Newborn Screening Advisory Committee Meeting
Meeting Minutes of Tuesday, April 21, 2015
1:30-4:30
Wilder Center
Auditorium A and B
451 Lexington Parkway North
Saint Paul, MN 55104

Committee Members Present:
Mark Bergeron    Steven Johnson    Kristin Loncorich    Michael Pryor
Susan Berry      Amy Karger       Richard Lussky       Annamarie Saarinen
Vikas Bhambhani  Julie Thompson Larson   Dietrich Matern  Kevin Sheridan
Jenny Bock       Jan Larson       Michael Nwaneri    Kathy Stagni
Kirsten Dummer   Candace Lindow-Davies Amy Powers Jennifer Ward
Robert Jacobson

Committee Members Absent:
Marianne Keuhn    Steve Nelson (notified he’d be absent) Kiki Sarafoglou

Guest Speakers:
Chet Whitley, PhD, MD    Amy Hietala    Dave Orren, Chief Legal Counsel for MDH
Amy Gaviglio    Tony Steyermark

Agenda Items:
1. Welcome and Introduction
   a. Mark McCann, Director of the Newborn Screening Program, welcomed the Committee members and guests to the Newborn Screening Advisory Committee meeting.
   b. Jan Larson, current Chair of the Newborn Screening Advisory Committee, welcomed the Committee members and guests and opened the meeting.
2. Vote on Revised Bylaws
   a. Jan introduced the redline version of the revised bylaws for the committee.
      i. Vice Chair would succeed the Chair after the Chair’s two year term expires
         1. This was discussed amongst the committee, many agreeing that this is a needed bylaw and would ensure continuity of leadership for the committee. Room was left to address individual needs separately,
should a Vice Chair not be able to fulfill their role as Chair when their
turn came.

b. Jan asked for a motion to accept the bylaws with the proposed amendments
   i. Susan Berry motioned to accept the bylaws with the proposed amendments
   ii. Mark Bergeron seconded
   iii. Chair called for a vote, all in favor, none opposed, motion was adopted.

3. Newborn Screening Updates
   a. The committee members briefly introduced themselves and indicated if they were a
      new or returning advisor. Rich Lussky is leaving clinical practice and wants to go into
      public health.
   b. Dave Orren gave a brief summary of the lawsuit that the Newborn Screening Program
      was involved in which lead to program and legislative changes. He also expressed
      concern about the Newborn Screening Reauthorization Act of 2014, specifically section
      12 which was added in the Senate and is limiting and possibly further problematic
      because the use of “research” is very broad, and research on blood spots is considered
      to be research on human subjects. This could possibly put the state at risk of another
      lawsuit if developing new tests could be considered “research.”
   c. Committee discussion:
      i. Dieter Matern: mentioned that this currently only applies to federally funded
         research, but we need to point out to advocates and the legislature how
         dangerous this could be to newborn screening, so that this doesn’t eventually
         include MDH “research.” ‘What would have happened to newborns over the
         past 50 years if this had happened? We would only have PKU testing, and many
         newborns wouldn’t have been screened and treated for other conditions.’
      ii. Annamarie Saarinen: ‘what you are saying is that there is a limited window of
         time during which you can influence laws and that we should be weighing in
         with federal agencies on this subject matter.’
         1. Susan Berry: ‘OHRP has a standardized policy on comments on changes
            in common rule. This is a very short window of time. We basically have
            two options, monitor their public comments and request for changes
            and work through your senator and congress members to bring some
            pressure to bear in Washington. Be aware that changes to the common
            rule encompass not just the blood spot, but anything material that came
            from a human. Common rule will probably be broader than what is
            currently in section 12.
      iii. Dave Orren: ‘We are trying to advocate the use of blood spots to create and
         validate new tests. The risk to old babies (those whose blood spots would be
         used) is practically zero, the benefit to new babies is monumental.
      iv. Sue: ‘Advancing knowledge for new tests is at risk. Current lab processes are
         most likely safe (quality control, validation of current tests) for now.

4. MnScreen
   a. Amy Gaviglio gave a report on MNScreen, a new electronic reporting tool for pulse
      oximetry and hearing screening results that pulls directly from the pulse oximetry or
      hearing screening equipment and enters it into MNScreen. It also merges information
from the infant’s electronic medical file to create a complete newborn screening record. MNScreen reporting will add security, improve workflow, reduce errors, and provide accurate demographic information for each birth. The goal is to have full implementation across MN by December 31, 2015.

b. Committee discussion:
   i. Dieter Matern recommended informing the public about it.

5. Chair and Vice-Chair Voting
   a. Current nominations are:
      i. Kathy Stagni, Vice Chair
      ii. Jan Larson, Chair
   b. Floor was opened for further nominations
      i. No further nominations were suggested
      ii. Susan Berry motioned to close nominations
      iii. Steve Johnson seconded
      iv. All in favor, nomination closed.
   c. Committee members completed their voting ballots, which were collected. The ballot votes unanimously elected Kathy Stagni as Vice Chair and Jan Larson as Chair.

6. 10-Minute Break

7. Lysosomal Storage Disorders
   a. Clinical Perspective
      i. Chet Whitely gave a presentation to the committee on LSDs, including case studies on diagnosis and treatment (if available) of LSDs.
      ii. Committee discussion:
         1. Sue Berry- What do we do for late onset or ones with no treatment? Emerging treatments? Chet-Optimistic that it will be cured/early treated in the future. There are always short coming with treatment. There are 6 disorders where there are drugs out there for treatment. Gene therapy will be used in the future. There is so much progress, but anything option fails if there is already damage. He thinks Genome sequencing will be the possible in the future.
         2. Kevin Sheridan- Late onset versus early onset in newborns. Is there a way to separate them? Chet-No. Use mutation analysis to determine which kind it is after seeing the late enzyme. They do DNA sequencing and the severity of the mutation. There is no full catalog of mutations for Fabry.
         3. Dieter Matern-There are many undiscovered mutations and related to the disease. There are a lot of times that you don’t know if it’s Hurler or what they are. Only time can tell.
      iii. Mark McCann asked Chet what his opinion was for what we should screen for based on the 6 LSDs on the current bill:
         1. Hurler: add to Newborn Screening
         2. Gaucher: add to Newborn Screening
         3. Niemann-Pick: no (no accepted treatment) – sometimes there are other reasons to add the disorders
4. Fabry: add to Newborn Screening  
5. Pompe: add to Newborn Screening  
6. Krabbe: Can’t say – wants to hear what other people think about it  

b. Newborn Screening Perspective  
   i. Amy Hietala from the Newborn Screening Lab gave a presentation on what the laboratory would need to do in order to begin testing for the 6 proposed LSDs and possible hurdles and costs associated with it. This included a comparison of two testing platforms and a review of the estimated annual incidence that MN could expect based on Missouri’s data.  
      1. Implementation considerations:  
         a. Cost/time involved in obtaining new equipment and making lab upgrades  
         b. Hiring staff for testing  
         c. Dealing with false positives and cases of pseudodeficiency  
         d. Challenges performing long-term follow-up  
         e. Low incidence of the disorder  
         f. Need to develop reporting and follow-up algorithms  
         g. Need to partner with specialists for LSDs in MN  
         h. Notify and educate primary care providers and parents  
         i. Partner with clinical diagnostic laboratories  
   ii. Amy Gaviglio presented what would need to be done to follow-up on infants with abnormal screens for LSDs. She presented information from New York, Missouri, and Illinois and their experience with screening for LSDs thus far, including lessons learned and advice about what things we should think about.  
      1. Things to consider for MN:  
         a. Estimated cost for testing and follow-up per infant: $6-8  
         b. Development of MDH follow-up protocols  
         c. Development of clinical diagnostic protocols  
         d. Availability of treatment (travel, treatment, cost)  
         e. Addressing family needs and late-onset/ambiguous results  
   iii. Tony Steyermark presented the committee with a list of things that will need to consider in order to provide the best support for families with LSDs found via newborn screening. This would include ensuring access to services, collection of outcomes data, and how to handle late-onset presentations.  
   iv. Committee discussion: $6-8 is a very rough estimate of what it would cost to test for 2 or 5 LSDs depending on the method chosen. This doesn’t include molecular testing or how much time would be needed for follow-up. Discussion included questions about role of primary pediatricians, cost impact for the family, impact of false positives on families, are the current tests sufficiently precise? LSDs are very expensive to treat, is it an appropriate use of healthcare costs? Do LSDs meet the guidelines that MDH has put together for consideration of addition to the NBS panel?  

c. Parents Perspective
i. Many parents and family members gave personal accounts of their struggle over the LSD diagnosis their child received and what their daily lives are like. Some asked the committee to approve adding LSDs to the newborn screening panel, some wanted to be able to say they wanted us to add it but didn’t feel like it was time, while one gentleman said he thought he knew what he was going to say but didn’t anymore.

d. Discussion

i. The committee discussed the topic, including the possibility of adding what is on the RUSP but agreed that there needs to be more discussion at the October 13, 2015 meeting.

1. Chair (Jan) called for a motion to add LSD discussion to the October 13, 2015 agenda.
   a. Dieter Matern motioned to add LSD discussion to the October 13, 2015 agenda.
   b. Sue Berry seconded

2. Motion adopted, LSD discussion will be added to the October 13, 2015 agenda.

8. Meeting adjourned