

Newborn Screening Advisory Committee Meeting

Meeting Minutes of Tuesday, October 10, 2017

Committee Members Present:

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|---------------|----------------|--------------------|---------------|
| Mark Bergeron | Jenny Jacobson | Vikas Bhambhani | Sheyhan Gelle |
| Terri Laguna | Dieter Matern | Amy Karger | Steve Johnson |
| Steve Nelson | Jan Larson | Annamarie Saarinen | Emilee Scheid |
| Kathy Stagni | Renee Temme | Courtney Jarboe | |

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

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| Roshini Abraham (N) | Susan Berry (N) | Kristi Borowski (N) | Kirsten Dummer (N) |
| Bob Jacobson (N) | Kristin Loncorich (N) | Kiki Sarafoglou (N) | Kevin Sheridan (N) |
| Jennifer Ward (N) | | | |

- 1) Welcome and Introductions:
 - a. Mark McCann
 - i. No formal call for a vote, but advisors have the authority to call for a vote.
 - b. Jan Larson
 - i. Announcement: Ed Jenkins and Martha Overby have both resigned from this committee.
- 2) Review and Approval of Minutes for April 2017
 - a. Review of minutes from April of 2017. It was noted that Steve Nelson was present. Steve Johnson motioned to approve the minutes with correction. Courtney Jarboe and Steve Nelson seconded. No discussion. Vote: Ayes, motion carries.
- 3) New Disorder Updates (MPS I and Pompe):
 - a. Amy Hietala
 - i. Provided an update to the Committee regarding the addition of three new disorders.
 - ii. Began screening on August 1, 2017.
 - iii. As of October 6, have screened 13,100 infants: 6 MPS1 and 10 Pompe presumptive positives.
 - iv. Upon Mayo's 2nd tier testing only 1 was found to have infantile onset Pompe. No cases of MPS I to date have been identified.
- 4) X-ALD – Early Experiences and Lessons Learned:
 - a. Amy Gaviglio and Holly Winslow
 - i. Reviewed X-ALD timeline from addition to panel to running specimens.
 - ii. As of Sept 30, have screened 45,095. We have identified 29 borderlines, 2 repeat borderlines, and 7 positives (4M, 3F).
 - iii. The 9 cases have gone to diagnostic testing, which has found 24 additional affected family members to date. We expected some additional family members but were surprised by the magnitude of extended family members being identified.
- 5) Spinal Muscular Atrophy
 - a. Maggie Dreon and Sondra Rosendahl
 - i. Review and update on the national activity at the ACHDNC and other states' activities. The New York Pilot on SMA data and information was shared.

Discussed that the ACHDNC was reviewing specific information about screening considerations, such as: carrier status detection and reporting, SMN2 copy number detection and reporting. Discussed the FDA approval of Nusinersen/Spinraza. Reviewed the next steps for the ACHDNC.

- ii. Review and update on SMA in Minnesota and Minnesota's readiness for SMA newborn screening.
 - iii. Public Comment:
 1. Carrissa Kiester – Presented the Committee with a petition signed by 3150 individuals supporting the addition of SMA to newborn screening in MN. Urged the Committee to take action now.
 2. Kara Forcier – Shared the story of her son Zachary. Urged Minnesota's committee to lead in SMA screening.
 3. Laura – shared the story of her son Andrew. Urged the Committee to vote today.
 4. Jayme – Shared the story of her daughter, Madison. Urged the Committee to consider adding SMA today.
 - iv. Discussion:
 1. Maggie Dreon and Sondra Rosendahl
 - a. Through 'Think, Pair, Share' facilitation, the Committee reviewed SMA using the Guidelines developed by the Committee previously. Discussion surrounded clinical considerations, details of the screening test/method, and the diagnosis, follow-up, treatment, and management for SMA.
 - v. Next Steps/Actions:
 1. Jan Larson
 - a. Shared several options for next steps, including tabling until another meeting. Asked the committee how they would like to proceed.
 - b. Courtney Jarboe motioned for the Advisory Committee to make a recommendation to the Commissioner to add SMA to the Minnesota newborn screening panel without waiting for the ACHDNC to add it to the RUSP.
 - c. Emilee Scheid seconded that motion.
 - d. Discussion was had about the role of the committee vs the role of the Commissioner vs the role of the ACHDNC/RUSP.
 - e. Vote: 13 Ayes, 1 Nay, motion carries.
- 6) Current Disorder Highlight – Severe Combined Immunodeficiency (SCID):
- a. Dr. Tamara Pozos (guest speaker)
 - i. Pediatric immunologist for Children's Hospitals and Clinics of MN.
 - ii. Shared with the committee the impact of newborn screening for SCID and the current state of SCID screening.
- 7) 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.