January 10, 2024

Dear Commissioner Cunningham,

On August 13, 2021, Dr. Peter Kang, a pediatric neurologist at the University of Minnesota, submitted a nomination to add Duchenne muscular dystrophy (DMD) to Minnesota's newborn screening panel. On May 31, 2022, the nomination was presented to the Advisory Committee on Heritable and Congenital Disorders (also known as the Newborn Screening Advisory Committee) where advisors voted to create a workgroup to assess DMD's readiness as a condition for newborn screening here in Minnesota. This scientific workgroup met four times to discuss the literature and assess readiness specific to Minnesota. Their findings are documented in the Condition Readiness Criteria document enclosed.

On October 23, 2023, the workgroup's findings were presented back to the advisory committee. Members from the Public Health Laboratory also presented on current efforts and feasibility of screening for DMD.

A few key elements of the discussion are highlighted below:

- Although no new equipment is needed, for redundancy in case of equipment failure, another Genetic Screening Processor (GSP) may be needed. No additional staff will be required within the newborn screening lab or follow up teams.
- First-tier screening using CK-MM is available from an FDA approved kit through Revvity, however the specificity of CK-MM alone is insufficient at minimizing false positive results. Second-tier molecular analysis of the *DMD* gene is available to reduce the false positive rate.
- Screening estimates based on the New York pilot outcomes (see included literature) combined with the average Minnesota birth rate show approximately 72 infants per year would have elevated CK-MM, and of those, nine would have a finding on molecular analysis requiring follow up.
- Current treatments include exon skipping therapy (30% of boys with DMD will be eligible), corticosteroids (standard of care – recommended to start after age 2), gene therapy (Elevidys - ~90% of boys will be eligible between 4-5 years of age). Treatments have been shown to prolong ambulatory age and modify phenotype in boys with DMD to be milder.
- Currently there is a known delay to diagnosis where boys typically present to specialty care at 5 years of age on average. Boys from rural areas or of lower socioeconomic status present even later after the eligible treatment window of currently available therapies.
- Concern for treatment coverage was brought up by advisors since the main available treatment, Elevidys, can't currently be administered until the age of 4 at the earliest. Exon skipping therapy and gene therapy are expensive treatments costing between $1.2-$3.4 million.

Following discussion, the Committee elected to proceed with a vote on the following:

To recommend the addition of Duchenne muscular dystrophy (DMD) to the Minnesota newborn screening panel as measurement of CK-MM and if elevated is followed by molecular testing to identify pathogenic or likely pathogenic variants in the dystrophin gene.

No = 0
Yes = 14
Abstain = 1

With a majority vote, the recommendation passes. Thank you for considering this recommendation.

Sincerely,

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Rae Blaylark, Chairperson