Memo

Minnesota Department of Health

Date: December 28, 2015

To: Commissioner Ed Ehlinger

Through: Paul Allwood, Assistant Commissioner, Health Protection Bureau
Joanne Bartkus, Public Health Laboratory Division, Director

From: Mark McCann, Newborn Screening Section Manager, Public Health Laboratory Division,
Maggie Dreon, Newborn Screening Genetic Counselor, Advisory Committee Coordinator
Sondra Rosendahl, Newborn Screening Genetic Counselor, Advisory Committee Coordinator

Subject: **Request for Response to the Recommendations made by the Advisory Committee on Heritable and Congenital Disorders**

The MDH Newborn Screening Program and its Advisory Committee on Heritable and Congenital Disorders would like you to consider the expansion of the Minnesota newborn screening panel. The mechanism for revising the list is described in Minnesota Statute, 144.125 "Tests of infants for heritable and congenital disorders," as follows:

"The list of tests to be performed may be revised if the changes are recommended by the advisory committee established under section 144.1255, if approved by the commissioner, and published in the State Register. The revision is exempt from the rulemaking requirements in chapter 14, and sections 14.385 and 14.386 do not apply."

Expansion is being sought because on the national level, the Advisory Committee on Heritable Disorders in Newborn and Children (ACHDNC), which advises the Secretary of the US Department of Health and Human Services (DHHS) has adopted Pompe disease as a national standard for newborn screening practice as part of the recommended uniform screening panel (RUSP). Two additional conditions, X-ALD and MPS I, have been recommended to be added to the RUSP by the ACHDNC and are currently waiting for a response from the DHHS Secretary. The DHHS Secretary has no deadline for responding to MPS I. However, the DHHS Secretary has to respond within 90 days for X-ALD as per the new Reauthorization of the Newborn Screening Saves Lives Act. Of the 32 disorders and 26 secondary disorders recommended for mandatory screening, MDH has met the standards for 57 of the 58 current conditions. We currently only lack Pompe disease, which was added to the RUSP in 2015. If X-ALD and MPS I are added to the RUSP by the DHHS Secretary, then we will lack those as well.

In addition to the national movements in lysosomal diseases, a group of lysosomal diseases were included in a bill introduced to the Minnesota legislature by Senator Jeremy Miller. This bill is called "The Super Gav Act" and includes the following diseases: Fabry, Gaucher, Krabbe, Alpha-L-iduronidase deficiency (MPS-I), Niemann-Pick A/B, and Pompe.

On April 21st, our Advisory Committee met to start discussions surrounding the diseases proposed in The Super Gav Act. Following this meeting, a work group was formed that was charged to provide expert guidance and recommendations to our Advisory Committee on whether or not these lysosomal diseases and X-ALD should be recommended for inclusion on Minnesota's newborn screening panel.

Our Advisory Committee met on October 13th to hear the outcomes of the work group and decide on whether or not these conditions should be recommended for inclusion on Minnesota's newborn screening panel. The Advisory Committee had three formal votes at this meeting, all of which passed. The first of which is specifically related to the Committee's roles and responsibilities as discussed in statute as it addresses the addition of new conditions to the Minnesota panel:

1) For the Advisory Committee to have a single vote to adopt the recommendations of the work group for inclusion on the Minnesota newborn screening panel. This would include Pompe immediately. MPS I and X-ALD would be added upon the addition to the RUSP, which is currently waiting for the DHHS Secretary's signature. **Motion carries – 15 for, 3 opposed**

In summary, the Advisory Committee would like to add Pompe disease to the Minnesota newborn screening panel. Pending the (likely) addition of MPS I and X-ALD to the RUSP by the DHHS Secretary, the Advisory Committee also recommends the addition of MPS I and X-ALD.

Given the extensive evidence reviews done by the ACHDNC, when conditions are nominated for addition to the RUSP, the Advisory Committee voted and passed the following recommendation:

2) Committee approves conditions that are added to the RUSP would be presumed to be added to the Minnesota panel upon a formal vote by the Advisory Committee and without the need to comission a work groups. Simply put, a condition that has been vetted though the formal ACHDNC process and is added to the RUSP, would be presumed to be recommended for addition to the Minnesota newborn screening panel and, upon a formal vote by the committee, to our MDH Commissioner, thereby eliminating the need for a work group to decide about conditions already vetted through the formal ACHDNC process. **Motion carries – unanimous**

Advisory Committee members understood that implementation of the two recommendations listed above would require MDH to secure resources (including staff expertise, training, and equipment), establish follow-up and connections to services for the affected families, and integrate the laboratory testing component into Minnesota's model for newborn screening public health practice. In addition, committee members understood the legal and political challenges that have been at play for the last few years. MDH will need to explore a request for a fee increase to implement and sustain the program in relation to this test addition.

While outside the directed scope of the Advisory Committee's role, the Advisory Committee felt specifically addressing infrastructure needs in Minnesota would be critical to the success of screening for lysosomal diseases and voted on and passed the following recommendation:

3) Committee recommends that the Department of Health ensure: 1) comprehensive long term follow-up of children identified as affected by newborn screening to provide a continuing means for determining the optimization of outcomes for diagnosed citizens; 2) increased awareness of the utility of long term follow-up; 3) and improved education to parents and providers about outcomes of newborn screening conditions and their long-term management. **Motion carries – unanimous**

The enclosed documentation highlights the benefits and risks of screening for Pompe, MPS I, and X-ALD and includes the national recommendations for state newborn screening programs. With early detection and medical intervention, current treatment outcomes pose the possibility to be curative. When detection and treatment are obtained prior to onset, long term survival and treatment options increase. Without this simple test, most children with Pompe, MPS I, and X-ALD are identified after significant morbidity and the inability to offer effective treatment.

The enclosed documentation also highlights the concerns of screening for Fabry, Gaucher, Krabbe, and Niemann-Pick A/B diseases, which are not recommended for newborn screening at this time Concerns surrounding these four conditions are included here due to the pending legislative bill, The Super Gav Act. Although the Advisory Committee is not recommending these conditions at this time, it was clear that the committee expects to stay abreast of these conditions as new developments (e.g., treatment advances) came to light so that they had an opportunity for potential reconsideration.

Request for Response

At present, we ask you to review the Advisory Committee's recommendations as outlined above. Please provide us with your response at your earliest convenience. For your awareness, the next Advisory Committee meeting will be held on April 26th, 2016. We remain available for questions and will await your response.

Thank you for your consideration.

Cc: Joanne Bartkus
Myra Kunas
Mark McCann

Encl: Outline of documents
Materials provided to Advisors regarding lysosomal diseases and X-ALD
Summary table outlining ACHDNC time line and process for each presented condition
Long term follow-up considerations
Fabry Disease: ACHDNC denial letter, brief condition outline
Gaucher Disease: Brief condition outline
Krabbe Disease: ACHDNC denial letter, formal evidence review documents (detailed)
MPS I - DHHS acceptance letter, letter by ACHDNC recommending addition to Secretary, formal evidence review (detailed)
Niemann-Pick Disease: ACHDNC denial letter, brief condition outline
Pompe Disease: DHHS acceptance letter, letter by ACHDNC recommending addition to Secretary, formal evidence review (detailed)
X-ALD: Formal evidence review slides as presented to ACHDNC in the absence of formal document as pending currently, slides presented to ACHDNC as formal recommendation by evidence review committee
Submission for the *State Register opt 1 (MPS I, Pompe, & X-ALD), opt 2 (MPS I & Pompe), opt 3 (X-ALD & Pompe), opt 4 (Pompe)*