

State Advisory Council
Hereditary and Congenital Disorders
Minutes (Draft)
January 24, 2023
On-line meeting

Members Present

Jamie Fraser, Chair
John McGing
Sharon Dols
Robert Brosius
David Myles
Dominique Sessa
Michelle Smith

Members Absent

Shannon Dixon
Gerald Raymond
House of Delegates seat (vacant)
Senate seat (vacant)

Ex-Officio Present

Fizza Majid
Johnna Watson
Robert Myers

Ex-Officio Absent

Stacy Taylor

MDH Staff

Lauren Whiteman
LaPortia Barrows
Monique Veney
Jody Sheely
Wendeline Frederic

Guests

Alexandra Bowen, UMD Genetics
Sofia Saenz-Ayala, UMD Genetics
Carol Greene, UMD Genetics
Paul Vetter, PerkinElmer Genetics
Erin Strovel, UMD Genetics
Mimi Blitzer, UMD Genetics
Ada Hamosh, Johns Hopkins Genetics

Called to order

Meeting called to order at 17:36. Meeting ground rules (meeting etiquette) were shared with the Council by Chairperson Fraser.

I. WELCOME & ROLL CALL

Johnna Watson conducted meeting roll call.

II. APPROVAL OF MINUTES

Draft meeting minutes were sent out to committee prior to meeting for review. Chairperson Fraser asked if there any concerns or corrections to be made. Motion to approve minutes made by John McGing and seconded by Sharon Dols. Minutes approved.

III. MEMBERSHIP UPDATE

Johnna Watson reports that an email has been sent today by the membership coordinator requesting members for the vacant Delegate and State Senator seats.

IV. NEW BUSINESS

Senate Bill 188

- Chairperson Fraser advised that Senate Bill 188 – Development of a Rare Disease Council has been introduced. This bill was briefly discussed at the last meeting but there were other agenda items that needed to be discussed and now this bill needs to be addressed at tonight’s meeting.
- The bill has been advocated by the National Organization for Rare Disorders (NORD) and it is believed that they are advocating for a Rare Disease Advisory Council (RDAC) in every state.
- Chairperson Fraser asked for input from MDH staff present in the meeting.
- Jody Sheely states that MDH does not have any further background information on the proposed bill and is here strictly to offer any advice should the Council decide they want to take a position on the bill.
- Sharon Dols asked for an explanation of the difference between the RDAC and this Council. Chairperson Fraser states there are rare diseases that are genetic and rare diseases that are non-genetic in nature.
- The Council members and guests continued discussion of the bill and expressed concern regarding possible overlap and authority over newborn screening. Discussion regarding expansion of the current Council to include all rare diseases was also undertaken.
- John McGing feels the Council should advocate for asking the legislature to take out the newborn screening part. He states this Council can give them advice or suggestions regarding the proposed RDAC but not ask to expand this Council’s function because the expectations of the RDAC are massive.
- Dominique Sessa and Michelle Smith expressed concern regarding the composition of the RDAC membership because it does not include enough members with rare diseases or families of people with rare diseases.
- Michelle Smith made a motion to write a letter expressing the Council’s concerns. John McGing seconded and volunteered to draft a letter for further Council review.
- Roll call vote taken and there were 7 ayes and 0 dissents.

CCHD Sub-Committee

- Johnna Watson reports that MDH needs to re-convene CCHD sub-committee to review new CCHD guidelines that have been introduced through AAP. Some members of the previous sub-committee are no longer at the institutions. Johnna will research old membership and reach out directly to the genetic centers for assistance in developing a new CCHD sub-committee.

V. OLD BUSINESS

- Chairperson Fraser made a motion to table MPS-II discussion until the next meeting. Robert Brosius seconded the motion. He also stated that in order to determine an appropriate score using the new criteria is to take a condition that is already on the panel and re-evaluate it using the new criteria to see where the “passing” score would lie.

VI. UPDATES

- **MCHB** (Lauren Whiteman)
 - The OGPSHCN is actively recruiting for a medical director. Link placed in the chat.

- **Laboratory (Dr. Fizza Majid)**
 - Dr. Majid reports there are still staffing issues. One staff member is starting in February.

- **Newborn Screening Follow-up (Johnna Watson)**
 - Johnna Watson reports the follow-up team is still trying to transition babies into Starlims. The NBS follow-up team is looking to hire an administrative aide to assist in this process.
 - Chairperson Fraser asked if there is a COOP plan in place should another disruption occur. Dr. Myers reports the lab is in the procurement process which will have PerkinElmer Genetics run the testing if there is another issue with the MD State Laboratory.

VII. NEXT MEETING DATE

- February 28, 2023 was proposed as the next meeting date.

VIII. ADJOURNMENT

- Michelle Smith moved to adjourn meeting, motion seconded by John McGing. Meeting adjourned at 18:57.

State Advisory Council
Hereditary and Congenital Disorders
Minutes (Draft)
February 9, 2023
On-line meeting

Members Present

Jamie Fraser, Chair
John McGing
Sharon Dols
Robert Brosius
Michelle Smith
Shannon Dixon
Gerald Raymond

Ex-Officio Absent

Fizza Majid
Stacy Taylor
Robert Myers

Ex-Officio Present

Johnna Watson

Members Absent

Dominique Sessa
David Myles
House of Delegates seat (vacant)
Senate seat (vacant)

MDH Staff

Lauren Whiteman
Monique Veney

Guests

Carol Greene, UMD Genetics
Mimi Blitzer, UMD Genetics
Ada Hamosh, Johns Hopkins Genetics
Erin Strovel, UMD Genetics

Called to order

Meeting called to order at 17:35. Meeting ground rules (meeting etiquette) were shared with the Council by Chairperson Fraser.

I. WELCOME & ROLL CALL

Johnna Watson conducted meeting roll call.

II. DISCUSSION REGARDING HB 302 & SB 188

- At the request of Chairperson Fraser, Lauren Whiteman gave a summary of the discussion of the Senate committee hearing on the bill.
- Discussion of the notes, including the proposed amendments which would remove newborn screening from the bill, add the Council chair from this Council to the RDAC and adding an additional care provider as well, ensued.
- After discussion, edits were made to the letter in real time.
- Motion was made to accept the letter/testimony as written by Gerald Raymond. Shannon Dixon seconded the motion. Vote was taken by roll call vote. 6 ayes and no dissents.
- Written testimony will be submitted by Sharon Dols.

III. ADJOURNMENT

- Gerald Raymond moved to adjourn meeting, motion seconded by John McGing.
Meeting adjourned at 19:00.

**State Advisory Council
Hereditary and Congenital Disorders
Minutes**

February 28, 2023

On-line meeting

Members Present

Jamie Fraser, Chair
Robert Brosius, Vice Chair
John McGing
Delegate Terri Hill
Shannon Dixon
Sharon Dols
David Myles
Gerald Raymond
Michelle Smith

Members Absent

Dominique Sessa
Senate seat (vacant)

Ex-Officio Present

Fizza Majid
Johnna Watson

Ex-Officio Absent

Stacy Taylor
Robert Myers

MDH Staff

Lauren Whiteman
Monique Veney
LaPortia Barrows

Guests

Carol Greene, UMMS Genetics
Erin Strovel, UMMS Genetics
Sofia Saenz-Ayala, UMMS Genetics
Mary Hackbarth, UMD 2nd yr med student
Christina Grant, CNMC Genetics

CALLED TO ORDER

Meeting called to order at 17:35. Meeting ground rules (meeting etiquette) were shared with the Council by Chairperson Fraser.

I. WELCOME & ROLL CALL

Johnna Watson conducted meeting roll call.

II. INTRODUCTIONS

The delegate seat has been filled by Delegate Terri Hill who was welcomed and asked to introduce herself to the Council.

All members of the Council introduced themselves in honor of Rare Disease Day.

III. APPROVAL OF MINUTES

January 24, 2023 minutes – John McGing motioned to approve and seconded by Shannan Dixon

February 9, 2023 minutes – Gerald Raymond motioned to approve and seconded by Sharon Dols

IV NEW BUSINESS

Johnna Watson reviewed new Council meeting procedures which are more in line with the other Councils associated with MDH.

- Members will discuss topics 1st, followed by a public comment period
 - Public comment period will be limited to 8 minutes total
 - Each guest will have a 2 minute limit, on screen timer to be used
 - Guest comment period can be lengthened at the Chair's discretion
 - Public comment can be placed in the chat if time runs out
 - Public comment can also be submitted in advance

V. OLD BUSINESS

Johnna Watson asked if there should be a recap of MPS-II for the benefit of Delegate Hill since this is her first meeting. Discussion took place and it was determined that Gerald Raymond would give a brief presentation on MPS-II. Previous presentation reviewed with highlights given.

Following the presentation:

Member discussion:

Discussion ensued regarding false positives and lack of second tier testing to reduce false positives from the Maryland State NBS laboratory. This is the same concern for other conditions on the Maryland panel as well.

Discussion also centered around the differences in outcomes between children treated early in life vs those who were treated after being diagnosed later in life, as well as the inaccessibility of enzyme replacement therapy in Maryland that crossed the blood brain barrier.

Discussion also ensued about the value of information for families to help them make decisions concerning care of their children.

Agreement amongst the members regarding each of these issues was not obtained.

Public Comment period:

Christina Grant expressed concern over the harm of false positives.

Carol Greene shared that University of MD has outreach clinics, so patients do not have to travel from the Eastern Shore for diagnostic testing.

Secondary to time constraint, review of the scoring criteria and potential vote was tabled until next meeting.

VI. NEXT MEETING DATE

April 4th was proposed, but this is the last week of legislative session so April 11, 2023 was selected.

VII. ADJOURNMENT

Michelle Smith moved to adjourn the meeting and seconded by Delegate Hill.

Meeting adjourned at 6:56 PM.

**State Advisory Council
Hereditary and Congenital Disorders
Minutes (Draft)**

May 9, 2023
On-line meeting

Members Present

Jamie Fraser, Chair
Delegate Terri Hill
John McGing
Michelle Smith
Shannon Dixon
Sharon Dols
David Myles
Gerald Raymond

Members Absent

Robert Brosius, Vice Chair
Dominique Sessa

Ex-Officio Present

Fizza Majid
Johnna Watson
Stacy Taylor
Robert Myers

MDH Staff

Laportia Barrows
Monique Veney

Guests

Allison Faila, CNMC Genetics
Paul Vetter, PerkinElmer Genetics

CALLED TO ORDER

Meeting called to order at 5:31 pm. Meeting ground rules (meeting etiquette) were shared with the Council by Chairperson Fraser.

I. WELCOME & ROLL CALL

Johnna Watson conducted meeting roll call.
(Fizza Majid joined the meeting after roll call)

II. APPROVAL OF MINUTES

Delegate Hill motioned to approve minutes from 4/11/2023; seconded by Michelle Smith. No objections. Minutes approved and will be posted on the website.
(Chairperson Fraser's name was corrected)

III. MEMBERSHIP UPDATE

Johnna Watson reported on the following:

- Michele Smith's renewal forms have been received.
- Dominique Sessa's term will still expire 6/2023 and no updates from her at this time.
- Senate seat still no update at this time.

IV. OLD BUSINESS

Review of MPS-II letters to the Secretary

- Chairperson Fraser thanked families that came out to the meeting. She said their presence was essential and helpful.

- The Committee reviewed the MPS-II letter to the Secretary
 - Chairperson Fraser had an addition to her name and otherwise agreed with everything else as it was written. She thanked Gerald Raymond for writing the letter and his initiative.
 - Michelle Smith added the letter was well written.
 - Chairperson Fraser made changes/edits to the letter as members mentioned during discussion. Letterhead updated.
 - Motion to approve letter Delegate Hill; seconded by Shannon Dixon. No objections. Motion passed.
- The Committee discussed the tiered testing implementation letter.
 - Gerald Raymond shared he changed the theme a little because tiered testing in general needs to be addressed and to not just focus on GAG's. Chairperson Fraser agreed.
 - Feedback requested from Fizza Majid on X-ALD and the tier testing. She indicates that the plan for X-ALD screening includes using a tower as the primary screening method so false positives should be minimized.
 - Council discussed other edits and suggestions to the body of the letter.
 - Motion to approve letter Michele Smith; seconded by Delegate Hill. No objections. Motion passed.

V. TOPICS FOR FUTURE MEETINGS

Presentation on Guanidinoacetate methyltransferase deficiency (GAMT)

- Chairperson Fraser gave a brief definition of the Guanidinoacetate methyltransferase deficiency (GAMT) that was added to the RUSP January, 2023.
 - Per new MD State law, The Council must consider a new condition for the MD screening panel within a year of addition to the RUSP.
 - For the next meeting, Chair Fraser proposed a basic presentation to talk about GAMT and to also hear from the lab about how they would implement GAMT screening and the expected timeline and additional costs.
 - Johnna Watson mentioned there was a presentation a few years ago given by Dr. Lisa Kratz.
 - Chair Fraser also says there is a GAMT expert who did a talk at ISMD for presentation if he is willing to come or share his slides, if Dr. Kratz is not available.
- Delegate Hill mentioned legislation just passed creating the Rare Disease Advisory.

VI. UPDATES

MCHB

- Stacy Taylor shared with the Committee there have been a lot of leadership changes within the Bureau due to the new Administration. She let the Committee know they can reach out to her if they need to know if a specific person is still in position. Stacy mentioned the Rare Disease Advisory just discussed will likely be assigned to OGPSHCN (Office for Genetics and People with Special Health Care Needs).

Laboratory

- Fizza Majid reported there is no X-ALD screening timeline at this moment but they are moving forward trying to identify the method. Part of the training has been done and they have been able to recruit and fill some of their positions. Fizza will have a GAMT update next meeting.

NBS Follow-up

- Johnna Watson shared they have been meeting with the lab and genetics centers. She thanked all for their input. NBS has been able to reduce the number of false positives. Impacting families have gotten much better and they are continuing to look at the cut-offs to improve that process as they go along. Johnna asked Dr. Raymond and Chair Fraser to meet with herself and NBS nurses to discuss protocols for X-ALD prior to screening implementation. Chair Fraser suggested adding Alli to that meeting as well.

VII. NEXT MEETING DATE

July 18, 2023 was selected as the next meeting date.

VIII. ADJOURNMENT

John McGing moved to adjourn the meeting and seconded by Gerald Raymond.
Meeting adjourned at 6:15 pm.

State Advisory Council
Hereditary and Congenital Disorders
Minutes

April 11, 2023

On-line meeting

Members Present

Jamie Fraser, Chair
Robert Brosius, Vice Chair
John McGing
Delegate Terri Hill
Shannon Dixon
Sharon Dols
David Myles
Gerald Raymond
Michelle Smith

Members Absent

Dominique Sessa
Senate seat (vacant)

Ex-Officio Present

Fizza Majid
Johnna Watson
Stacy Taylor
Robert Myers

MDH Staff

Lauren Whiteman
Monique Veney

Guests

Carol Greene, UMMS Genetics
Erin Strovel, UMMS Genetics
Christina Grant, CNMC Genetics
Allison Faila, CNMC Genetics
Kristin McKay, Project Alive
Sharon Kauders, MPS-II parent
Mike Hu, Columbia University
Ivana Thomas, GC student
Paul Vetter, PerkinElmer Genetics

CALLED TO ORDER

Meeting called to order at 17:35. Meeting ground rules (meeting etiquette) were shared with the Council by Chairperson Fraser.

I. WELCOME & ROLL CALL

Johnna Watson conducted meeting roll call.

(Robert Brosius, Michele Smith and David Myles joined after completion of roll call.)

II. APPROVAL OF MINUTES

John McGing motioned to approve minutes from 2/28/2023; seconded by Sharon Dols. No objections. Minutes approved and will be posted on website.

III. MEMBERSHIP UPDATE

Prior to membership update, Johnna Watson reported that SB188 did pass with approved amendment as discussed at previous meetings. It is not known at this time which MDH office will be responsible for staffing the newly created Rare Disease Advisory Council. The Office of Budget and Management will be making that determination in the next 3-6 months.

Johnna Watson reported on the following:

- Michele Smith needs to submit application to renew term which expired in 6/22

- Dominique Sessa's term will expire 6/2023 and she is eligible for renewal.
 - application to be sent to both of these members.
- Senate seat – no update at this time. Chairperson Fraser asked Delegate Hill to consider approaching Senators she may know or worked with in the past regarding joining the Council

IV. OLD BUSINESS

Continued discussion of MPS-II using criteria for inclusion on Maryland NBS Panel

- Chairperson Fraser asked for public comments first and reviewed time limits
 - Sharon Kauders – son with Hunter's testified regarding son's diagnosis and admission into a trial in North Carolina.
 - Mike Hu – two sons with Hunter's. Older child diagnosed at 3 years of age and the younger at 1 year of age. The younger child is benefiting more from treatment because of age treatment started
 - Kristin McKay – Director of Project Alive, son was diagnosed prenatally and started treatment at 1 month. No symptoms in her child at 4 years of age.
 - Christina Grant, CNMC Genetics – presented information regarding lack of FDA approved treatments and the number of false positives for LSD in MD
- Gerald Raymond acknowledged the family members who publicly testified. Chairperson Fraser also thanked the family members.
- Delegate Hill asked Dr. Grant to provide a little more detail regarding her concerns about the false positive rates in MD for LSD.
 - Follow-up information provided by Dr. Grant indicated the referral rate for MPS-II is predicted to be twice as much as the national referral rate predicted by the RUSP. Discussion followed regarding how referrals are addressed by each of the genetic centers.
- Gerald Raymond advised that the consideration is whether the Council feels MPS-II is a good condition to be on the panel as a public health measure.
- John McGing concurred that the ability for the lab to reduce false positives should not be a consideration.
- Voting members then discussed the NBS Criteria as it relates to MPS-II and scored each criteria as a group.
- Discussion also centered around whether or not the blood spots can be sent from the lab to an outside lab for 2nd tier test as a part of screening.
- Chairperson Fraser proposed adding MPS-II to the Maryland screening panel only if there is a 2nd tier test, such as GAGs, as part of the screen, and X-ALD should be implemented before screening starts for MPS-II.
 - Discussion centered around this topic, and it was determined that the second tier testing and implementation should be separate from the decision to screen for MPS-II. A separate recommendation to the Secretary for inclusion of GAGs in the screening was discussed as well.
- John McGing proposed a vote on just adding MPS-II to the Maryland NBS panel, without conditions. Gerald Raymond seconded the motion.
- Roll call vote taken with 8 yes votes and 1 no vote.
- Delegate Hill moved to send a letter to the Secretary explaining the need to include GAGs in the screening process. Shannan Dixon seconded the motion.
- Roll call vote taken and there were 9 yes votes and 0 no votes.
- Gerald Raymond will draft a letter to the Secretary recommending MPS-II should be added to the Maryland newborn screening panel, as well as a second letter recommending GAGs be added to the screening for MPS-II.

V. UPDATES

- MCHB – Lauren Whiteman: no updates
- Laboratory – Fizza Majid reports that training for X-ALD will be conducted with available staff over the next two weeks. Chairperson Fraser asked if there is a disaster plan in place to handle future disasters.
- NBS Follow-up – Johnna Watson: inquired about length of time of training for X-ALD and was advised training would be 2 weeks but with the staff shortages implementation will not be at the end of training.

VI. NEXT MEETING DATE

May 9, 2023 was selected as the next meeting date.

VII. ADJOURNMENT

Gerald Raymond moved to adjourn the meeting and seconded by Sharon Dols.
Meeting adjourned at 7:15 PM.

State Advisory Council
Hereditary and Congenital Disorders
Minutes (Final)
July 18, 2023
On-line meeting

Members Present

Jamie Fraser, Chair
Robert Brosius, Vice Chair
John McGing
Gerald Raymond
Michelle Smith
Sharon Dols
Shannon Dixon
Delegate Terri Hill

Members Absent

David Myles
Dominique Sessa
Senator- Vacant

Ex-Officio Present

Fizza Majid
Robert Myers
Johnna Watson
Stacy Taylor

MDH Staff

Lauren Whiteman
Laportia Barrows
Tatum Cox
Monique Veney

Guests

Paul Vetter, PerkinElmer Genetics
Allison Faila, CNMC Genetics
Carol Greene, UMD Genetics
Lisa Kratz, KKI

CALLED TO ORDER

Meeting called to order at 5:36 pm. Meeting ground rules (meeting etiquette) were shared with the committee by Chairperson Jamie Fraser.

I. WELCOME & ROLL CALL

Laportia Barrows conducted meeting roll call.

II. APPROVAL OF MINUTES

Draft meeting minutes were sent out to the Committee prior to meeting for review. Chairperson Fraser asked were there any corrections to be made. Two corrections were noted and corrected. Motion to approve amended minutes made by Dr. Raymond and seconded by Sharon Dols. No opposing votes. Minutes approved.

III. MEMBERSHIP UPDATE

- Laportia Barrows reported to the Committee:
- Michele Smiths term has been renewed.
 - Dominique Sessa's membership forms are pending.
 - No update on the vacant Senate seat.

IV. OLD BUSINESS

- Status of MPS-II letters to the Secretary
- The letters have been completed and submitted to the Secretary.

V. NEW BUSINESS

Chairperson Fraser inquired with the Council if anyone was seeing heat impacts on false positives with the recent mass heat waves. She was wondering if MD needed to be updated on anything being as though DC and VA have not been having the same outcomes. Johnna added that the state lab has been receiving an increase of specimen's coming in that were unsatisfactory heat related and wanted to discuss that at the next meeting with the genetic centers.

Presentation on Guanidinoacetate methyltransferase deficiency (GAMT)

- Chairperson Fraser gave a brief definition of the Guanidinoacetate methyltransferase deficiency (GAMT) that was added to the RUSP, January 2023 and introduced Dr. Lisa Kratz for her GAMT presentation.
 - Dr. Kratz began her presentation sharing an outline of GAMT.
 - GAMT deficiencies, clinical features and diagnosis were shared with the committee.
 - Dr. Kratz shared and explained different cases and treatments.
 - Different GAMT studies were shared with the Council.
 - GAMT questions to Dr. Kratz were asked and discussed.
 - The Council discussed with the MD Lab a timeline to add GAMT to existing platforms.
 - Dr. Katz was asked to forward her presentation so that the Council can review for future meeting.

VI. UPDATES

- MCHB – Lauren Whiteman officially announced Johnna Watson's retirement and Laportia's Barrows new position as the NBS program manager. Her NBS follow up nurse position will be posted soon if anyone is interested.
- Laboratory – Dr. Majid reported to the Committee that the goal is to have X-ALD screening by August.
- NBS Follow-up – Laportia Barrows shared with the Council they are seeing an uptick with the GALT and Biotinidase abnormalities as well as unsatisfactory specimens as mentioned earlier. They do meet monthly with the Genetic centers and the lab to work on protocols and cut offs to help the false positive rates. She also mentioned NBS is recruiting now for a NBS follow up nurse and an administrative aid to assist the follow-up team may be starting soon.

VII. FUTURE TOPICS

Johnna Watson mentioned she was approached by a group of Audiologist to discuss blood spot CMV's. The group would like to present to the Council. It was agreed to have them present in September. The Committee discussed CMV.

VIII. NEXT MEETING DATE

August 22, 2023, was selected as the next meeting date.

IX. ADJOURNMENT

Dr. Raymond moved to adjourn the meeting and seconded by Michelle Smith. Meeting adjourned at 6:15 pm.

State Advisory Council
Hereditary and Congenital Disorders
Minutes (Final)
August 22, 2023
On-line meeting

Members Present

Jamie Fraser, Chair
Robert Brosius, Vice Chair
John McGing
Gerald Raymond
Delegate Terri Hill
David Myles
Shannon Dixon

Members Absent

Michelle Smith
Sharon Dols
Dominique Sessa
Senator- Vacant

Ex-Officio Present

Fizza Majid
Robert Myers
Stacy Taylor
Johnna Watson

MDH Staff

Lauren Whiteman
Laportia Barrows
Monique Veney

Guests

Dr. Carolyn Jenks, Consortium
Carolyn Horton, Aud., CCC-A, Consortium
Ruth S. Martin, PhD, CCC-A, Consortium
James D. Campbell, MD, MS, UMMS
Dr. Roberta DeBiasi, CNMC
Matthew Miyamoto, UMD
Mickey Kuo- NIH Fellow
Paul Vetter, PerkinElmer Genetics
Allison Faila, CNMC Genetics
Carol Greene, UMD Genetics
Amanda Devereaux
Mark Schlesis, Univ. Minn.
Erin Strovel, UMD

CALL TO ORDER

Meeting called to order at 5:40 pm. Meeting ground rules (meeting etiquette) were shared with the committee by Chairperson Jamie Fraser.

I. WELCOME & ROLL CALL

Laportia Barrows conducted meeting roll call.

II. APPROVAL OF MINUTES

Draft meeting minutes were sent out to the Committee prior to meeting for review. Chairperson Fraser asked were there any edits to be made. Motion to approve amended minutes made by Dr. Raymond and seconded by John McGing. No opposing votes. Minutes approved.

III. MEMBERSHIP UPDATE

Laportia Barrows reported to the Committee:

- Dominique Sessa's membership forms submission is pending.
- The Senate seat has been filled and his name is Senator Johnny Ray Sailing of Baltimore County.

IV. NEW BUSINESS

Presentation on Screening for Congenital Cytomegalovirus (cCMV)

- Ruth Marin, PhD, Carolyn Horton and Dr. Carolyn Jenks of the Maryland chapter of the Mid-Atlantic CMV Consortium presented:
 - Carolyn Horton began the presentation sharing participants and what cCMV is and who is affected by this common virus.
 - The transmittal through bodily fluids is how cCMV is contracted.
 - Statistics and a comparison with other congenital disorders were shared with the Council.
 - Hearing loss is the most common complication of cCMV. It occurs often.
 - There are no cures for cCMV but treatment is on the horizon.
 - Targeted vs Universal cCMV screening was shared. Targeted screening being inadequate was explained.
 - A map was shared with the Council displaying cCMV screenings throughout the U.S.
 - Universal screening is important and the Consortium shared the clinical trials are showing promise.
 - The Consortium shared the cCMV screening methods and financial benefits.
 - In conclusion, a list of cCMV educators and references were shared with the Council.
- The floor was opened to the Council for questions.

Presentation on Congenital Cytomegalovirus (cCMV) Overview with Emphasis on Considerations for Newborn Screening.

- Dr. James Campbell presented on additional cCMV data.
- After Dr. Campbells presentation, the floor was opened for questions. The Council began a discussion.

V. UPDATES

- MCHB – Lauren Whiteman announced Johnna Watson’s last day officially will be Friday. Laportia Barrows will be taking over the position. The NBS f/u nurse position has closed and interviews are being scheduled for that position.
- Laboratory – Dr. Majid reported no updates at this time. She shared there are five positions still to be filled and 3 to 4 starting within the month. X- ALD screening still on hold.
- NBS Follow-up- Laportia Barrows thanked Johnna Watson for her years of service and assured the Council the MD babies will continue to be taken care of.

VI. FUTURE TOPICS

GAMT and cCMV discussions.

VII. NEXT MEETING DATE

September 12, 2023

VIII. ADJOURNMENT

John McGing moved to adjourn the meeting and seconded by Delegate Hill.
Meeting adjourned at 7:02 pm.

State Advisory Council
Hereditary and Congenital Disorders
Minutes (Final)
October 24, 2023
On-line meeting

Members Present

Jamie Fraser, Chair
Robert Brosius, Vice Chair
John McGing
Sharon Dols
Delegate Terri Hill
David Myles
Shannon Dixon
Senator Johnny Ray Salling

Members Absent

Michelle Smith
Gerald Raymond
Dominique Sessa

Ex-Officio Present

Fizza Majid
Robert Myers
Stacy Taylor

MDH Staff

Lauren Whiteman
Laportia Barrows
Monique Veney
Theresa Wavra

Guests

Carol Greene, UMD
Jelili Ojodu, NBS
Paul Vetter, Revvity (PerkinElmer)
Erin Strovel, UMD
Kevin Brown, NBS
Prakash Paudyal, NBS
Rodney Hargraves, NBS
Mimi Blitzer, UMD

CALL TO ORDER

Meeting called to order at 5:33 pm. Meeting ground rules were shared with the committee by Chairperson Jamie Fraser.

I. ROLL CALL & INTRODUCTION

Laportia Barrows conducted the meeting roll call and attendees introduced themselves.

II. APPROVAL OF MINUTES

Draft meeting minutes were sent out to the Committee prior to the meeting for review. Chairperson Fraser asked were there any edits to be made. Motion to approve minutes made by John McGing and seconded by Delegate Hill. No opposing votes. Minutes approved.

III. MEMBERSHIP UPDATE

Laportia Barrows reported to the Committee:

- Dominique Sessa's reappointment has been approved.
- All other seats filled.

IV. OLD BUSINESS

The Committee agreed to table the GAMT and cCMV letters at this time.

V. NEW BUSINESS

Newborn Screening Program Presentation

- Laportia Barrows introduced her NBS team along with new staff starting soon and gave an overview of NBS bloodspots:
 - What newborn bloodspot screening was explained.
 - Over 50 disorders are screened for in MD.
 - PKU is just one disorder although it is referenced for all the disorders. NBS is in the process of transferring that language to NBS or NB Metabolic screen so that people are not confused.
 - NBS is offered to all babies born in Maryland and in 2008 consent was changed.
 - Blood spot screening was detailed along with their schedules.
 - The NBS follow up teams role was explained in the screening process.
 - Protocol for NBS abnormal and unsatisfactory results was shared.
- The floor was open for questions. The Council discussed Newborn Screening.

VI. UPDATES

- MCHB – Lauren Whiteman mentioned no updates at the moment.
- Laboratory – Dr. Majid reported XLD should be going live by the end of the year, GAMT and MPS II to follow. It was also reported that ALD may be on the Newborn Screen by 2024.

Dr. Meyers shared with the Council that Dr. Majid will be retiring before the end of this year. Dr. Paudyal was introduced as the acting Division Chief during this transition. Kevin Brown was also introduced and will be responsible for the operations of the NBS lab. Dr. Meyers assured the Council that they will be fully supported during this transition and a national search will be held to find a new Division Chief.

- NBS Follow-up- Laportia Barrows had no additional NBS updates at this time.

VII. FUTURE TOPICS

GAMT and cCMV letters.

VIII. NEXT MEETING DATE

November 14, 2023

IX. ADJOURNMENT

John McGing moved to adjourn the meeting and was seconded by Delegate Hill.
Meeting adjourned at 6:32 pm.

State Advisory Council
Hereditary and Congenital Disorders
Minutes (Draft)
December 5, 2023
On-line meeting

Members Present

Jamie Fraser, Chair
Robert Brosius, Vice Chair
John McGing
Sharon Dols
Delegate Terri Hill
David Myles
Gerald Raymond

Members Absent

Senator Johnny Ray Salling
Michelle Smith
Shannon Dixon
Dominique Sessa

Ex-Officio Present

Roberts Myers

MDH Staff

Lauren Whiteman
Laportia Barrows
Monique Veney
Theresa Wavra
Luz Del Valle Sanchez
Kevin Brown
Prakash Paudyal
Rodney Hargraves

Guest

Carol Greene, UMD
Erin Strovel, UMD
Paul Vetter
Allison Faila, CNMC Genetics
Mickey Kuo
Sofia Saenz-Ayala

CALL TO ORDER

Meeting called to order at 5:58 pm. Meeting ground rules were shared with the committee by Chairperson Jamie Fraser.

I. ROLL CALL & INTRODUCTION

Laportia Barrows conducted the meeting roll call and attendees introduced themselves.

II. APPROVAL OF MINUTES

Draft meeting minutes from the October 24th meeting were sent out to the Committee prior to the meeting for review. Chairperson Fraser asked were there any edits to be made. Motion to approve minutes made by Gerald Raymond and seconded by David Myles. No opposing votes. Minutes approved.

III. MEMBERSHIP UPDATE

Laportia Barrows reported to the Committee that all positions are filled.

IV. OLD BUSINESS

Status of GAMT letter

- The GAMT letter was sent out with revisions to the Council for review.
- Chairperson Fraser asked if there were any additional edits.
- A motion to approve the GAMT letter as drafted with the edits to send to the Secretary was made by Delegate Hill and seconded by Gerald Raymond.
- The Committee voted to approve the letter.

Status of cCMV letter

- The cCMV letter was distributed to Council members to review.
- Chairperson Fraser asked if there were any additional comments to add to the text of the letter as it currently stands.
- Motion to approve the cCMV letter was made by Deleat Hill and seconded by John McGing
- The Committee voted to approve the letter.

V. NEW BUSINESS

Process for a disorder to be added to the RUSP.

- Allison Failla- Genetic Counselor, Children’s National Medical Center presented.
 - Alison gave an overview of the processes of adding disorders to the RUSP.
 - A diagram of steps was shared with and explained to the Committee.
 - The six page nomination of a condition form was displayed for the Council to review.
- The Committee discussed some presentation items, the nominations form and some legislature.

VI. UPDATES

- MCHB – Lauren Whiteman referenced no other updates aside from introducing new NBS staff and legislative questions to add to the NBS mentioned earlier.
- Laboratory – Dr. Meyers mentioned that Dr. Prakash is the acting Division Chief at this time until Dr. Majids replacement can be found and Kevin is assisting him with managing operations. Posting for the position went up last week. Dr. Myers also said that the ALD screening should be up by the end of the year which is their goal and he will also have an update on some other test they are working on the first part of the year.
- NBS Follow-up- Laportia Barrows had no NBS updates.

VII. FUTURE TOPICS

Update on NBS II.

VIII. NEXT MEETING DATE

January 9, 2024

IX. ADJOURNMENT

Gerald Raymond moved to adjourn the meeting and was seconded by John McGing .
Meeting adjourned at 6:28 pm.