

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

**February 15, 2022**

(Virtual meeting secondary to Covid-19)

**Members Present**

Erin Strovel, Chair  
Jamie Fraser, Vice Chair  
Michelle Smith  
Jamie Fraser  
Robert Brosius  
Meral Gunay  
Delegate Karen Lewis-Young  
David Myles

**Members Absent**

John McGing  
Senator Edward Reilly  
Sharon Dols  
1 vacant health unrelated member

**Ex-Officio Present**

Johnna Watson  
Robert Myers  
Stacy Taylor

**Ex-Officio Absent**

Fizza Majid

**Guests**

Ada Hamosh  
Allison Shaw, CNMC Genetics  
Chelsea Kadish, Delegate Lewis-Young's  
office

**MDH Staff**

Monique Veney  
Laportia Barrows

**I. WELCOME & ROLL CALL**

Roll call completed and quorum present.  
Dr. Strovel, Council Chair, welcomed everyone to the meeting.

**II. DISCUSSION OF HB 109**

Dr. Strovel gave an introduction of HB 109.

Delegate Lewis-Young states she is on the committee that is hearing this bill and reports that Delegate Shetty was not aware of the Advisory Council on Hereditary and Congenital Disorders.

HB 109 is requiring Maryland to screen for all core conditions on the Federal Recommended Universal Screening Panel (RUSP) within a 2-year timeframe.

Discussion centered around the requirement of screening for conditions on the RUSP without input from the Advisory Council on Hereditary and Congenital Disorders. Delegate Lewis-Young states there is still some time to prepare letters to the Chairs of the committees in both houses.

Motion made to submit a position statement on behalf of the Advisory Council by Jamie Fraser, seconded by Michelle Smith. Vote showed no opposition. Position statement will be drafted and shared in an open meeting.

Plan is to post a meeting for Thursday, February 17, 2022, to review the draft position statement.

Motion to adjourn made by Michelle Smith, seconded by Dr. Myles. Meeting adjourned at 6:30 PM

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

**February 17, 2022 5:30 PM**

(Virtual meeting secondary to Covid-19)

**Members Present**

Erin Strovel, Chair  
Jamie Fraser, Vice Chair  
Michelle Smith  
Meral Gunay  
David Myles  
Delegate Karen Lewis-Young  
John McGing (late)

**Members Absent**

Robert Brosius  
Senator Edward Reilly  
Sharon Dols  
1 vacant Health Unrelated

**Ex-Officio Present**

Johnna Watson  
Stacy Taylor

**Ex-Officio Absent**

Fizza Majid  
Robert Myers

**Guests**

Carol Greene, UMMS Genetics  
Ada Hamosh, Hopkins Genetics  
Paul Vetter, PerkinElmer Genetics

**MDH Staff**

Monique Veney

**I. WELCOME & ROLL CALL**

Roll call completed and quorum present.

Dr. Strovel, Council Chair, welcomed everyone to the meeting.

**II. DISCUSSION OF HB 109**

Dr. Strovel shared a copy of the draft position statement. The position statement recommends amending the bill to include screening for all conditions on the RUSP, under the advisement of the Council, and to include sufficient funding and an expedited process for the procurement of necessary equipment and staffing to enable the proper and timely implementation of new conditions added to the newborn screen.

Motion made to submit the position statement on behalf of the Advisory Council by Jamie Fraser, seconded by John McGing. Vote showed no opposition. Position statement to be forwarded to legislature via Office of Support Services at MDH.

Plan to meet again in 2-3 weeks, March 1st or 3rd, prior to the end of the session.

Motion to adjourn made by Dr. Fraser, seconded by Dr. Strovel. Meeting adjourned at 6:30 PM

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

**February 28, 2022 5:30 PM**

(Virtual meeting secondary to Covid-19)

**Members Present**

Erin Strovel, Chair  
Jamie Fraser, Vice Chair  
Michelle Smith  
Jamie Fraser  
Meral Gunay  
John McGing  
Senator Edward Reilly  
Delegate Karen Lewis-Young  
Sharon Dols  
Robert Brosius

**Members Absent**

David Myles  
1 vacant health unrelated member

**MDH Staff**

Monique Veney  
Laportia Barrows

**Ex-Officio Present**

Johnna Watson  
Stacy Taylor

**Ex-Officio Absent**

Fizza Majid  
Robert Myers

**Guests**

Ada Hamosh, Hopkins Genetics  
Carol Greene, UMMS Genetics  
Jody Sheely, Office of Support Services, MDH  
Maggie McGinnis, Office of Support Services  
Shelly Choo, MCHB, MDH

**I. WELCOME & ROLL CALL**

Roll call completed and quorum present.

Dr. Strovel, Council Chair, welcomed everyone to the meeting.

**II. LEGISLATIVE UPDATE/TRAINING**

Presentation given by Jody Sheely and Maggie McGinnis describing the legislative process in Maryland.

Discussion ensued regarding how the Council will keep up with proposed bills for future legislative sessions. However, it was recommended that, based on time, review of the amendments as discussed between Dr. Strovel and Delegate Shetty should be completed at this time and the previous discussion was tabled for a later meeting.

Dr. Strovel reviewed the most recent amendments received after speaking with Delegate Shetty over the phone. She also reports that Delegate Shetty appears very willing to work with the Council on appropriate amendments.

Discussion regarding the newly proposed amendments ensued. Language change discussed to clarify what the annual review process would entail. Motion was made by Dr. Fraser to approve the language changes and to approve Dr. Strovel to continue discussions regarding amendments with Delegate Shetty and bring back results of the discussion to the Council for a formal position vote. Seconded by John McGing. Vote was 6-0 in support.

Tentative meetings scheduled for 3/2/2022 and 3/3/2022 to allow time for discussion with Delegate Shetty.

Meeting adjourned at 6:30 PM

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

**March 2, 2022 5:30 PM**  
(Virtual meeting secondary to Covid-19)

**Members Present**

Erin Strovel, Chair  
Jamie Fraser, Vice Chair  
Robert Brosius  
Jamie Fraser  
Sharon Dols  
Meral Gunay  
John McGing

**Members Absent**

David Myles  
Senator Edward Reilly  
Delegate Karen Lewis-Young  
Michelle Smith  
1 vacant health unrelated position

**Ex-Officio Present**

Johnna Watson  
Stacy Taylor

**Ex-Officio Absent**

Fizza Majid  
Robert Myers

**Guests**

Ada Hamosh, Hopkins Genetics  
Carol Greene, UMMS Genetics  
Shelly Choo, MCHB, MD  
Paul Vetter, PerkinElmer Genetics

**MDH Staff**

Monique Veney  
Laportia Barrows

**I. WELCOME & ROLL CALL**

Roll call completed and quorum present.

Dr. Strovel, Council Chair, welcomed everyone to the meeting.

**II. LEGISLATIVE UPDATE**

Dr. Strovel shared current proposed amendments to HB 109 that were recently discussed between her and Delegate Shetty. Changes in current proposed amendments were reviewed.

Clarification is needed on when the two-year time period begins - either after the condition is on the RUSP or the Maryland newborn screening panel -was discussed. The name of the Council in incorrect in the Bill. Suggested changes to the name of the Council and clarification regarding the 2-year time period were entered into the document with track changes to share back with Delegate Shetty.

Dr. Strovel received an email during this meeting from Delegate Shetty indicating she is meeting with the advocates tomorrow. The Council does not feel a letter of support should be given until proposed amendments are finalized since there are still discussions with the advocates.

Letter of support was edited to reflect support of the current proposed amendments in anticipation of submission after the Delegate's meeting with the advocates, with the addition of the name change and the timeframe starting after the condition has been approved by the Secretary of Health for inclusion on the Maryland newborn screening panel.

Motion made by Dr. Fraser to approve the letter of support as written now, unless there are significant changes to the amendments, after Delegate Shetty meets with the advocates. Motion seconded by John McGing. Roll call vote taken with 6 yes and 0 no votes. Motion was also made by Dr Fraser to allow staff to make any technical changes necessary to the letter. Motion seconded by John McGing and approved.

Planned tentative meeting currently set for 3/3/2022 will remain scheduled pending response from the advocates. Dr. Fraser also recommended that future meetings be planned for every month or every other month to keep on track with the timeline.

Dr. Strovel will communicate the suggested changes and request the bill version from Delegate Shetty and forward the letter. John McGing motioned to adjourn and Dr. Fraser seconded.

Meeting adjourned at 7:01 PM

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

**March 9, 2022 5:30 PM**  
(Virtual meeting secondary to Covid-19)

**Members Present**

Erin Strovel, Chair  
Jamie Fraser, Vice Chair  
Robert Brosius  
Meral Gunay  
Sharon Dols  
John McGing  
Dominique Sessa

**Members Absent**

David Myles  
Michelle Smith  
Senator Edward Reilly  
Delegate Karen Lewis-Young

**Ex-Officio Present**

Johnna Watson  
Stacy Taylor  
Fizza Majid

**Ex-Officio Absent**

Robert Myers

**Guests**

Carol Greene, UMMS Genetics  
Paul Vetter, PerkinElmer Genetics

**MDH Staff**

Monique Veney

**I. WELCOME & ROLL CALL**

Roll call completed and quorum present.

Dr. Strovel, Council Chair, welcomed everyone to the meeting.

New Health-Unrelated member, Dominique Sessa, was introduced.

**II. LEGISLATIVE UPDATE**

Dr. Strovel shared that she had a phone call with Delegate Shetty last night. Del. Shetty agreed in principle with recommended changes, but she pushed back on the timeline concern. She feels the two-year timeline should start with the addition of the condition on the RUSP. However, she had not heard back from the advocates prior to this discussion. Proposed amendments, with language from the advocates, were shared with the Council. These have not been reviewed by MDH or the Delegate at this time. Dr. Strovel reports Delegate Shetty wants the Council's feedback regarding the language in the amendments.

The proposed amendments from the advocates were discussed and suggestions made from the Council for Dr Strovel to take back to Delegate Shetty. Original language presented to the Delegate clarifying the annual review process by the Council is recommended. For implementation, the Council recommends including staffing in the language, in addition to the procurement of equipment. Comments were placed in track changes in the original document to be sent back to Delegate Shetty.

Motion made by Dr Fraser asking Dr. Strovel to forward the document with the suggestions back to Delegate Shetty by email. Seconded by John McGing. Hearing no objections, Erin Strovel states she will forward the document to Delegate Shetty by email to incorporate into the proposed amendments.

Dr. Fraser moved to adjourn the meeting. John McGing seconded the motion. There is a tentative meeting scheduled for 3/10/2022 if needed.

Meeting adjourned at 7:06 PM

**State Advisory Council**  
**Hereditary and Congenital Disorders**  
**Minutes**  
**April 19, 2022**  
(Virtual meeting secondary to Covid-19)

**Members Present**

Erin Strovel, Chair  
Jamie Fraser, Vice Chair  
Gerald Raymond  
Michelle Smith  
John McGing  
Senator Edward Reilly  
Robert Brosius

**Members Absent**

Delegate Karen Lewis-Young  
Dominique Sasas  
David Myles  
Sharon Doyle

**Ex-Officio Present**

Fizza Majid  
Stacy Taylor

**MDH Staff**

Laportia Barrows  
Monique Veney

**Guests**

Allison Shaw  
Shelly Cho  
Carol Greene  
KeriAnn Kuperman  
Paul Vetter  
Delegate Emily Shetty

**Called to Order** – 5:38 PM. Meeting ground rules (meeting etiquette) were shared with the committee by Stacy Taylor.

**I. WELCOME**

Dr. Erin Strovel, Council Chair welcomed and thanked all for joining the call.

**II. APPROVAL OF MINUTES**

Draft meeting minutes sent prior to meeting for review. Motion to approve minutes made by Dr. Jamie Fraser and seconded by John McGing. No opposing votes. Minutes approved.

**III. MEMBERSHIP UPDATES**

In Johnna Watson's absence, Erin welcomed new council member Dr. Gerald Raymond who will be representing Johns Hopkins and taking over Dr. Meral Gunay's position. Dr. Raymond introduced himself and gave a brief background.

All positions are currently filled. Delegate Karen Lewis-Young's status needs to be confirmed.

Several terms are expiring June 30, 2022. Dr. Strovel announced her term being one. She will not be eligible for renewal. Jamie Fraser is up for renewal in June, 2022 and accepted the request to renew. Robert Brosius and Michelle Smith have also accepted renewal.

**IV. OLD BUSINESS**

- Delegate Emily Shetty introduced herself to the committee and thanked the Council for the quick turn around on feedback to make HB 109 significantly stronger. HB 109 passed, as did the Senate cross-filed companion Bill. Delegate Shetty stated they are waiting to hear if the Governor will sign it or allow it to become law without his signature.
- Dr. Strovel initiated discussion on a letter supporting coverage of expanded carrier screening. Dr. Fraser expressed the role of the carrier screening has become ever more important. The Council discussed and agreed that the letter is important and worthwhile to move ahead on. The

Council will follow up on the process. A vote at the last meeting was approved to write the letter.

- The Council discussed criteria selection process for adding conditions to the Maryland (MD) Newborn screening panel. It had been voted on and agreed that conditions shall be added to the Recommended Uniform Screening Panel (RUSP) prior to consideration to be added to the MD NBS panel. Dr. Strovel will do some follow up for more clarification.

#### **V. NEW BUSINESS**

- Council meeting scheduling was discussed. It was agreed to meet every other month and to add an additional meeting during Legislative Session if needed.
- Dr. Fraser introduced discussion on family experiences with newborn screening and introduced Allison Shaw, Children's National Hospital to present compiled data.
- Allison presented:
  - Lysosomal Storage Disorders Newborn Screening: Delays in Diagnosis in Maryland
    - Described current processes and proposed improvements. Comparison of how various States do screening. Explained the MD processes now.
  - Mucopolysaccharidosis I (MPS-1) NBS at Children's National Hospital. Visuals of outcomes were shared. What comes after Newborn Screen. Pompe Disease and screening cases were shared. Benefits of an automated system along with resources were shown.
- The council discussed bettering services to the families, consent, follow up and barriers.

#### **VI. UPDATES**

- **MCHB** (Stacy Taylor)
  - No updates at this time.
- **Laboratory** (Dr. Fizza Majid/Dr. Robert Myers)
  - Dr. Myers mentioned that projected timing of reassuming normal operations in the lab is beyond their control. Everyone is working overtime to get restored. No firm timeline while restoration is being worked on.
  - Dr. Majid added they are working with the restoration team and before they can go live, pieces will need to be added. Instruments and some test will need to be re-validated.
  - Dr. Majid shared with the Council that once they are live, it would be much easier to update X ALD projection implementation.
  - Dr. Greene asked would NBS like a letter to be sent to the Secretary recommending the NBS Lab is given advances on permanent positions. Dr. Majid and Dr. Myers both welcomed that suggestion. Dr. Fraser motioned to write a letter supporting the lab and Dr. Raymond seconded the motion. The Council voted and it was approved to write the letter to the Secretary in support of the NBS Lab.
- **Newborn Screening Follow-up** (Monique Veney)
  - NBS is continuing to operate status quo.
  - They referred and identified their first MPS-1 baby in the State of Maryland.
  - Johnna has been able to go in the lab to work through Starlims to isolate some older reports and work on the block grant.

#### **VII. FUTURE TOPICS FOR DISCUSSION**

- Dr. Strovel recommended discussing the most recent addition to the RUSP which is MPS 2 at the next meeting.

#### **VIII. NEXT MEETING DATE**

- June 14, 2022 was unanimously proposed as the next meeting date.

#### **IX. ADJOURNMENT**



- Jamie Fraser moved to adjourn, motion seconded by Michelle Smith. Meeting adjourned @ 7:18 PM.

# State Advisory Council on Hereditary and Congenital Disorders

## Minutes

June 21, 2022

(Virtual meeting secondary to Covid-19)

### Members Present

Erin Strovel, Chair  
Jamie Fraser, Vice Chair  
Robert Brosius  
Gerald Raymond  
Robert Brosius  
Dominique Sessa  
Sharon Dols  
John McGing

### Members Absent

Delegate Karen Lewis-Young  
Senator Edward Reilly  
David Myles  
Michelle Smith

### Ex-Officio Present

Fizza Majid  
Stacy Taylor  
Johnna Watson

### Ex-Officio Absent

Robert Myers

### MDH Staff

Laportia Barrows  
Monique Veney

### Guests

Carol Greene  
Paul Vetter

Called to Order –5:35 PM

### I. WELCOME & ROLL CALL

Meeting ground rules (meeting etiquette) were shared with the committee by Dr. Erin Strovel. Johnna Watson conducted roll call.

### II. APPROVAL OF MINUTES

Draft meeting minutes sent to committee prior to meeting for review. Motion to approve minutes made by Dr. Erin Strovel and seconded by Dr. Jamie Fraser. No opposing votes. Minutes approved.

### III. MEMBERSHIP UPDATES

- Dr. Strovel announced Senator Reilly is not returning to the Senate and will no longer be attending the Council meetings. She recognized him for all his help over the years with the Committee.
- Delegate Karen Lewis-Young's status was unclear and Johnna Watson gave an update on that. Delegate Lewis-Young requested to be off of the Council. She put in a request to the speaker to have Delegate Shetty become a member of the Council. Dr. Strovel acknowledged Delegate Lewis-Young as well for all her help over the years.
- Robert Brosius has renewed his membership.
- Dr. Jamie Fraser is in the process of renewing her membership.
- Michelle Smith status is unclear. She has been sent reminders to renew her seat.
- Dr. Strovel stated her nine year term is expiring and ineligible for renewal. She recommended some replacements. In a month or two hopefully, someone will be appointed to represent University of Maryland in an official capacity.

- The Council held nominations and votes for new Chair and Vice Chair.
  - Dr. Strovel nominated Dr. Jamie Fraser for Chair and Robert Brosius for Vice Chair
  - Dr. Raymond seconded the nominations.
  - Vote taken and both positions were approved.

#### **IV. OLD BUSINESS**

- Dr. Strovel announced to the Council that Taryn Couture from Access to Equitable Carrier Screening reached out about the status of the letter regarding expanding the carrier screening coverage on MD Medicaid. She explained the letter would be addressed and provided directly to the Secretary of Health. After the letter is completed and sent to the Secretary, a copy can be provided to the advocacy group.
- The Council voted back in December to write the letter regarding the need to update the coverage policy to include expanded carrier screening. Dr. Strovel asked the advocacy group to send an outline of pertinent points to facilitate writing a draft. Dr. Strovel will try to get a draft together and send it out to the group for review. The group was planning to meet again with Medicaid in January 2022. It is not known at this time if the meeting actually took place so Dr. Strovel will follow-up regarding this question. Dr. Fraser asked if Dr. Strovel would be willing to continue working on this issue after her term ends, and Dr. Strovel agreed to see this through.
- Dr. Strovel also mentioned drafting a letter regarding lab positions. A need for permanent positions had been discussed previously with the lab. Dr. Strovel requested that Dr. Majid provide an update during the laboratory update.

#### **V. NEW BUSINESS**

- The Council discussed and made suggestions for the GAMT presentation and scheduling of either Dr. Lisa Kratz at Kennedy Kreiger or Dr. Sofia Saerz Ayala at University of MD. Presentation will be later in the fall and Dr. Strovel will reach out to Dr. Sofia Saerz Ayala regarding providing the presentation.
- Dr. Raymond will present his MPS-II presentation at the next staff meeting.

#### **VI. UPDATES**

- **MCHB** (Stacy Taylor)
  - No new updates at this time other than thanking the lab and the NBS follow-up staff for continuing to ensure babies are screened during the network security incident.
- **Laboratory** (Dr. Fizza Majid)
  - Dr. Majid reports there are no new updates except gearing up to go live soon. Dr. Fraser asked about lead time for notification of when the State lab begins to screen. Dr. Majid stated she should know about a week prior to resuming operations. The goal is to go live by the end of July.
  - Dr. Strovel asked if there is continued difficulty with getting positions filled. Dr. Majid states she has lists but no appropriately qualified persons are on the list.
  - Dr. Fraser asked if there is sufficient staff to go live currently. Dr. Majid reports the lab has recently lost two more staff members, but that will not impact going live. It will impact new testing going online.
  - Dr. Fraser asked if MPS-II could be implemented in the next year if approved. Dr. Majid said it could be started rather quickly because it is being added to a platform that is already in place.

- **Newborn Screening Follow-up** (Johnna Watson)
  - NBS follow up is moving along receiving a lot of results from PerkinElmer.
  - Trying to work with two databases and trying to see how much access they will have remotely to StarLIMs
  - Dr. Fraser asked about the delay in results currently and if that will continue once the lab is operational again. Johnna states the delay is related to processing the samples through PerkinElmer and this will be eliminated when the lab returns to screening.

#### **VII. NEXT MEETING DATE**

- September 13, 2022 was agreed upon as the next meeting date. Start time will be 5:30 PM.
- Dr. Fraser also asked to have a call list available and asked all members to send direct contact information to Johnna Watson via email.

#### **VIII. ADJOURNMENT**

- Dr. Erin Strovel moved to adjourn the meeting, motion seconded by Dr. Jamie Fraser. Meeting adjourned @ 6:20 PM.

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

**September 13, 2022**

(Virtual meeting secondary to Covid-19)

**Members Present**

Jamie Fraser, Chair  
John McGing  
Robert Brosius  
Gerald Raymond  
Shannan Dixon  
Sharon Dols  
Dominique Sessa

**Ex-Officio Present**

Fizza Majid  
Johnna Watson

**MDH Staff**

Monique Veney

**Members Absent**

Michelle Smith  
David Myles  
Senator Edward Reilly  
Delegate Karen Lewis-Young

**Guests**

Carol Greene  
Paul Vetter  
Erin Strovel

**Called to order- 5:30 PM**

**I. WELCOME & ROLL CALL**

Johnna Watson conducted meeting roll call.

Shannan Dixon was welcomed as the new representative from University of Maryland Genetics

**II. APPROVAL OF MINUTES**

Draft meeting minutes were sent out to committee prior to meeting for review.

Motion to approve minutes made by Gerald Raymond and seconded by John McGing.

No opposing votes. Minutes approved.

**III. OLD BUSINESS**

- Dr. Strovel updated the Council on the status of the letter regarding expanded carrier screening coverage through Maryland Medicaid. She was sent an outline from Taryn Couture that will assist in drafting the letter. It was agreed that Dr. Strovel will draft the letter. She asked Dr. Greene to assist with editing and then it will be sent to Johnna for distribution to the Council. The Council discussed the process of approving the letter after the completion. Robert Brosius made a motion to email completed letter for review by members and conduct a vote through email for approval and Dr. Raymond seconded the motion.
- Dr. Strovel was advised by Taryn Couture that the Maryland chapter of American College of Obstetricians and Gynecologists (ACOG) will ask MD Medicaid to cover

expanded coverage screening and a letter from this Council may help get MD Medicaid on board.

- Dr. Fraser asked Dr. Majid if she was still in need of the letter of support from the Council in regards to lab positions. Dr. Majid said six or seven positions are currently open but the State cannot compete with the money being offered to applicants. It was suggested the letter of support recommend incentives to applicants and permanent positions offered. The Council discussed the letter of support and what incentives the State of MD could offer applicants without resolution. The letter of support will be revisited the next staff meeting.

#### **IV. NEW BUSINESS**

- **Mucopolysaccharidosis Type II** – presented by Dr. Gerald Raymond
  - Mucopolysaccharidosis, also known as Hunter Syndrome was explained, including symptoms, frequency of the disorder and available treatment.
- Following the presentation, the Council discussed this rare condition and the new legislation passed in 2022. Under this legislation, this Advisory Council must review new core conditions added to the Recommended Universal Screen Panel (RUSP) within 1 year and make recommendations to the Secretary as to whether or not the condition should be added to the MD panel.
- Dr. Fraser asked to have MPS-II reviewed using the screening criteria developed by this Council last year. Johnna will distribute the screening criteria to the Council. Robert Brosius made a motion to table this discussion until the next meeting, and Dr. Raymond seconded the motion.

#### **V. UPDATES**

- **MCHB** (Stacy Taylor)
  - No new updates at this time in Stacy Taylor's absence.
- **Laboratory** (Dr. Fizza Majid)
  - Dr. Majid announced ALD will not be online by the end of the year because the lab still does not have the staff. They have the instruments but can't move forward without staff.
  - After the transfer back from Perkin Elmer, the lab is handling everything ok Dr. Majid stated.
- **Newborn Screening Follow-up** (Johnna Watson)
  - Johnna mentioned they are in the process of working from two databases and migrating babies back into the Starlims database.

#### **VI. NEXT MEETING DATE**

- November 1, 2022 was proposed as the next meeting date.

#### **VII. ADJOURNMENT**

John McGing moved to adjourn meeting, motion seconded by Sharon Dols.  
Meeting adjourned @ 7:05.



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

**December 20, 2022**

(Virtual meeting secondary to Covid-19)

**Members Present**

Jamie Fraser, Chair  
John McGing  
Sharon Dols  
Robert Brosius  
Gerald Raymond  
David Myles

**Ex-Officio Present**

Fizza Majid  
Robert Myers  
Stacy Taylor

**Ex-Officio Absent**

Johnna Watson

**Members Absent**

Dominique Sessa  
Shannon Dixon  
Michelle Smith  
House of Delegates seat (vacant)  
Senate seat (vacant)

**MDH Staff**

Lauren Whiteman  
LaPortia Barrows  
Monique Veney

**Guests**

Alexandria Bowen, UMD Genetics  
Carol Greene, UMD Genetics  
Paul Vetter, PerkinElmer Genetics  
Christina Grant, Children's Genetics  
Erin Strovel, UMD Genetics (late arrival)

**Called to order**

Meeting called to order at 17:40. Meeting ground rules (meeting etiquette) were shared with the committee by Chairperson Fraser.

**I. WELCOME & ROLL CALL**

LaPortia Barrows conducted meeting roll call.

**II. APPROVAL OF MINUTES**

Draft meeting minutes were sent out to committee prior to meeting for review. Chairperson Fraser asked were there any concerns or corrections to be made. Sharon Dols mentioned her name is incorrect. Motion to approve minutes made with correction by Dr. Raymond and seconded by John McGing. No opposing votes. Minutes approved.

**III. MEMBERSHIP UPDATE**

The Committee had no updates regarding membership for the vacant Delegate and State Senator seats. Stacy Taylor, MDH mentioned she hasn't heard of any updates but will reach out to their OSS to see what they can do to solicit some new members.



#### IV. OLD BUSINESS

- Chairperson Fraser mentioned the letters that were being drafted by Dr. Strovel related to coverage of prenatal carrier testing, but since Dr. Strovel is not in attendance, the status of the letters will be tabled.
- Dr. Raymond presented updated information to the Committee related to MPS-II using the criteria for inclusion on Maryland NBS panel.
  - Mucopolysaccharidosis Type II (MPS-II or Hunter syndrome)
    - Clinical features most commonly develop between 18 months – 4 years of age.
      - Coarse facial features
      - Frequent ear infections
      - Hepatosplenomegaly
      - Joint Contractures
      - Umbilical hernia
      - Short stature
    - Diagnosis cannot be made only on clinical features.
    - MPS-II is not characterized by acute crisis.
    - Condition is progressive. Lifespan for untreated severe is 10-20 years.
    - Establishing the diagnosis is based on absent or reduced iduronate-2-sulfatase (I2S) in plasma, leukocytes, fibroblasts or dried blood spots and there are well-established methods using fluorometric and mass spectrometry testing to measure the I2S.
    - Evaluation/monitoring is needed with multispecialty care regardless of whether identification of MPS-II is in newborn period or later
    - Enzyme replacement therapy (ERT) has been FDA-approved for >10 years.
      - Weekly IV dose
      - Improves somatic signs/symptoms of all individuals
      - Survival is improved
      - Does not cross the blood-brain barrier
      - Infusion-related reactions may occur with some rare severe events
    - Long term experience with ERT in MPS-II patients with a severe phenotype showed improvement/stabilization in the following areas
      - Liver and/or spleen size
      - Frequency of respiratory infections
      - Texture of hair and skin
      - Disease related hospitalizations
      - Skeletal and cardiac disease
    - Timing of initiation of ERT related to symptom development is demonstrated in several reports of siblings in which the younger sibling is diagnosed and treated earlier because the older sibling was already diagnosed. The younger siblings have less symptoms and higher developmental scores than their older siblings, even though ERT does not cross the blood brain barrier
    - There is an ERT that is currently being studied in Japan and Europe that is expected to cross the blood brain barrier.
    - Dr. Grant raised a question regarding the age differences in the siblings in the studies which are 3 to 7 years and whether the better developmental scores in the younger siblings are related to the lack of progression of the disease in the younger sibling. Dr. Raymond indicates the age of follow-up in the last study by Grant et al compares the development of the

- siblings at the same age, but it is possible the differences in ages at the time of the developmental follow-up could be a factor.
- Dr. Fraser asked if there is any further follow-up on these siblings because the most recent study presented is from 2014 and whether long term survival rate has been improved. Dr. Raymond states he is not aware of any currently but will investigate.
  - Continuation of the presentation indicates bone marrow transplant (BMT) may be showing increasing effectiveness in Hunter syndrome treatment.
    - Overall, experience is limited.
    - One case was presented which shows a child who was treated at 10 days of age with ERT and then at 70 days of age had hemopoietic stem cell transplant (HSCT) with umbilical cord blood from an unrelated donor. At 7 years of age, he has normal growth and motor development.
  - Newborn Screening and MPS II
    - Based on measuring I2S in dried blood spots (DBS)
    - May be multiplexed
    - Second tier screen using DBS GAGs is being used by most states that are currently screening for MPS II
    - Added to the RUSP in August 2022
    - Follow-up testing is available
      - Repeat enzyme and urine GAGs
      - Dried blood spot GAGs
      - DNA determination
    - Unknowns
      - Sensitivity of newborn GAGs for attenuated forms
      - Are pseudo deficiency determinations valid?
      - Value of extended family testing
    - Newborn Screening Population projections (data determined by experts studying data for inclusion on the RUSP)
      - Incidence is 1.6/100K so if all babies in US were screened
        - 480 positive screens nationwide
        - about 59 cases of MPS II
        - about 59 cases still undergoing diagnostic testing at 1 year of age
        - about 322 false positives
      - Without newborn screening, estimated only 24 cases come to attention clinically.
  - Dr. Fraser asked if anyone on the Council had questions.
    - Robert Brosius asked if the screening could be determined through other testing that is already conducted. Dr. Raymond states it can be multiplexed with the other LSD screens that are currently in progress but there is no other testing that could identify MPS II in the NBS
    - Dr. Grant raised the issue that Invitae has been discontinuing some of their sponsored panels for DNA and there may be a high pseudo deficiency rate for MSP II without a method to determine if they are pseudo deficiency or affected.
    - Dr. Raymond strongly recommends second tier testing at the newborn screening level to reduce the pseudo deficiency referral rate if and/or when screening is implemented. Dried blood spot GAGs would be helpful for both MPS-I and MPS-II.

- Dr. Fraser also states that molecular testing as second tier would be beneficial in reducing uncertainty and time of diagnosis.
- Dr. Grant states Children's is currently at a 300 to 1 referral rate for MPS I for referrals from Maryland. Dr. Fraser suspects the same referral rate may be seen in MPS II.
- Dr. Myles asked if the cost of treatment is a factor to be considered in the Excel spreadsheet or otherwise. Dr. Fraser states the cost of treatment specifically is not in the criteria but the benefits to society and family is included in that early diagnosis provides benefit to society, and so far, she does not feel we have enough data to show that treatment with ERT alone is enough to prevent mortality but may just prolong the natural history of the disorder. Dr. Raymond states there is definitely data that shows increased survivability and reduced morbidity with ERT. Dr. Fraser referred to a report reviewed prior to this meeting that shows mortality was improved just to 30 years of age and concluded that since the ERT does not cross the blood brain barrier there is still a shorter lifespan.
- Robert Brosius asked if ERT is supported by Medicaid and Medicare and response from Dr. Raymond is that no one has been denied ERT based on their ability to pay, with the exception of undocumented individuals.
- Dr. Fraser states she has a concern regarding lack of access to ERT that crosses the blood brain barrier and the limited study information regarding HSCT and the inability to perform HSCT in MD which should be a factor in not recommending newborn screening for MPS II at this time and should be reconsidered in another 2-3 years.
- Dr. Raymond states that MPS II is on the RUSP and he would advocate for it going forward but he does not see it getting implemented for another 3-4 years and other issues may be settled at that time.
- Dr. Grant raised the question that since MPS II would be multiplexed if the timeline for implementation would be relatively short to which Dr. Majid from MDH lab responded implementation for MPS II could be less than a year.
- Dr. Fraser states that specificity and sensitivity is too high for this condition. Dr. Raymond states that the same issue is present for MPS I. False positive rate has been high in Maryland for MPS I but some of that is due to the lack of second tier testing at the state laboratory level.
- Dr. Grant states that for other conditions with high referral rates there are tests that can be performed easily to rule out the condition versus molecular testing. She also discussed that there is more family trauma with false positives for a condition where the child may only live to 20 years of age versus a condition in which the child may have to be on diet therapy.
- Dr. Fraser led a line by line discussion regarding MPS II using the scoring criteria for selection for a condition on the MD screening panel.
- Floor was opened after review of the criteria using the scoring criteria.
  - Robert Brosius brought up the question of whether the scoring of the criteria has been re-evaluated after the removal of 3 criteria and he began the process of this re-evaluation.
  - John McGing asked if there are other family members that need to be tested based on the newborn's screening results to which Gerald Raymond responded yes since it is an x-linked condition. The follow-up testing for family members is relatively straightforward. However, Dr.

Fraser brought up the point that extended family members may not have enough insurance coverage for the follow-up testing.

- A motion to table further discussion for next meeting given time constraints made by Gerald Raymond and seconded by Robert Brosius. All members approved.

## **V. NEW BUSINESS**

- A proposed legislation regarding creation of rare disorders council was brought to the Council. Stacy shared that MDH received an email from Senator Clarence Lam's office considering introducing a Bill to establish a Rare Disease Advisory Council (RDAC) to study and make recommendations on issues that impact individuals with rare diseases. The National Organization for Rare Disorders (NORD) helps support and set up these councils in various States. NORD has information on their website about RDACs. Senator Lam's Legislative Assistant reached out as a courtesy to inform of this potential legislation.

## **VI. UPDATES**

- **MCHB** (Stacy Taylor)
  - Stacy Taylor informed the Committee that the Office for Genetics and People with Special Health Care Needs has a new office director, Lauren Whiteman. Lauren introduced herself to the Committee.
- **Laboratory** (Dr. Fizza Majid)
  - Dr. Majid shared with the Council that the lab has x-ALD instruments set up and ready to go. They will be active once staff starts training and will move forward after that. The lab has had some luck with staff but are waiting to see if they accept the open positions.
- **Newborn Screening Follow-up** (LaPortia Barrows)
  - LaPortia Barrows shared with the Committee that there is a new reporting system for the grant management system. The mid-year and full term reports will be in REDCap. Monthly meetings with the Genetic Centers will also be initiated to discuss and review current protocols this year.

## **VII. NEXT MEETING DATE**

- January 24, 2023 was proposed as the next meeting date.

## **VIII. ADJOURNMENT**

- John McGing moved to adjourn meeting, motion seconded by Sharon Dols. Meeting adjourned at 1910.