

State Advisory Council on Hereditary and Congenital Disorders

Minutes January 5, 2021

(Virtual meeting secondary to Covid-19)

Members Present

John McGing, Chair
Ben Smith
Hilary Vernon
David Myles
Jamie Fraser
Erin Strovel
Senator Edward Reilly

Members Absent

Marjorie Shulbank
Michelle Smith
Delegate Karen Lewis-Young
Robert Brosius

MDH Staff

Monique Veney
LaPortia Barrows

Ex-Officio Present

Johnna Watson (scribe)
Stacy Taylor
Fizza Majid
Robert Myers

Guests

Carol Greene
Danielle Pantalone, Cullari Communications

Called to Order – 5:04 pm

I. WELCOME

John McGing, Council Chair, extended a welcome to all attendees. In virtual meeting environment introductions around the table were not conducted. However, attendees attending by phone were identified.

II. APPROVAL OF MINUTES

Minutes from meeting on August 18, 2020 were approved and will be posted on website.

III. MEMBERSHIP UPDATES

- Johnna Watson reported Delegate Karen Lewis Young has asked to be taken off of this committee so a new delegate member will be sought.
- One health unrelated member position is open. Johnna received an email from someone interested in that position so an application will be sent out to them,

IV. OLD BUSINESS

- Letter re. genetic testing techniques not covered by Medicaid.
 - Johnna Watson reported that the letter has been completed and released to the secretary.
- Letter re. hiring new staff for the newborn screening laboratory to perform X-ALD screening
 - Discussion between Ben Smith and Dr. Majid about need for letter and details of need. Ben asked about a hiring freeze. Dr. Majid stated the lab has not received any new PIN's for positions at this time. The lab is down 5 positions and recruitment has been taking a while, but these positions are for regular lab operations, not for adding X-ALD screening. Ben asked if the letter should address the wider issue of manning the entire lab instead of just addressing staffing for testing of one disorder. Dr. Majid thinks the focus of the letter should be addressing the additional staff needed for X-ALD.
 - Ben will begin drafting this particular letter to be sent out to the committee for review.
 - Question from Jamie Fraser asking if staffing issues are why there is still no 2nd tier testing for MPS-1. Dr. Majid indicates development of 2nd tier testing is very

challenging when all positions are not filled. Staffing is needed to implement XALD and then the lab can start looking at 2nd tier testing, not only for MPS-I, but others as well. Erin Strovel states UMMS Biochemical lab is also having trouble hiring, particularly since private labs can pay higher.

V. UPDATES

MCHB

- Stacy Taylor reports Secretary Neal retired effective Dec. 1, 2020.
- Dennis Schrader is now the Acting Secretary who has been in this role in the past.
- Dr. Jinlene Chan is still serving as Acting Deputy Secretary for Public Health, which was the role Fran Phillips held before her retirement.
- There no specific updates on Covid-19 related matters, but just be aware there is a significant impact to all of our staff. We are not specifically involved in any of the vaccine issues, but Governor Hogan is holding a press conference currently about the vaccine updates so everyone is welcome to check out the summary after this meeting.

Laboratory

- Fizza Majid reports the lab is currently trying to keep up with the work due to staff shortage and no new PIN's. Discussion ensued about whether or not the State can increase pay to hire and keep staff. Decisions related to increasing pay are at a higher level within the State.
- Robert Myers alluded to the tremendous amount of Covid work being done by the lab. Specimen volume is 10,000 specimens a week, as opposed to normally 7,000 specimens weekly. There is a lot of sequencing work also happening with Covid to identify variants and a lot of staff is being pulled into this testing.
- Robert Myers further states that Dr. Majid is trying to acquire the mass spectrometers needed to implement X-ALD. The reserve fund may need to be tapped for this procurement, and after that, a fee increase may need to be asked for. An analysis will be done to see. The lab is trying to hire staff, but the job market for lab science people is pretty competitive now.

Newborn Screening Symposium

- Fizza Majid shared that she was able to attend part of the Symposium which focused on:
 - increasing second tier testing and sequencing
 - changing shifts around due to Covid so that Newborn Screening does not shut down because of concerns.
- Johnna Watson reports that NBS Follow-up had a workshop with other Follow-up units across the country and most teams are operating remotely successfully.

Advisory Committee on Heritable Disorders in Newborns & Children

- Johnna Watson shared that the meeting was held on December 1, 2020 and the committee is reviewing their processes on how they are scoring new conditions.

VI. Future Topics for Discussion

- Johnna Watson would like to review process for adding conditions to the Maryland newborn screening panel. Hilary Vernon and Erin Strovel will give a presentation at the next meeting regarding the criteria that was developed by a sub-committee of this council for adding conditions.
- Funding for the laboratory was brought up by David Myles. Further discussion ensued regarding funding for the laboratory and whether or not the Council should be involved with asking for more funding, particularly to add 2nd tier testing. John McGing recapped at the end of the discussion the need to address current issues and processes. Ben Smith will work on a letter to make a position statement on the need to hire staff to begin implementation of X-ALD screening. He hopes to have a draft within a week.
- John mentioned if any further topics for discussions come up, they can be submitted to Johnna, and they will be added to the agenda.

- Ben Smith asked for a presentation from Dr. Myles to see how the military handles newborn screening across different states. Discussion ensued regarding there are not only differences between military babies across the world but also between states which is the reason that the Federal Advisory Council has developed the recommended universal screening panel (RUSP) which creates a baseline that all states should include in their panels.

VII. NEXT MEETING DATE

April 20, 2021

VIII. ADJOURNMENT

John McGing moved to adjourn. Motion seconded by Jamie Fraser.
Meeting adjourned 5:56 PM

State Advisory Council on Hereditary and Congenital Disorders

April 20, 2021 Meeting Minutes

(Virtual meeting secondary to COVID-19)

Members Present

John McGing, Chair
Hilary Vernon
Michelle Smith
David Myles
Jamie Fraser
Erin Strovel

MDH Staff

Monique Veney
Scott Riley

Ex-Officio Present

Johnna Watson (scribe)
Stacy Taylor
Fizza Majid

Members Absent

Marjorie Shulbank
Robert Brosius
Delegate Karen Lewis-Young
Senator Edward Reilly

Guests

Danielle Pantalone, Cullari Communications
Emily McLoughlin, Baebies
Ada Hamosh

Called to Order – 5:05 pm

I. WELCOME

John McGing, Council Chair, extended a welcome to all attendees. In virtual meeting environment introductions around the screen were conducted.

II. APPROVAL OF MINUTES

Hilary Vernon made a motion to approve the minutes from the January 5th meeting. The motion was seconded by Jamie Fraser. No opposing votes were received so minutes were approved and will be placed on the website.

III. MEMBERSHIP UPDATES

Johnna Watson reported there are two health unrelated member positions open, one of which Ben Smith occupies. The other member, Marjorie Shulbank no longer wants to be a member of the council. Johnna will be reaching out to Genetic Centers and Sickle Cell families to see if there is anyone interested in being on the Council. She said if anyone knows anyone that may be interested to let her know. Jamie Fraser said she may have a few families who may be interested in serving on the Council.

One House of Delegates member is needed as well. David Myles' term is expiring in June and is eligible for renewal. He states he is interested in serving another term so Johnna will send him the link to reapply. Hilary Vernon's term is expiring in June and is not eligible for renewal. Johns Hopkins has submitted Dr. Gunay to be a part of the Council.

John McGing thanked Hilary Vernon for her service and David Myles for agreeing to another term. Jamie Fraser also expressed thanks to Hilary Vernon.

IV. OLD BUSINESS

Pending letter to Secretary regarding hiring of newborn screening laboratory personnel

- Draft was not presented to the Council as discussed at the last meeting.
- John McGing asked if there is still a need for a letter.

- Dr. Majid reported some of the lab positions have been unfrozen. A few positions have started recruitment. Hiring new staff and one additional staff member.
- Jamie Fraser motioned to table the letter on hiring additional NBS lab personnel at this time and John McGing seconded the motion. There were no opposing votes.

V. NEW BUSINESS

New Condition Review procedure discussion by Erin Strovel and Hilary Vernon (copy of criteria with suggested edits attached with the minutes)

- Erin provided a summary of the review of the criteria used over the past several years to determine which tests or disorders should be added to the newborn screening panel. Some suggestions may have been made after reading through them and they would love some feedback and discussion(s).
- Recommended changing the criteria from “Disorder must not present clinically within the first 48 hours of life” to “Disorder must not typically present clinically within the first 48 hours of life.” There have been MSUD (maple syrup urine disease) cases that clearly present in that time frame prior to a NBS result being available.
- Two bullet points were combined into one. “Must have a test that has cut-offs with acceptable sensitivity and specificity.”
- Discussion item: whether the cost of treatment should remain on the criteria because some of the recommended and provided treatments are costly, particularly for some of the newly added disorders. It was felt that it may be too difficult to determine a cut off cost for treatments.
- Proposed “Benefit of therapy” instead of “Simplicity of therapy” vs risk of harm of therapy.
- Proposed an asterisk be placed beside cost of screening and testing because ideally new screening tests can be included on an existing platform in the newborn screening lab and diagnostic testing costs can be offset by part of the State grants to the genetic centers.
- Hilary adds the main goal is simplification of the criteria list. The list will be distributed with edits made so the changes can be seen clearly. There was a brief literature review and there has not been any new publications regarding criteria for newborn screening since the list was first developed.
- John McGing suggests that the last two bullets related to cost of testing for the NBS lab and the diagnostic testing may be considered secondary criteria and not primary criteria.
- American College of Medical Genetics guidelines were shared as well. Discussion indicates that Cost of Treatment, Incidence of Condition and Simplicity of Therapy should be crossed off of these criteria as well.
- John McGing states the plan is to review the recommendations presented today and to discuss at the next Council meeting. The recommendations will be sent via email to Council members.

VI. UPDATES

MCHB (Stacy Taylor)

- Dennis Schrader has officially been confirmed as Maryland Secretary of Health.
- Stacy mentioned COVID as well as staffing challenges are causing delays in some operations and programs.
- The Maternal Child Health Bureau (MCHB) is currently engaged in strategic planning efforts. It will involve communication and request for input from external stakeholders so you may be asked to add some input.

Laboratory (Fizza Majid)

- They have put in the paperwork for procurement for equipment for X-ALD. So far, they have approval for one position and have requested two more positions.
- Jamie Fraser asked if there is a timeline for X-ALD at this time. Fizza Majid responds there is no timeline yet because there is no answer from procurement at this time for equipment.

Newborn Screening Follow-up (Johnna Watson)

- Johnna Watson has been working on the 2020 data for the MCHB block grant.

- In 2020, there were 53 babies identified with primary conditions on the newborn screening panel, excluding hemoglobin babies. Hemoglobinopathy is the biggest category out there, usually between 90-100 yearly

VII. FUTURE TOPICS FOR DISCUSSION

- Selection Criteria will be discussed at the next meeting.
- If there are other topics, they can be emailed prior to the next meeting

VIII. NEXT MEETING DATE

- John McGing proposed Sept 14, 2021 at 5pm as the next meeting date and time, with Sept. 21st as a back-up date if something comes up in the interim.

IX. ADJOURNMENT

- Jamie Fraser moved to adjourn. Motion seconded by Hilary Vernon. The meeting was adjourned at 5:46 PM

State Advisory Council Hereditary and Congenital Disorders Minutes

September 14, 2021

(Virtual meeting secondary to Covid-19)

Members Present

John McGing, Chair
Michelle Smith
David Myles
Jamie Fraser
Erin Strovel
Senator Edward Reilly
Robert Brosius
Meral Gunay

MDH Staff

LaPortia Barrows
Scott Riley

Ex-Officio Present

Johnna Watson (scribe)
Stacy Taylor
Fizza Majid
Robert Myers

Members Absent

Delegate Karen Lewis-Young
Marjorie Shulbank
Ben Smith

Guests

Allison Shaw
Ada Hamosh
Carol Greene
Kami Skurow-Todd
Paul Vetter

Called to Order – 5:11 PM, after quorum obtained

I. WELCOME

John McGing, Council Chair will extend a welcome later in the meeting to all attendees.

II. APPROVAL OF MINUTES

Motion to approve minutes from meeting on April 20, 2021 made by Senator Ed Reilly and seconded by Erin Strovel. No opposing votes so minutes approved and will be placed on website.

III. MEMBERSHIP UPDATES

Johnna Watson introduced new council member, Dr. Meral Gunay. Dr. Gunay is representing Johns Hopkins and has been working with NBS follow up for the past several years.

There are still two health unrelated positions open at this time. Ben Smith's term has expired, but he can still continue serving until new member is added. However, he has not responded to any efforts to contact hm.

The council is actively searching for new members, trying to go through different organizations to see what family members might be interested in joining the council. John McGing asked if anyone knows of anyone that may be interested to contact Johnna.

Jamie Frasier mentioned a PhD from the community that was not a health care provider applied for the open position but did not qualify. Johnna states that the applicant's qualifications were discussed with the Attorney General's office and the determination was that the applicant did not qualify for a health unrelated position. Stacy Taylor said she will reach out to the contact for the administration for councils and committees that handles recruitment efforts to see if anything can be done in regards to qualifications.

IV. ELECTION OF NEW CHAIR/VICE-CHAIR

John McGing announced Erin Strovel had been nominated as new Chair replacing him and Jamie Fraser nominated as new Vice-Chair. John opened the floor to anyone interested in nomination(s).

Since no additional nominations were received, Senator Ed Reilly motioned to unanimously support the two candidates and give them the honor of serving the committee. With no adverse comments, Erin and Jamie were acclimated.

V. OLD BUSINESS

Erin Strovel began her new seat with following up on the discussion of criteria selections.

- Council is looking at the process of adding conditions to the NBS panel.
- The main goal is simplification of the criteria. Literature research that was performed did not reveal any new criteria since development of this list.
- Council members received criteria through email on June 4th and no feedback was received.
- Senator Reilly asked if there was any place for an evaluation that talks about the cost savings of human stress and unknown issues that can be quantified in this formula? He also asked was there any way to factor in the long term benefit, the long term financial benefit and the non long term financial benefit to having these issues discovered at an early age? Erin states these factors are addressed throughout all of the criteria, however they are not quantifiable.
- There was also discussion prompted by Allison Shaw regarding inclusion of the condition on the federal Recommended Universal Screening Panel (RUSP) as part of the criteria for consideration for screening by Maryland. Discussion of the RUSP as being based on opinions from experts in newborn screening and public health and the regimented review of each condition which could be beneficial and simplify the burden on this Council. Data used for consideration of inclusion on the RUSP could be added to the information presented by the clinician who presents the condition to this Council.
- Erin proposed adding the criteria of being on the RUSP and taking another look at the document with this addition to simplify the process for consideration on whether to include new conditions on the Maryland panel. Jamie reports that the RUSP website has legacy data for each condition.
- Erin will send current criteria with RUSP addition to Jamie for review and then will send it to Johnna for distribution to the Council.

VI. NEW BUSINESS

Jamie Fraser presented on Ehlers-Danlos Syndrome (EDS) and Tyrosinemia type I case.

- Ehlers-Danlos Syndrome (EDS)
 - Newborn Screening (NBS) was summarized in general as a public health program in which conditions can be identified through biochemical markers via blood spot collection and treatment can be started early to reduce or eliminate adverse effects of the condition.
 - Ehlers-Danlos Syndrome (EDS) was explained in full. What it is, testing process, diagnosis, signs and symptoms. Visuals were shared. EDS is a group of connective tissue disorders. EDS does not present in the newborn period. Most affected individuals start to show symptoms when they are school aged or older. Diagnosis is based on clinical symptoms and usually requires DNA analysis.
 - EDS is a clinical diagnosis that is not amenable to NBS based on current accepted clinical practice and public health guidelines. There is no blood test for EDS that is conducive for inclusion in testing on the newborn screen.
- Tyrosinemia Type I
 - Biochemical attributes and break down of Tyrosine were briefly explained. Tyrosine is obtained through diet and the breakdown of Phenylalanine. Tyrosine is an amino acid which is used for making new proteins, melanin, thyroxine and catecholamines. The

inability to break down tyrosine can cause liver and kidney damage. A byproduct of the inability to break down tyrosine is succinylacetone.

- Treatment involves limiting tyrosine in the diet and use of nitisinone to prevent the most toxic metabolites from forming.
- Symptomatic presentation involves acute liver failure, failure to thrive, hepatosplenomegaly and cirrhosis or cancer of the liver, as well as kidney disease.
- On the NBS, succinylacetone is recommended as 1st tier testing.
- A contrast of 2 cases (babies) were presented.
 - 1st baby had initial normal NBS and no repeat screen. Parents were known to be second cousins. At 1 year of age, failure to thrive and abnormal liver function with lesions on the liver, requiring a liver transplant.
 - 2nd baby was identified at 3 weeks of age on NBS using succinylacetone. At 1 year of age, normal liver function and normal growth and development.
- If succinylacetone is used as primary marker for Tyrosinemia on NBS, better job of helping patients in care and keeping them in care would have made a better outcome for the babies, Representatives from all 3 genetic centers concurred that succinylacetone should be added as a primary screen for tyrosinemia type I.
- Fizza Majid announced succinylacetone as a primary marker will be available very soon. Timeline is not currently available.

VII. UPDATES

- **MCHB** (Stacy Taylor)
 - With the increasing case numbers for RSV significantly higher, Synagist has been pre-authorized through Medicaid starting September 13th. It was previously not pre-authorized until Nov. but through the combined efforts of a few offices within the Maternal and Child Health (MCH) bureau working with Medicaid, it will be pre-authorized earlier this year. Memo will be sent to Johnna to share.
- **Laboratory** (Fizza Majid/Robert Myers)
 - Lab manager position has been filled. His name is Kevin Brown with 33 years of quality assurance experience with Quest labs.
 - X-ALD procurement documents have been submitted in April to OPAS. No confirmed date to place order. No timeline available at this time. Discussion ensued regarding the length of time since the documents have been submitted.
- **Newborn Screening Follow-up** (Johnna Watson)
 - LaPortia Barrows has come up on her NBS follow-up one year anniversary.
 - At the last Federal Secretary's Advisory Council meeting, GAMT, which was presented at a meeting about 2 years ago, was pushed into full review, which is a 9 month process.

VIII. Future Topics for Discussion

- Request to present by an organization at the next meeting regarding the recent American College of Genetic and Genomes Practice Guidance screening was discussed. General consensus was agreed upon to invite the organization for a brief presentation at the next meeting.

IX. NEXT MEETING DATE

- December 7, 2021 was unanimously proposed as the next meeting date.

X. ADJOURNMENT

- John McGing moved to adjourn, motion seconded by Erin Strovel. Meeting adjourned @ 7:05 PM

State Advisory Council Hereditary and Congenital Disorders Minutes

December 7, 2021

(Virtual meeting secondary to Covid-19)

Members Present

Erin Strovel, Chair
Jamie Fraser, Vice Chair
Meral Gunay
Michelle Smith
John McGing
Sharon Dols
Robert Brosius
David Myles

Members Absent

Delegate Karen Lewis-Young
Senator Edward Reilly

MDH Staff

Monique Veney
Laportia Barrows

Roll Call

I. WELCOME

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

II. APPROVAL OF MINUTES

Motion to approve minutes from meeting on September 14, 2021 made by Michelle Smith and seconded by John McGing. No opposing votes so minutes approved and will be placed on the website.

III. MEMBERSHIP UPDATES (Johnna Watson)

- There is still one Health Unrelated position open.
- New Health Unrelated member Sharon Dols introduced herself to the Council.
- Dr. Strovel's term will expire June 30, 2022.
- Delegate Karen Lewis-Young has expressed that she wishes to be replaced on the Council so there will be one House of Delegates member needed.

IV. NEW BUSINESS

Presentation from Access to Equitable Carrier Screening Coalition
(Taryn Couture and Katelyn Sagaser).

- The Access to Equitable Carrier Screening (AECS) Coalition is a multi-stakeholder coalition focused on ensuring all women of child bearing age and their partners have access to expanded carrier screening (ECS). They work closely with some advocacy groups.
- American College of Obstetricians and Gynecologists (ACOG) carrier screening guidelines have historically placed a person's ethnicity at the center. Limitations to ethnicity-based carrier screenings exist causing a large percentage of potential carriers of inherited disease to be missed because self-reported ancestry can be inaccurate.

Ex-Officio Present

Johnna Watson
Stacy Taylor
Robert Myers

Ex-Officio Absent

Fizza Majid

Guests

Katelyn Sagaser, Hopkins Genetics (formerly)
Allison Shaw, CNMC Genetics
Carol Greene, UMMS Genetics
Paul Vetter, PerkinElmer Genetics
Taryn Couture, Powers Law

- Expanded Carrier Screening (ECS) offers carrier screening for more childhood onset diseases to expecting parents and parents considering pregnancy regardless of ethnicity or family history giving them the opportunity to identify their potential at risk offspring to make good decisions for their future.
- American College of Medical Genetics and Genomics (ACMG) Practice Resource, 2021, recommends carrier screening paradigms should be ethnic and population neutral and more inclusive of diverse populations.
 - There are 4 tiers of carrier screening proposed and ACMG is recommending all pregnant patients and those planning pregnancy should be offered Tier 3 screening.
 - Tier 3 is screening for conditions with carrier frequency $\geq 1/200$ “in *any ethnic group* with reasonable representation in the United States”
- According to available data, 75% of people eligible for MD Medicaid identify as other than white.
- Offering Tier 1 screening, which is for cystic fibrosis and spinal muscular atrophy only, for Medicaid recipients is not providing adequate screening for non-white patients.
- MD Medicaid currently does not have a written policy for carrier screening. There are a few codes on the fee schedule for single gene screening, but there is no code for an expanded carrier screening.
- AECS is currently engaging with Maryland Medicaid to consider updating coverage policy to include expanded carrier screening.
- AECS is asking for a letter of support from the State Council on Hereditary & Congenital Disorders to bolster their current efforts with Maryland Medicaid.
- Following the presentation, the Council discussed the topic. Motion to write a letter was made by Dr. Fraser and seconded by Dr. Gunay. All voting members present voted in favor of writing a letter. A writing group consisting of Dr. Strovel, Allison Shaw, Dr. Fraser and Dr. Greene was established to complete the letter.

V. OLD BUSINESS

- Discussion of criteria selection process for adding conditions to the MD Newborn screening panel.
 - Dr. Strovel proposed this be pushed to the next meeting. The most recent document was sent to Dr. Fraser just today. Plan is to distribute the document out to other members prior to the next meeting.

VI. UPDATES

- **MCHB** (Stacy Taylor)
 - If you have anything that needs to be brought to the attention of the Department while Johnna is on leave, please email Stacy.Taylor@maryland.gov
 - The MD Department of Health has recently experienced a network security incident. MDH servers are offline. The MDH website is back up now, but internal servers are currently offline. The impact is being assessed and additional information will be provided as it becomes available. Newborn Screening (NBS) has been identified as the highest priority system to return to service from the Public Health Services Unit.
- **Laboratory** (Dr. Robert Myers)
 - A recovery team is presently working at the MD Department of Health (MDH) lab trying to reconfigure their servers so that they can resume testing operations in the lab.
 - The lab is currently making arrangements to have specimens shipped to PerkinElmer to be tested. Paul Vetter of PerkinElmer said they can handle the lab's capacity of specimens. The process, logistics and trying to get an agreement in place are being discussed.
- **Newborn Screening Follow-up** (Johnna Watson)

- During this MDH network incident, Newborn Screening Follow-Up has been setting up systems that they can follow up with babies with a paper trail. Their biggest hurdle is identifying babies that currently are in the database to determine if follow-up has been completed.

VII. Future Topics for Discussion

- Comprehensive LSD screening and cut offs.
- Follow up discussions on future emergencies. Back up plans and keeping track of said back up plans.
- X -linked ALD screening status
- Update on a start date for using succinylacetone for tyrosinemia type I

VIII. NEXT MEETING DATE

- April 19, 2022 was unanimously proposed as the next meeting date.

IX. ADJOURNMENT

- John McGing moved to adjourn, motion seconded by Jamie Fraser. Meeting adjourned @ 7 PM