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Joint Advisory Committee Maine Newborn Bloodspot Program Meeting Minutes

Wednesday, October 13, 2021 1:30-4:00 p.m. ZOOM

Attendance: Tom Brewster, Abigail D'Ambruoso, Megan Dumas, Megan Haynes, Nola Metcalf, Colleen McCorkell, Jean Moreno, Jodi Philippon, Holly Richards, Wendy Smith, Dan Sobel, Kassi Swallow, Anne Watson, Melissa Whitcomb, Ana Cairns

Guest: Elizabeth Hall (medical student)

Welcome and Introductions	Introductions	Abigail D'Ambruoso
Committee Business	<p>Review and acceptance of minutes from 5-12-21 meeting</p> <ul style="list-style-type: none">Minutes were unanimously approved. Kassi made the motion and Jodi seconded. <p>2020 Infographic is now posted on the Maine NBS Bloodspot website.</p> <p>Membership</p> <ul style="list-style-type: none">Anne stated we still need to get another parent to join.New member will be Dr. Sandra Ilaka-Chibuluzo, Pediatric Endocrinology at NL EMMC, replacing Dr. Ann Boniface <p>Refusals</p> <ul style="list-style-type: none">Anne states a majority are coming from home births. The program will continue to track. <p>Confidentiality Agreements</p> <ul style="list-style-type: none">Worked with the Attorney General's office, these are not required. This is a public forum; we can go into a private session if we need to discuss something confidential.	Abigail D'Ambruoso Anne Watson
New Conditions	4 new conditions added April 1. Holly stated providers are paying the increased fees for the filer papers and she has only had a few midwives complain. The refusal rates don't appear to be increasing despite the increase in fees.	Holly Richards Anne Watson Jodi Philippon

	<p>Anne was contacted by a member of NERGG concerning supporting communities with higher rates of refusal due to financial issues. We discussed how financial issues extend to any needed follow-up and treatment in the context of access to care. She will be continuing this discussion with NERGG in December.</p> <p>Jodi reported a couple of possible pseudo-deficiencies for MPS-I that are being investigated, one X-ALD case identified (with siblings that have the disorder or a related disorder), and 1 possible late-onset Pompe. No OORs for SMA.</p> <p>Wendy Smith noted that the OORs in the immigrant population are more difficult to sort out due to how variants may emerge in these groups. In addition, Wendy Smith has had discussions with Anne Comeau at the UMMS lab about the detail in the reports for the new conditions. Wendy and Megan will use the report mostly as a screen and then proceed to do their own work-up. The rapid expansion of the technology for screening will continue to be an issue to be discussed. We are lucky that the UMMS lab is doing the DNA analyses so that insurance company authorizations do not need to be as involved in follow-up. But the detail can be cumbersome. Wendy has concerns that the lab is devoting too many resources to analyses that are not useful to the providers. Follow-up may be needed with the other states participating in NENBS and the lab itself.</p>	
<p>Maine, National and Clinic Updates and Highlights</p>	<p>Anne provided a brief summary of NBS Current/Future Needs that were discussed at the HRSA Virtual NBS Stakeholders Engagement Meetings. Issues raised were 1) need for more detailed and organized long term follow-up, 2) interoperability across health care systems, labs, and screening programs, 3) concerns about lack of resources for both staff and operations, 4) health equity – difficulty navigating the health care system for non-English speakers, insurance problems with adequate coverage of needed treatment, and access issues for rural areas and low SES families.</p> <p>Tom Brewster talked about recent legislation that was passed (LD 1601) that created a gene-editing research advisory panel and the development of a Precision Medicine and Genomic Medicine program at Maine Med. The bill was introduced by physician Sam Zager who has been following technology advancements (e.g., CRISPR) that will enable gene editing to treat genetic disorders like inborn errors of metabolism. Issues involve how this new technology will be successfully integrated with existing systems, the ethics of both disease treatment and modification of the genome of not only humans, but plant and other</p>	<p>Wendy Smith Meghan Dumas Holly Richards Anne Watson</p>

	<p>animal life. These are concerns being raised in all states. Wendy reported that national metabolic groups believe that biochemical markers of genetic disorders will remain extremely useful in screening and diagnosis given the complexity of genetic variants and their expression. Holly and Anne will contact the group established by the legislation and Anne will send out the materials that Tom discussed.</p> <p>Dr. Ana Cairns presented information on a recent CF Foundation meeting, specifically a session on quality improvement and education. An overriding theme involved educating PCPs on CF: multiple types of CF, when to call for sweat tests, state program workflows, lack understanding of IRT levels, inconclusive sweat chloride testing, low SES families may have difficulty with access to care, transformative effect of new medication in treatment, need for full CF mutation testing in father where the mother has CF (involve OBs too), complete sequencing in NICU babies, additional mutations added to CF panel. Ana has offered to provide a multidisciplinary grand round to MMC that could be advertised and sent out to the whole state. Get the word out that the outlook for CF is much more positive than before.</p> <p>Discussion around difficulty with follow-up on an abnormal screen by a provider. Outstanding effort on the part of everyone in NBS on this case.</p>													
NBS Program	<ul style="list-style-type: none">• We promoted NBS Screening Month in September through CDCs Facebook and twitter pages. Holly will send what was shared when the minutes go out.• Holly noted that COVID still is very much the focus at the health department but currently staff are not redeployed to COVID work related activities but could be deployed at any moment,• The program’s medical secretary, Amy Fair left in July 2021 and we are still actively recruiting to fulfill this position. Holly thanked the clinic for their patience as our program works to complete those tasks along with everyone’s current job responsibilities. <p>APHL Data Repository/Confirmed Cases 2020 – Preliminary</p> <table><tr><th>Conditions</th><th>N</th></tr><tr><td>Congenital Adrenal Hyperplasia</td><td>1</td></tr><tr><td>Congenital Hypothyroidism</td><td>23</td></tr><tr><td>Cystic Fibrosis</td><td>4</td></tr><tr><td>PKU</td><td>3</td></tr><tr><td>Hb-SS</td><td>4</td></tr></table>	Conditions	N	Congenital Adrenal Hyperplasia	1	Congenital Hypothyroidism	23	Cystic Fibrosis	4	PKU	3	Hb-SS	4	Holly Richard and Anne Watson
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	Classic Galactosemia	1	
	Total	36	
	Jodi presented preliminary 2021 confirmed case data.		
Public Comments	<p>Dates for upcoming meetings (Meetings are the 2nd Wednesday from 1:30-4 in May & October)</p> <ul style="list-style-type: none"> • May 11, 2022, 1:30-4:00 pm • October 12, 2022 1:30-4:00 pm 		
Wrap-Up			Anne Watson