

Newborn Screening Advisory Committee Meeting

Meeting Minutes of Tuesday, October 11, 2016

1:00-4:00

Wilder Center

Auditorium A - B

451 Lexington Parkway North

Saint Paul, MN 55104

Committee Members Present:

Bob Jacobson	Kristi Borowski	Ed Jenkins	Emilee Scheid
Dieter Matern	Vikas Bhambhani	Terri Laguna	Kevin Sheridan
Mark Bergeron	Steve Johnson	Jan Larson	Kathy Stagni
Julie Thompson Larson	Susan Berry	Martha Overby	Renee Temme
Amy Karger	Roshini Araham	Annamarie	Jennifer Ward
Kirsten Dummer	Jenny Jacobson	Saarinen	Kristin Loncorich

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

Michael Nwaneri (N)	Courtney Jarboe (N)	Kiki Sarafoglou (N)
Steve Nelson (N)	Sheyhan Gelle (N)	

Guest Speakers:

Peter Karachunski	Terri Laguna	Ashley Asher
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- 1) Welcome and Introduction
 - a. Jan Larson (Chair) welcomed committee and introduced Mark McCann, program manager.
 - b. Mark McCann: Welcomed on behalf of MDH & PHL
 - a. In addition to spinal muscular atrophy (SMA), future disorders on watch list: guanidinoacetate N-methyltransferase (GAMT) and Duchenne muscular dystrophy (DMD)
 - b. Thanked Dr. Matern for email regarding performance metrics. Encouraged advisors to contact Maggie and Sondra with any additional thoughts/comments they would like considered.
- 2) Business of Committee
 - a. Review of minutes from April of 2016. It was noted that year needed to be adjusted to 2016 and Julie Thompson Larson was in attendance. Jan called for motion to approve with those amendments. Mark Bergeron motioned. Martha Overby seconded. No discussion. Vote: Ayes, motion carries.
- 3) MN Committee Coordinators Updates
 - a. Maggie Dreon & Sondra Rosendahl:
 - i. Newsletter – let them know if there are any questions
 - ii. Lysosomal disease and leukodystrophy implementation workgroup to meet November 9th, over 20 specialists involved.
 - iii. Format different today - emerging condition: spinal muscular atrophy (SMA), and a 'look back' at a previously added condition: Cystic Fibrosis (CF).
 - iv. Dr. Michael Nwaneri resigned from committee due to moving out of state.
- 4) Emerging Condition – Spinal Muscular Atrophy (SMA)

- a. Guest speaker - Peter Karachunski, Pediatric Neurologist at the University of MN
 - i. Provided a comprehensive review of Spinal Muscular Atrophy (SMA): clinical features, diagnosis, genetic variations, treatment, clinical trials. Discussed SMA as it relates to newborn screening – not currently on the screening panel for any state. Discussed a study in which parents of children with SMA believed that it would be a valuable addition to newborn screening.
 - ii. Questions and comments by the committee.
- 5) 'Look Back' on Current Disorder – Cystic Fibrosis
- a. Speaker – Dr. Terri Laguna, advisor and Pulmonologist at the University of MN
 - i. Case example of CF before screening, the CF workflow, advantages, challenges, current drug developments for CF and why newborn screening is important for these trials, no ultimate cure yet- still have work to do. Survival improved for CF patients due to newborn screening, but needs to improve more.
 - ii. Questions and comments from the committee.
- 6) Family Story
- a. Guest speaker - Ashley Asher
 - i. Son Carson (4 yrs old). Identified prenatally to both be carriers of CF and possible affected child. May 30th, 2012 Carson was born. Found out at 9 days old that Carson has CF. 1st year was the hardest, but currently he is doing well. CF has come a long way, but they are waiting for the day for when CF stands for Cure Found. Appreciate the care they received at the University of Minnesota and being identified through newborn screening.
- 7) Advisors Updates
- a. Susan Berry – kudos to Kristi Bentler who authored paper on MCAD
 - b. Dieter - Kentucky newborn screening update on 35,000 babies screened (1 true positive MPS-1, 1 false positive MPS-1 that did not have 2nd tier testing, and 1 late-onset Pompe)
 - c. Annamarie – invitation to attend the Newborn Foundation meeting @ U of MN (Kauffman Union) on Friday, Oct. 14, and the Gala on Saturday, Oct 15th. Sue Berry will be receiving an award.
 - d. Terri Laguna - attended (along with Mark & Sondra) NewSTEPS 360 meeting on CF timeliness of newborn screening; MN is a model state.
 - e. Steve Johnson - MN PKU foundation has much activity, including a golf tournament to help subsidize children and families going to PKU camps and a picnic. They've also recently updated their website: www.MNPKU.org
 - f. Kathy Stagni - Organic Acidemia Association is working on creating a patient directed registry through NORD & FDA, creating new treatments, traveling to Virginia.
 - g. Jan Larson - 20th anniversary of the phone call they received regarding their son's newborn screen, now a celebration (son is now 23 yrs old). Thank you to all for everything you do to help children.
- 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.