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

**Minutes October 22, 2019**

**Members Present** **MDH Staff**

John McGing, Chair Jed Miller (phone)

Michelle Smith Jennifer Taylor

Hilary Vernon (phone) Linda Lammeree, (scribe)

Delegate Karen Lewis-Young (phone) Monique Veney

Erin Strovel **Ex-Officio Present**

Sarah Viall (phone) Robert Myers

 Fizza Majid

 Johnna Watson

 **Guests**

**Members Absent** Paul Vetter, (phone)

Ben Smith Ann Moser

Senator Ronald Young Hannah Baer

David Myles Sarah Hash

Rebecca Furman Dan Shattuck

 Dr Carol Greene

 Jasmine Kretzer Noronha

 Francis Rossignoc

**Called to Order** – 5:18 pm

**I. Welcome and Introductions**

Members and guests introduced themselves. Guests Sarah Hash and Hannah Baer are representing Coalition for Access to Prenatal Screening and will be presenting at the meeting this evening.

**II. Approval of Minutes**

Minutes from meeting on April 23, 2019 were approved and will be posted on website.

**III. New & Old Business**

Presentation: Coalition for Access to Prenatal Screening

* Sarah Hash, certified genetic counselor, and Hannah Baer, research associate, representing the Coalition for Access to Prenatal Screening presented evidence supporting the adoption of noninvasive prenatal screening and how policy changes can ensure all women have equal access to prenatal screening. The power point presentation was sent to Johnna Watson, Chief, NBS Follow up. The presenters shared the following key points:
	+ Cell free DNA based noninvasive prenatal screening is increasingly utilized across all pregnancy risk groups. Since 2011, it has offered improved detection of fetal chromosomal abnormalities.
	+ Cell free DNA based noninvasive prenatal screening is extensively studied in the general population (15+ studies with 88,000+ patients) and shows a very high positive predictive value compared to traditional screening with equal or better negative predictive value for all major aneuploidies.
	+ Noninvasive prenatal screening provides better detection of Trisomy 21, 18, and 13. Its lower false positive rate, compared to traditional screening, leads to fewer invasive follow up procedures and procedure-related losses.
	+ All major professional societies endorse or recognize cfDNA-based noninvasive prenatal screening as a clinically valid screening option for all pregnancies.
	+ There is a clear disparity in access to noninvasive prenatal screening for many women enrolled in Maryland Medicaid. There should be a single standard of high quality care of all pregnant women.
* The Coalition is asking for the Council’s support in the form of a letter of support to the State Medicaid program advocating for access to noninvasive prenatal screening for all pregnancies, not just high risk pregnancies.
* General discussion following the presentation indicated a letter of support could be seen as consistent with scope of Advisory Council. However, further information would be needed before a letter could be considered further, since an effective letter of support would need to address why access is not currently included and address those points, including fiscal, social, and political ramifications. The Coalition was invited to re-address the Council in the future with this information.
* Dr Majid provided an update on the new screenings added to the Maryland NBS panel as was requested at last Advisory Council meeting:
	+ SMA (Spinal Muscular Atrophy) screening commenced May 30, 2019. A total of 31,900 infants have been screened for SMA. Dr. Majid discussed the status of presumptive positive screens and confirmed cases.
	+ Screening for Pompe disease, Fabry disease and Mucopolysaccharidosis Type 1 (MPS 1) started June 17, 2019. Approximately 25,769 screenings for lysosomal storage disorders have been performed.
	+ Dr Majid was pleased to announce that second tier testing for Cystic Fibrosis (CF) will begin in early 2020. Infants with elevations in the immunoreactive trypsinogen level on two screenings will then have DNA analysis performed for common CF mutations.
	+ In addition, Dr Majid reported that second tier testing for Pompe disease (DNA sequencing) and MPS 1 (glycosaminoglycans assay) may also be available in 2020 as a send out to another lab. Second tier testing may help reduce the referrals to the genetics center.
	+ As was discussed at last Advisory Council meeting, the lab has performed a cost analysis for X-linked Adrenoleukodystrophy (ALD) screening and is estimating it will cost between $10-15.00 per infant. This will cover the cost of additional instruments and staff for X-linked ALD screening. It will be necessary to request a fee increase for newborn screening which could add one year to the implementation time frame.
* There was discussion regarding confirmed cases of any of the lysosomal storage disorders.
	+ Johnna Watson, Chief, NBS Follow up, reported approximately 24 infants referred to genetics centers for possible Fabry disease, 70 referrals for possible Pompe disease and 45 referrals for possible MPS 1.
	+ So far, there are several cases of possible late onset Pompe disease and no confirmed cases of infantile onset Pompe disease. There are no confirmed cases of MPS 1 to date. Sarah Viall, CNMC Genetics reported that there has been one case of possible late onset Pompe disease in Washington, D.C., which started screening in 2017. Virginia started screening in Jan 2019 and has had one confirmed case of MPS1.

**IV: Member Updates**

* Membership Update
	+ Currently there are two health unrelated vacancies in the Advisory Council membership. Additionally, it is likely that Senator Young may not be able to continue his position on the Advisory Council and so there will likely be a new legislative member vacancy.

* + Johnna Watson reported that the statutory authority for the Federal Advisory Committee on Heritable Disorders in Newborns and Children expired as of September 30, 2019. Currently there is no standing committee or work group. There is more information on their website.
	+ Johnna Watson stated that an email has been received indicating the Attorney’s General office has advised that members of the Advisory Council must be physically present at the meeting in order to vote. There was discussion about the impact this has on accomplishing even relatively uncomplicated tasks such as meeting minutes approval. Johnna also mentioned that a recent draft on attendance requirements at advisory committees permits attendance by phone. The Advisory Council would like more clarification on this issue.

**V. Next Meeting Date:**

Next meeting is planned for April 21, 2020 at 201 W Preston St.

**VI. Adjournment**

Meeting adjourned at 6:22 PM.