children just approved the addition of Spinal Muscular Atrophy (SMA) to the recommended uniform screening panel. The Maryland Advisory Council sent a letter to the Maryland Secretary of Health in March 2018 recommending SMA be included on the Maryland newborn screening panel. It is too soon for a response.

VI. Next meeting

- A doodle poll will be sent out to set next meeting date.
- Johnna Watson stated that the Advisory Council meetings need to be posted in the Maryland Register, so more lead time might be needed to meet this requirement. Meeting will be scheduled for some time in June 2018.
- Dr. Vernon stated John McGing will begin as new chair 07/01/2018.

VII. Adjournment

Meeting adjourned at 6:14 PM.

State Advisory Council on Hereditary and Congenital Disorders

Minutes October 9, 2018

Members Present
John McGing, Chair
Hilary Vernon (phone)
Erin Strovel
David Myles (phone)
Anne Eder
Michelle Smith (phone)
Sarah Viall
Ben Smith (phone)

Members Absent
Delegate Karen Young
Senator Ronald Young

MDH Staff
Jed Miller

Ex-Officio Present
Fizza Majid
Johnna Watson (scribe)
Robert Myers

Guests
Paul Vetter, (phone)
Mimi Blitzer (phone)
Richard Jones
Carol Greene
Ann Moser

Called to Order – 5:10 pm

I. Welcome and Introductions

Sarah Viall is welcomed as newest member of the Council. Ann Moser and Richard Jones are present to discuss the status of X-ALD screening in Maryland.

II. Approval of Minutes

Minutes from meeting on June 12, 2018 were approved with the following changes:

- DHMH changed to MDH
- Under the heading Meeting Schedule, "legislation" and "Executive order" to be changed to "statute".

III. Old Business

Status of addition of new disorders

- Dr. Myers reports that the current plan is to be on-line for the 3 lysosomal disorders (LSDs) and spinal muscular atrophy (SMA) either by the 1st of January or the 1st of February of 2019. There have been significant issues delaying implementation secondary to staff turnover, but after a year long process, two new contractual staff have been hired. The LSDs using the Seeker system should be on-line by January 1st and SMA, which will be integrated into the molecular program currently performing SCID screening, should be on-line by February 1st.
- For X-ALD there has been a preliminary cost analysis, and the lab thinks it can be done with about $8 a test. There is a surplus currently since LSD and SMA but the screening have not been implemented yet. After the first quarter of the year, the lab will have a better idea as to whether or not a fee increase will be needed to implement X-ALD and to keep it sustainable into the future.
- John McGing asked if a fee increase requires a change in regulation or legislation. Dr. Myers indicates that a fee increase does require a regulation change and support from MDH. An emergency request for regulation change took about 3-4 months for the last increase. The lab needs to justify the reason for the increase, if needed. The lab also needs to be cautious in moving forward with screening and incurring a deficit in the budget which would be difficult to cover. Dr. Majid has done some initial cost analysis with PerkinElmer Genetics for reagent rental that does not require a capital investment. The reagent rental will still need to go through a procurement process.
- Dr. Carol Greene asked why the lab does not just go ahead and ask for the fee increase since $8 a baby would be difficult to cover with the current fee. Dr. Myers states there is a surplus in the budget currently, due to the delay in implementing the 4 new disorders and due to military

contract providing additional income. Therefore, the lab has to see how the books balance out once LSDs and SMA screening is started. The lab cannot ask for a fee increase and create an even larger surplus. The lab is starting screening for LSDs and SMA without a fee increase, but it is unknown if they can start X-ALD without a fee increase at this time.

- Dr. Richard Jones asked what percentage of the $8 estimate is based on the cost of the kit. Dr. Majid states there is no kit at this point. The plan is to obtain the reagents from PerkinElmer and have an in-house procedure. Dr. Myers adds that staffing needs are not completely determined yet for X-ALD. The lab may be able to move people around since X-ALD will be mass spec or they may need to hire personnel. The LSDs, since it is a different platform, requires 2 dedicated staff members. SMA will be performed by staff currently performing SCID screening so no new staff is needed for SMA.
- Johnna Watson asked what follow-up plans have been determined by the genetic centers. Dr. Majid states they will have a committee of all of the genetic centers involved to set up a protocol to respond to the new disorders on the screen. Sarah Viall states Children's already has protocols in place because DC has started screening for MPS-I and Pompe. She will share these protocols. Dr. Majid will send out a Doodle poll to determine date and time for meeting.
- John McGing provided a summary at the end of the discussion. Recapping that screening for LSDs and SMA is planned to be on line in January/February 2019 and the 5th disorder, X-ALD, needs more budgetary analysis prior to implementation.
- Dr. Ann Moser offered her assistance in setting up screening for X-ALD and KKI will be available for any follow-up testing that is needed. She also indicated Dr. Ali Fatemi at Hopkins is also running a clinic seeing patients with X-ALD so he is available for consult as well.
- Dr. Greene asked if diagnostic testing can be sent to KKI regardless of insurance coverage. Dr. Jones states the testing can be done through the grant KKI has with the State of Maryland to cover costs of the testing if needed.
- Dr. Greene states that the Society for Inherited Metabolic Disorders meeting will be happening during the next scheduled meeting time of April 9th. Sarah Viall states the National Newborn Screening Symposium is April 7th-10th, as well. John McGing indicates we will reschedule the next meeting and we will discuss this issue again at the end of the meeting.

Letter regarding reimbursement for genetic counselors

- Johnna Watson reports the letter regarding reimbursement for genetic counselors was finalized and mailed to Dennis Schrader, CFO for Medicaid, on August 9, 2018. There has been no response at this time.
- Discussion ensued regarding the change of the intent of the letter which initially was to address the lack of coverage for genetic testing through Maryland Medicaid (MA). Dr. Greene states microarrays are not being covered by Maryland Medicaid (MA). Sarah Viall states they were having this issue as well and they talked to MA. There appears to have been a mistake in Maryland MA and the wrong codes were being used. Some codes are being covered, and she will share with others.
- John McGing encouraged the genetic centers to share the codes with each other to determine if the problem has been resolved.

IV: Member Updates

- Laboratory Administration
  No additional updates provided

- MCHB
  Dr. Jed Miller reported that Dr. Howard Haft, former Deputy Secretary for Public Health Services, is now with a new program called Maryland Primary Care. Fran Phillips has been named as the Deputy Secretary for Public Health Services. Ms. Phillips was in this position from 2008-2013, and she has been re-appointed under Secretary Neall.

- Johnna Watson reports that the Federal Advisory Committee is meeting in 2 weeks and they are now looking at a new disorder, cerebrotendinous xanthomatosis (CTX) to see if this disorder should be moved forward into review process. Dr. Moser states this is a lipid disorder and babies with this disorder can have malabsorption and liver dysfunction. If left untreated, dementia can develop in the teens. Testing is through bile alcohol measurement through mass spectrometry. Dr. Moser is working on setting up a combined screen for X-ALD and CTX.

- Sarah Viall reports that Virginia is going live with screening for Pompe and MPS-I on January 1, 2019. They have voted to include SMA but it may be another year. They are still working on X-ALD.
- Dr. Miller reports that SMA was added to the RUSP in July, 2019.
- Michelle Smith states she has been on the newborn screening website, and some of the links are not working. Dr. Majid reports the lab website is currently being updated.

V. Future Topics for Discussion

- John McGing asked if members had any topics they would like addressed at future meetings. No response noted.
- Dr. Greene requested that the issue of reimbursement be discussed at the next meeting. She suggested inviting some of the genetic counselors who work in the clinics to come in to discuss the issue. Dr. Hilary Vernon states that she was told there were some codes that were not working but different codes were working. John McGing states if it is just an issue of codes needing to be changed, then it is not an issue for this Council. He reiterated the need for discussion among the genetic centers to share the appropriate codes and if the issue is not resolved, then the Council could hear a presentation on the issue to get a better idea of what the problem is and if there is anything that can be done.
- Sarah Viall asked if there could be a summary of the implementation of LSDs and SMA screening at the next meeting since it will be a couple months in process.

VI. Next Meeting Date:

- The members of the Council selected April 23rd as the next meeting date instead of April 9th as originally planned to avoid conflicts with the two national meetings. Meeting will be held April 23, 2019 at 5:00-7:00 pm at 201 W. Preston Street. Reminder and call-in information will be sent prior to this meeting.

VII. Adjournment

Meeting adjourned at 5:48 PM.