

Newborn Screening Advisory Committee Meeting

Meeting Minutes of Tuesday, April 25, 2017

1:00-4:00

Wilder Center

Auditorium A - B

451 Lexington Parkway North

Saint Paul, MN 55104

Committee Members Present:

Bob Jacobson	Jan Larson	Renee Temme	*Theresa Hirte-
Dieter Matern	Annamarie	Jennifer Ward	Runtsch
Amy Karger	Saarinen	Kristin Loncorich	
Vikas Bhambhani	Emilee Scheid	Courtney Jarboe	
Steve Johnson	Kevin Sheridan	Kiki Sarafoglou	
Susan Berry	Kathy Stagni	Steve Nelson	

*Non-voting representative sitting in for Julie Thompson Larson due to retirement

Committee Members Absent: (N = Notification that s/he would be absent)

Roshini Araham (N)	Mark Bergeron (N)	Jenny Jacobson	Kristi Borowski
Kirsten Dummer (N)	Sheyhan Gelle	Ed Jenkins (N)	Terri Laguna (N)
Martha Overby			

Guest Speakers:

Amy Hietala, MS	Joanne Bartkus, PhD	Carrie Wolf, MPH	Peter Karachunski, MD
Joline Dalton, CGC	Carissa Keister		

- 1) Welcome and Introduction
 - a. Jan Larson (Chair) welcomed committee and guests
 - b. Announcement: Theresa Hirte-Runtsch is here in place of Julie Thompson Larson due to her retirement and will be standing in as a non-voting member to represent birth hospitals on our committee.
- 2) Review and approval of minutes for October 2016
 - a. Review of minutes from October of 2016. It was noted that Jenny Bock was now known as Jenny Jacobson. Steve Nelson motioned to approve minutes with correction. Sue Berry seconded. No discussion. Vote: Ayes, motion carries.
- 3) New Disorder Update
 - a. Amy Hietala, MDH Newborn Screening Laboratory supervisor, provided an update to the Committee regarding the addition of three new disorders.
 - i. On April 18, 2016, the Commissioner of Health accepted the recommendation of this committee and added X-linked adrenoleukodystrophy (xALD), Pompe, and mucopolysaccharidosis I (MPS I) to the Minnesota newborn screening panel.

- ii. In June 2016, the lab finalized a contract, and received three tandem mass spectrometers and reagents. The instruments were installed by PerkinElmer, scientists were trained, and the lab began validating for all three conditions in October 2016.
- iii. Pompe and MPS I validation was delayed due to intermittent hardware issues with the primary instrument (which was rebuilt the last week of March 2017). Validation began again on April 2, 2017. It is estimated that MDH will begin routine screening by the end of June 2017.
- iv. xALD validation was completed in January 2017 and screening began on February 6, 2017. As of April 19, 2017 the lab has screened 14,332 babies for xALD. Six borderlines (all male), five of these normalized upon repeat, one repeated as borderline again and was referred to specialist. Four positives (two males confirmed to have xALD, 1 female and 1 male pending follow-up) have been found.

4) Legislative Updates

- a. Joanne Bartkus, Division Director of the Public Health Laboratory, provided a legislative update.
 - i. The MDH Bonding bill is still uncertain, but includes a NextGen sequencing instrument for the public health lab.
 - ii. Policy bills of interest to this committee and introduced after deadlines had passed:
 - a) [House 2497](#), authored by Rep. Moran (65A), bill to establish a G6PD (Glucose-6-phosphate dehydrogenase) awareness program.
 - b) [House 2574](#), authored by Reps. Dean (38B), Albright (55B), Zerwas (30A), Youakim (46B), and Murphy (64A), bill to establish an advisory council on rare diseases to advise the Commissioner of Health of issues related to rare diseases (including research, treatment, education).
 - iii. Policy Bills—Not Yet Introduced:
 - a) Coverage for metabolic disorder nutrition
 - b) [House 2653](#), bill to direct the Commissioner of Health to educate healthcare providers and the public about cCMV.
- b. Discussions and questions by the committee.

5) State Nomination Process Discussion

- a. Maggie Dreon provided information to the Committee regarding the current approach and potential options for a state nomination process. Discussed the current approach employed by MDH, a possible RUSP alignment only approach, and a possible Minnesota specific nomination process.
- b. Discussions and questions by the committee.
- c. The committee decided to form a workgroup to explore possible ideas for a Minnesota specific nomination process.

6) Landscape of Spinal Muscular Atrophy (SMA) Panel Discussion

- a. Carrie Wolf, MDH Newborn Screening Laboratory supervisor, provided information about the current screening methods for SMA and what the lab has found thus far in

preliminary investigations. Next steps are to obtain IRB approval to study technical and operations feasibility

- b. Joline Dalton, CGC, SMA Clinic, discussed the phenotypes seen in SMA. Discussed Spinraza treatment method and specific studies of clinical trials demonstrating improvement with this treatment.
 - c. Dr. Peter Karachunski, MD, SMA Clinic, discussed treatment tolerance and approach. He discussed current case volume and clinical interest in treatment. Stated he felt this treatment “is effective and can change the course of the life of someone with SMA. Protects and renews nerves, doesn’t just stabilize disease, but it helps affect individuals progress with motor skills.”
 - d. Carissa Keister, Parent of a Child with SMA, shared her story of her daughter, Evie, who passed away from SMA I three years ago. Discussed their diagnostic journey and experience with SMA. Shared the experience of other families who have been benefited by Spinraza.
 - e. Discussions and questions by the committee.
 - f. The Committee decided to see what the ACHDNC does at its next two meetings and will discuss again at the next Committee meeting.
- 7) Advisor Updates
- a. Dietrich Matern - Mayo has been screening for Pompe, Krabbe, and MPS I for Kentucky and have added two more positive cases, 2 Pompe, 1 Krabbe, 1 MPS I. Mayo has been screening some Minnesotans for the full RUSP using their supplemental newborn screen.
 - b. Steve Nelson - Entering third year of STORM (Sickle Treatment and Outcomes Research in the Midwest). Discussed purpose of STORM, current activities, and focus of this grant.
- 8) Closure
- a. Jan Larson – we stand adjourned
 - b. As always, please email Maggie/Sondra or entire committee if you have any updates or things you’d like to share.