



## Meeting Minutes: Advisory Committee for Heritable and Congenital Disorders

April 17, 2018

Minutes Prepared By: Jessica Cavazos, Newborn Screening Program Health Educator

Location: Wilder Center

### Advisors Present

- Roshini Abraham
- Steve Johnson
- Emilee Scheid
- Vikas Bhambhani
- Amy Karger
- Kevin Sheridan
- Kristi Borowski
- Jan Larson
- Kathy Stagni
- Sheyhan Gelle
- Dieter Matern
- Jennifer Ward
- Courtney Jarboe
- Steve Nelson

### Advisors Absent

- Mark Bergeron
- Bob Jacobson
- Annamarie Saarinen
- Sue Berry
- Jenny Jacobson
- Kiki Sarafoglou
- Kirsten Dummer
- Kristin Loncorich
- Renee Temme

### Summary of Decisions Made

- Decision: The October 10, 2017 meeting minutes approved.
- Decision: Jan Larson was voted in as Chair and Emilee Scheid was voted in as Vice Chair.

### Summary of Action Items

- Convene a work group to update the advisory committee's current bylaws.
- Convene a work group of advisors for the Disorder Nomination and Consideration Model Development to provide input on when/how the advisory committee will be engaged when new disorders are nominated to Minnesota's newborn screening panel.

### Agenda

- Welcome and Introductions
  - Reviewed MDH's disorder watch list.

- Family Story – Phenylketonuria (PKU)
  - Becca Williams – Discussed the story of her twins that were identified with PKU through newborn screening. She shared about the news, how it impacted her family, and what living with PKU looks like.
- Committee Business
  - Review and Approval of October 2017 Meeting Minutes
    - Motion to approve minutes by Steve Johnson, Emilee Scheid second. Vote: all ayes, motion passes
  - Bylaws Changes –
    - Call for work group to review and update the committee bylaws.
    - Steve Johnson, Emilee Scheid, Dieter Matern, and Courtney Jarboe offered to help.
  - Vote on Chair/Vice Chair
    - Nominations: Jan Larson (Chair), Emilee Scheid (Vice Chair).
    - Steve Johnson moves to approve slate of nominations. Call for additional nominations. Motion to close nominations by Dieter Matern, Jennifer Ward second.
    - Jan Larson voted in as Chair and Emilee Scheid voted in as Vice Chair.
    - Thank you to Kathy Stagni for service as Vice Chair.
  - Resignations and Expiring Terms
    - Terri Lagnua and Kirsten Dummer have resigned and are moving out of state. Please email coordinators if you have suggestions for new advisors.
    - Half of the advisors’ terms will be expiring at the end of the year and advisors will be notified by the committee coordinators.
- Follow-up for X-ALD, MPS I, and Pompe Disease – A NewSTEPS and MDH Partnership
  - Elise Holmes, MPP, Planner (MDH)
    - See slides for details. Elise discussed the details of the grant they received, their plan for implementation, and their progress to date.
    - Discussion:
      - Steve Nelson: Looking at definitions of long-term outcomes and maximizing health outcomes, what age do you stop being involved with these patients?
      - Elise: We reach out at 3 months, 1 year, and have a plan or assessment by age 4. California goes up to age 5 (longest term), sometimes described as 18-23, but in terms of NBS follow-up we haven’t gotten there yet.
      - Amy Dahle (MDH staff member): how are you recruiting families?
      - Elise: family support groups for these conditions, baby’s first test and genetic counselors (via Baby’s First Test).
- Co-coordinators and Manager Updates
  - Maggie Dreon
    - SMA Update:
      - On October 10<sup>th</sup>, the committee voted to recommend SMA to the Commissioner.
      - On December 19<sup>th</sup>, the commissioner approved. MDH went live with screening on March 1<sup>st</sup>.

- MDH has screened 9,300 newborns for SMA and identified the first positive case on April 1<sup>st</sup>.
    - Mark McCann
      - Legislative Update:
        - Three bills introduced last session.
          - 1) G6PD – education bill; probably not moving forward,
          - 2) Rare Disease Bill – creation of advisory council on rare disease,
          - 3) CMV education
- Disorder Nomination and Consideration Model Development
  - Steve Johnson (parent, advisor)
    - See slides for details. Steve discussed the plan for how the Disorder Nomination and Consideration Model Development work group will be conducted and shared how advisors will be engaged in this activity.
    - Discussion with advisors pertained to what questions should be asked of the community members, and how best to inform the public about MDH’s process so that someone who has an interest in nominating a disorder to MN’s panel that reasonable expectations are laid out for them.
- Advisors Updates and Closure
  - Kathy Stagni:
    - Shared that the organic acidemia parent support group is having a conference in Bloomington this summer. Susan Berry is going to be speaking. We expect over 300 families to attend.
  - Roshini Abraham:
    - There is a meeting on the 4<sup>th</sup> of May for SCID NBS. Clinical is having a hard time triaging results since NBS values cannot be reported; Shared that they are working with lab to come up with solutions. Will be leaving in July. Shared her thanks to everyone at MDH for allowing her to participate in NBS, and that she has learned a lot and have enjoyed working with everyone.
  - Emilee Scheid:
    - Has been with the Newborn Hearing Screening Advisory Committee (been with it for 5 years now). She has been working with providers on guidelines and giving presentations at conferences regarding medical home updates for providers. It is the 10 year anniversary of EHDI.
  - Steve Nelson:
    - Shared that there have been many advances in sickle cell disease (SCD) treatment and curative therapies, but that the most important intervention that has affected outcomes is NBS. The average survival went from 12 to mid 40s (in the US). But the new crisis is the peak mortality is between 18 and 25, mortality for adults at this age has increased by 1% per year. All these adults have SCD, but have nowhere to go for care. Transition to adult care is an issue. Discussed a grant (details in the newsletter) they received and are engaging with UMN adult hematology group to create an adult SCD clinic. Shared his thanks to MDH for helping us with this. Also, the Sickle Cell Foundation of MN is now in

its second year. They are having their first fundraiser. There will be a walk on June 16<sup>th</sup> and a gala in September.

- Dieter Matern:
  - Stated his hope that MDH is happy with second tier testing for MPS I and Pompe. Shared that they are screening Krabbe for Kentucky. Have had 1 abnormal result from over 120,000 screened. Child has been transplanted. Earlier this year there was an article from a taskforce (Hunters Hope) to look at 2008 evidence review. They have found a way to screen and identify those affected, but the problem remains how to treat them. 18 patients who were treated early in life but 5 have died due to treatment complications; much higher than typical treatment rates.
- No other committee business; adjourn.

## **Next Meeting**

Date: October 9, 2018

Time: 1:00 – 4:00 PM

Location: Wilder Center