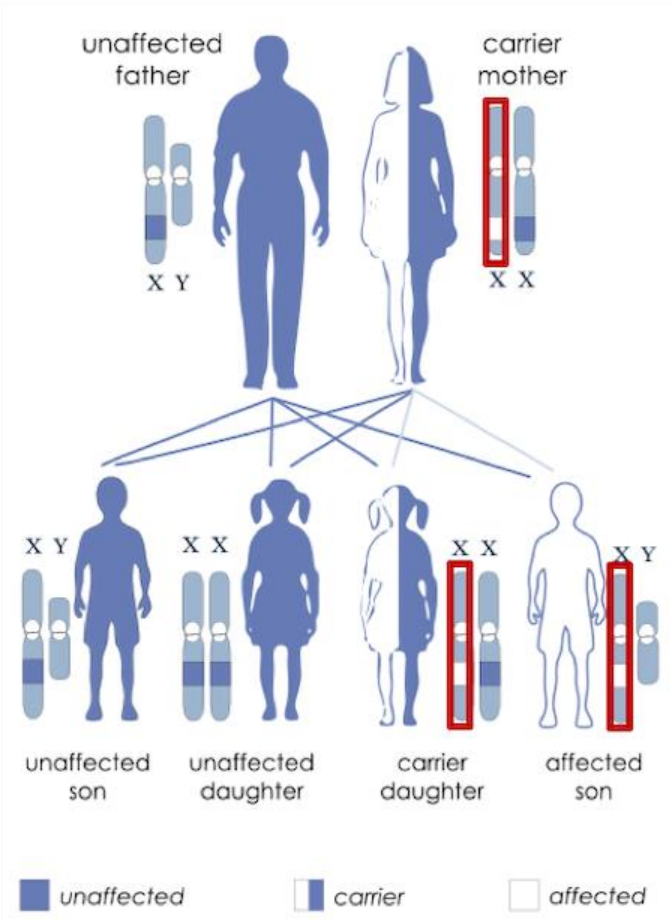

# Minnesota's Experience of Screening for Duchenne Muscular Dystrophy

# Duchenne Muscular Dystrophy – Background

- Prevalence – 1 in 5,000 live male births
- Onset – early childhood, usually 2-3 years of age; almost exclusively affects boys (X-linked recessive)
  - About 1/3 are de novo
- Symptoms
  - First sign is muscle weakness
  - Enlarged calves, waddling gait, and scoliosis
  - Heart and respiratory muscles are affected later
  - Survival into early 30s
- Muscle breakdown leads to elevated creatine kinase (CK) levels



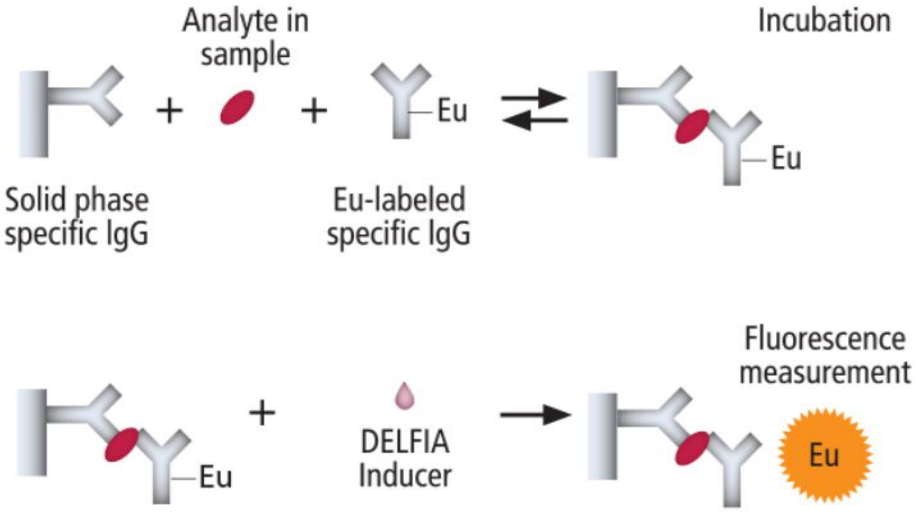
# Minnesota Timeline

- August 2021- DMD Nominated
- May 2022- Presented at the Advisory Committee and approved to move on to evidence review
- October 2023- Evidence Review presented to Advisory Committee and Vote
- January 2024- Commissioner Packet Review and Approval
- February 24, 2025- MN started Screening for DMD

# Laboratory

# DMD assay

- Started screening on 2/24/25
- GSP Neonatal Creatine Kinase –MM kit
  - FDA approved kit by Revvity measuring CK-MM levels.
  - Kit is a solid phase, two-site fluoroimmunometric assay based on the direct sandwich technique.
  - Analyte is bound to monoclonal CK-MM specific antibodies.
  - Fluorescence signal is proportional to the analyte concentration in the sample being tested.



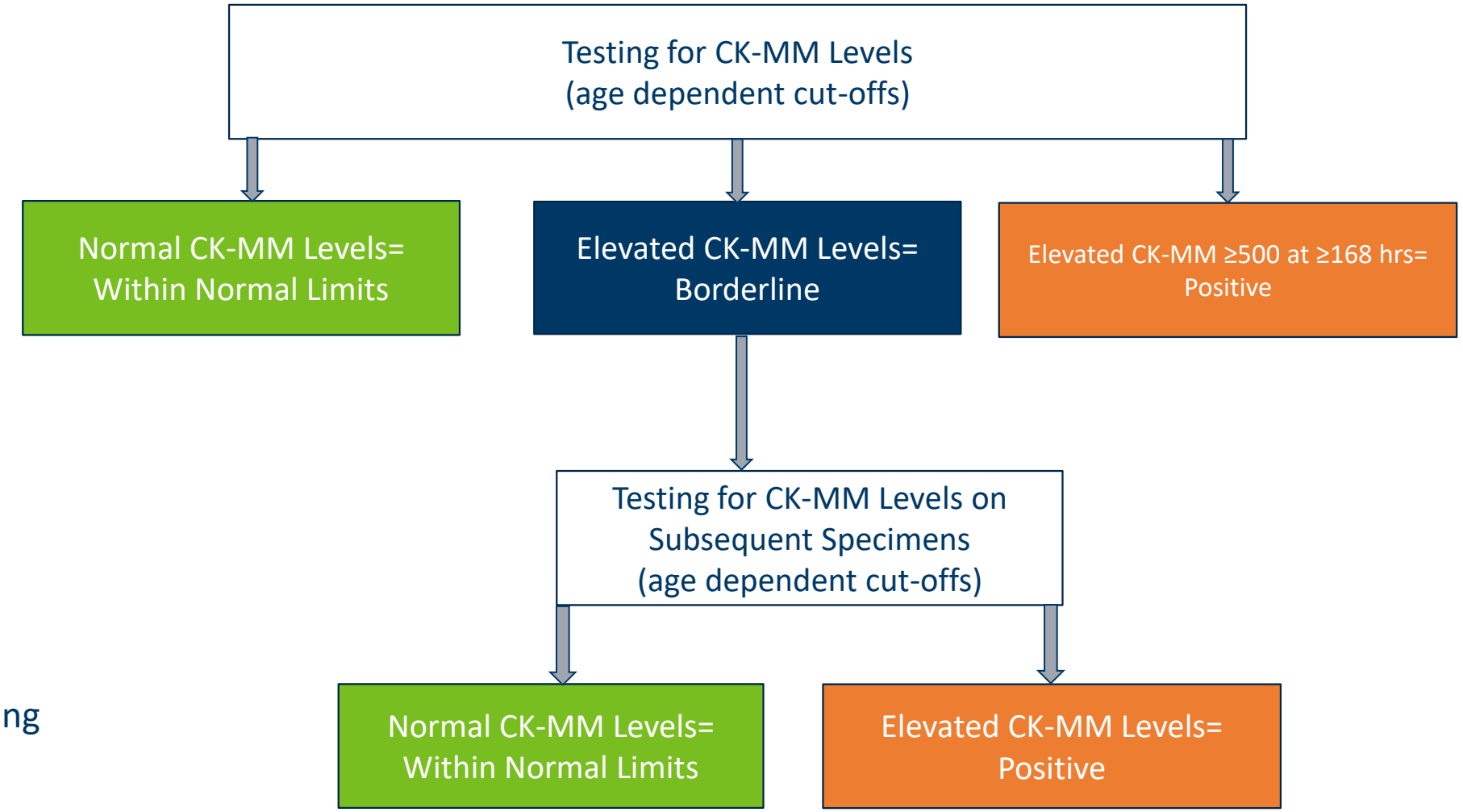
# Cut-Offs

## Established CK-MM cut-offs for DMD assay

| Age at Time of Collection (hours) | Borderline (ng/mL) | Positive (ng/mL) |
|---|---|---|
| 0 – 47 | ≥ 1700.0 | NA |
| 48 -71 | ≥ 1500.0 | NA |
| 72 - 167 | ≥ 500.0 | NA |
| ≥ 168 | NA | ≥ 500.0 |

Range 29.2 – 8000 ng/mL

# Algorithm




*We are not currently doing molecular testing

# Abnormal Result Interpretations

| | |
|---|---|
| Borderline | DUCHENNE MUSCULAR DYSTROPHY RESULT INTERPRETATION: This newborn screen shows elevated creatine kinase-muscular isoform (CK-MM). This finding is likely due to muscle injury. Send a repeat newborn screening specimen at 14 and 30 days of age. |

| | |
|---|---|
| Borderline | DUCHENNE MUSCULAR DYSTROPHY RESULT INTERPRETATION: This newborn screen shows elevated creatine kinase-muscular isoform (CK-MM). This finding is likely due to muscle injury. Collect a total CK after child is two weeks of age and fax the results to MDH at 651-215-6285. Consult with a neuromuscular specialist if clinical testing is abnormal. |

| | |
|---|---|
| 2nd Borderline | DUCHENNE MUSCULAR DYSTROPHY RESULT INTERPRETATION: Multiple newborn screening specimens, including the current specimen, have elevated creatine kinase-muscular isoform (CK-MM). This finding may indicate muscle injury or disease, such as Duchenne muscular dystrophy. Further diagnostic testing is recommended. Contact a neuromuscular specialist within one week. |

| | |
|---|---|
| Positive | DUCHENNE MUSCULAR DYSTROPHY RESULT INTERPRETATION: This specimen has elevated creatine kinase-muscular isoform (CK-MM). This finding may indicate muscle injury or disease, such as Duchenne muscular dystrophy. Further diagnostic testing is recommended. Contact a neuromuscular specialist within one week. |

# Follow-up

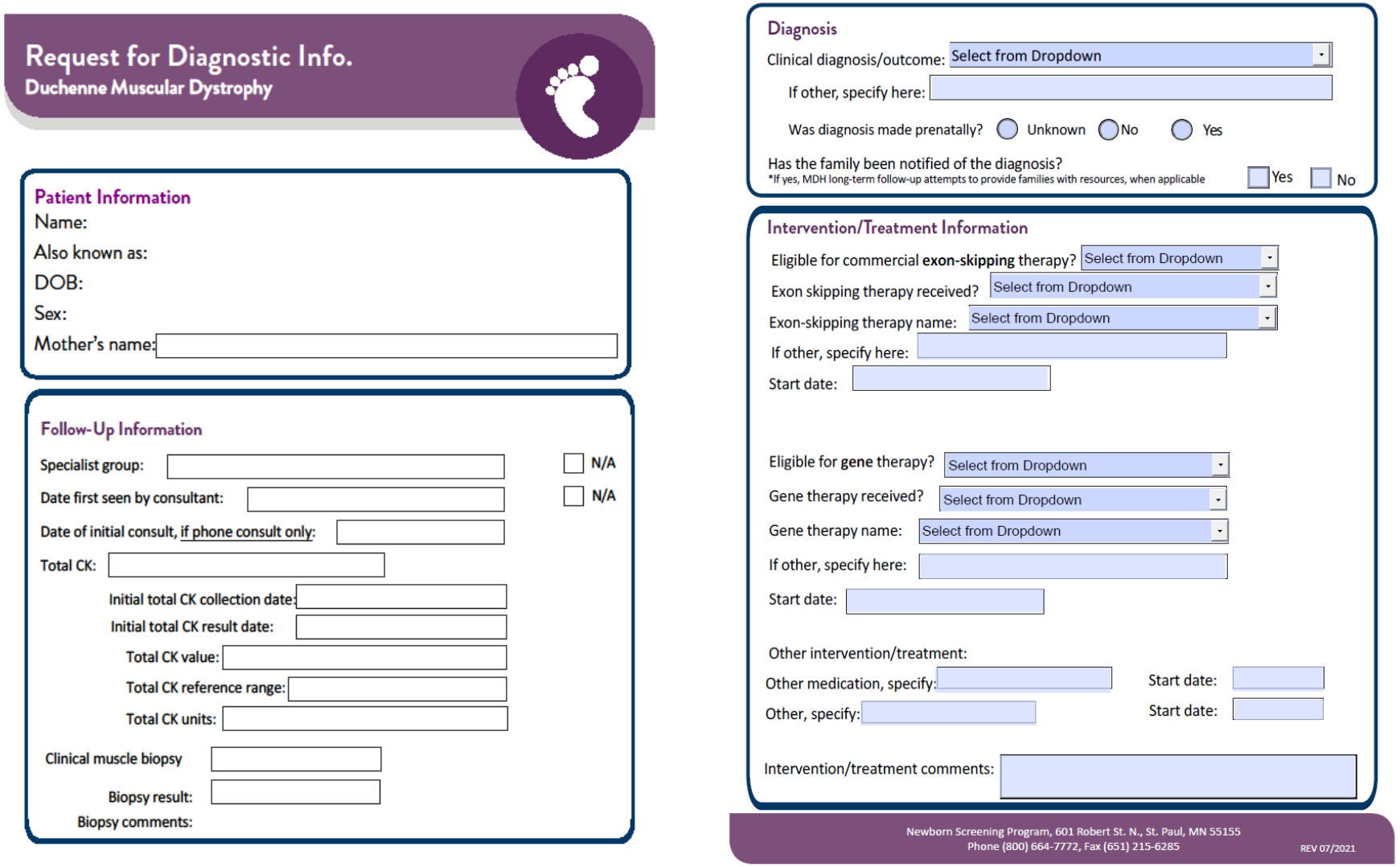
# Notification

- Abnormal result notifications made to primary care clinic/provider, midwife, or NICU if baby is admitted
- Provide “just-in-time” education
- Recommend ~~repeat newborn screen~~ **total CK** after two weeks of age
- Fax screening report, informational fact sheets for both provider and the family, and contact list for neuromuscular specialists

# Follow-up

- Persistently elevated CK on repeat newborn screen OR abnormal total CK labs prompts recommendation to consult with neuromuscular specialists within 1 week
- Referral sent to specialists through Natus that gives them access to the screening report and a brief note from us
- Diagnostic form completion is requested

# Diagnostic Form – sample data (not all we collect)



## Request for Diagnostic Info.
**Duchenne Muscular Dystrophy**

### Patient Information
Name:
Also known as:
DOB:
Sex:
Mother's name:

### Follow-Up Information
Specialist group: ☐ N/A
Date first seen by consultant: ☐ N/A
Date of initial consult, if phone consult only:
Total CK:
Initial total CK collection date:
Initial total CK result date:
Total CK value:
Total CK reference range:
Total CK units:
Clinical muscle biopsy
Biopsy result:
Biopsy comments:

### Diagnosis
Clinical diagnosis/outcome: Select from Dropdown
If other, specify here:
Was diagnosis made prenatally? ◯ Unknown ◯ No ◯ Yes
Has the family been notified of the diagnosis? ☐ Yes ☐ No
*If yes, MDH long-term follow-up attempts to provide families with resources, when applicable

### Intervention/Treatment Information
Eligible for commercial **exon-skipping** therapy? Select from Dropdown
Exon skipping therapy received? Select from Dropdown
Exon-skipping therapy name: Select from Dropdown
If other, specify here:
Start date:

Eligible for **gene** therapy? Select from Dropdown
Gene therapy received? Select from Dropdown
Gene therapy name: Select from Dropdown
If other, specify here:
Start date:

Other intervention/treatment:
Other medication, specify: Start date:
Other, specify: Start date:

Intervention/treatment comments:

Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155
Phone (800) 664-7772, Fax (651) 215-6285
REV 07/2021

# Outcomes

# Data Included:

- Newborns with an abnormal DMD screen received by MDH between 2/24/25–8/31/25
- As of 9/22/25

# Between February 24 and August 31

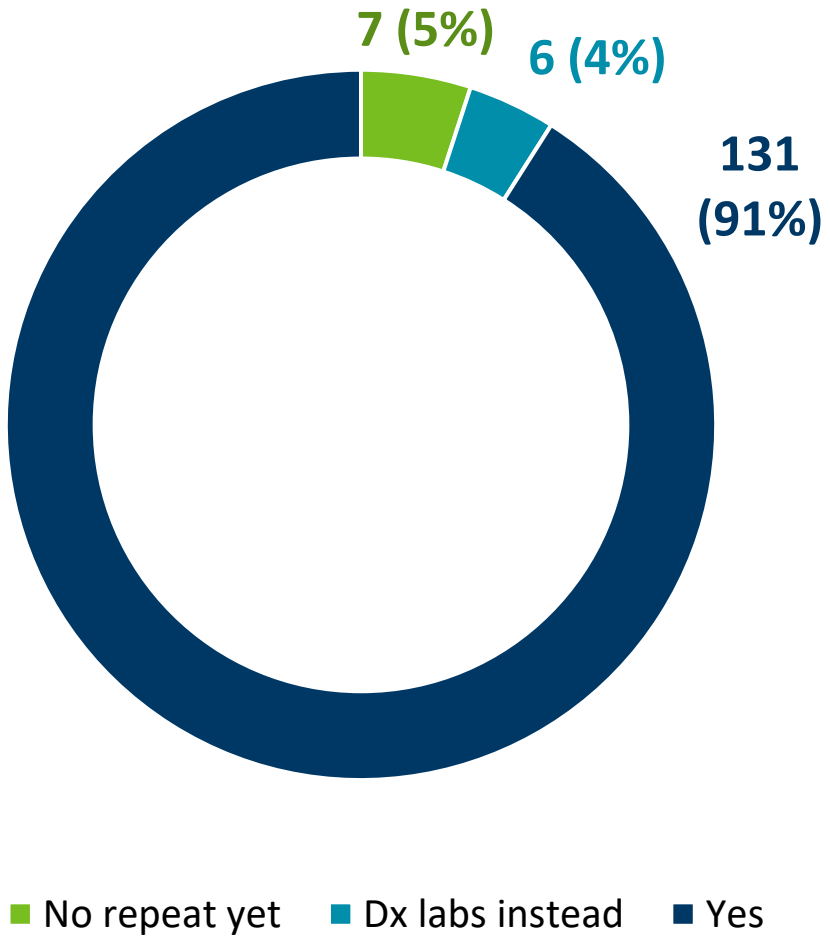
# 0.44%

Had a **borderline DMD** result

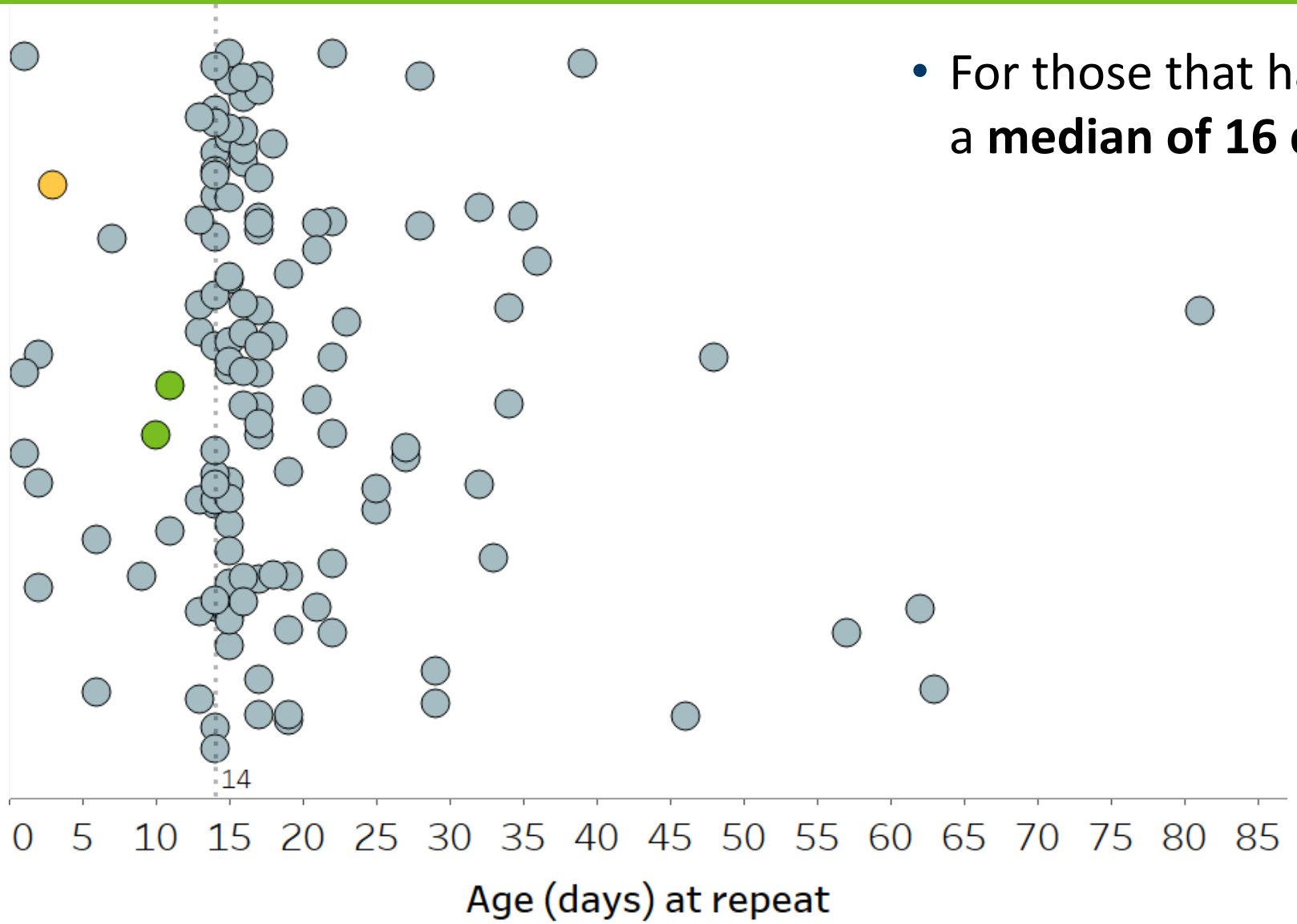
- 32,758 newborns screened
- 144 borderline DMD results

# 94% have follow-up CK testing after a borderline screen

- Most are getting a **repeat newborn screen** collected and sent to MDH
- Some have opted for **diagnostic CK labs** instead
- Of the 7 **without a repeat**, 3 are still pending, 4 have been closed due to lack of response in our follow-up time frame (3 females, 1 male with normal rapid whole exome)



# All repeats collected >14 days have been WNL

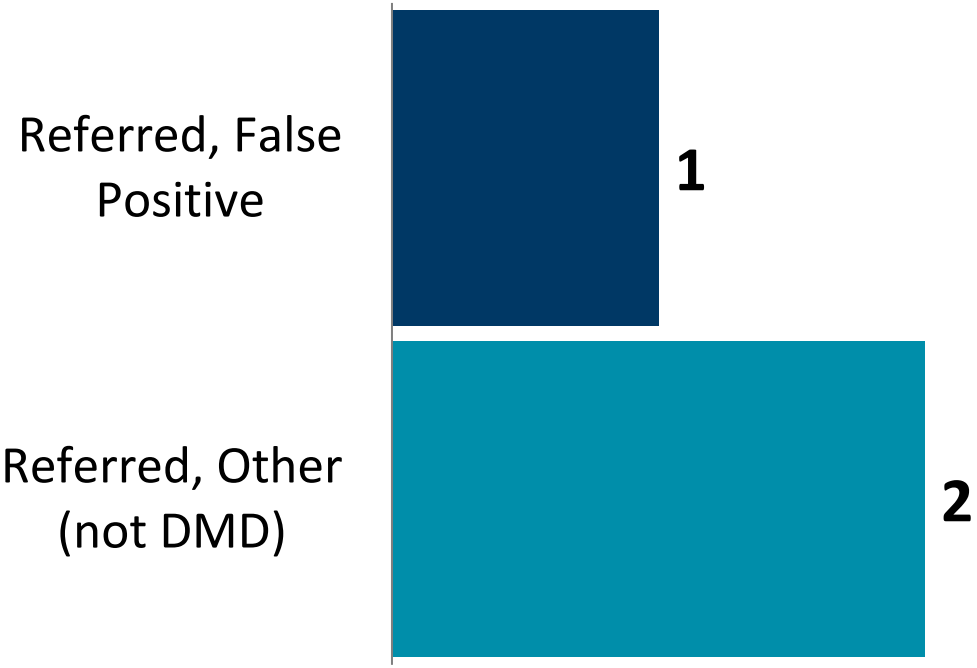



- For those that have a repeat, they are collected a **median of 16 days after birth (1–81 days)**

Outcome of repeat screen
- Positive
- Borderline
- WNL

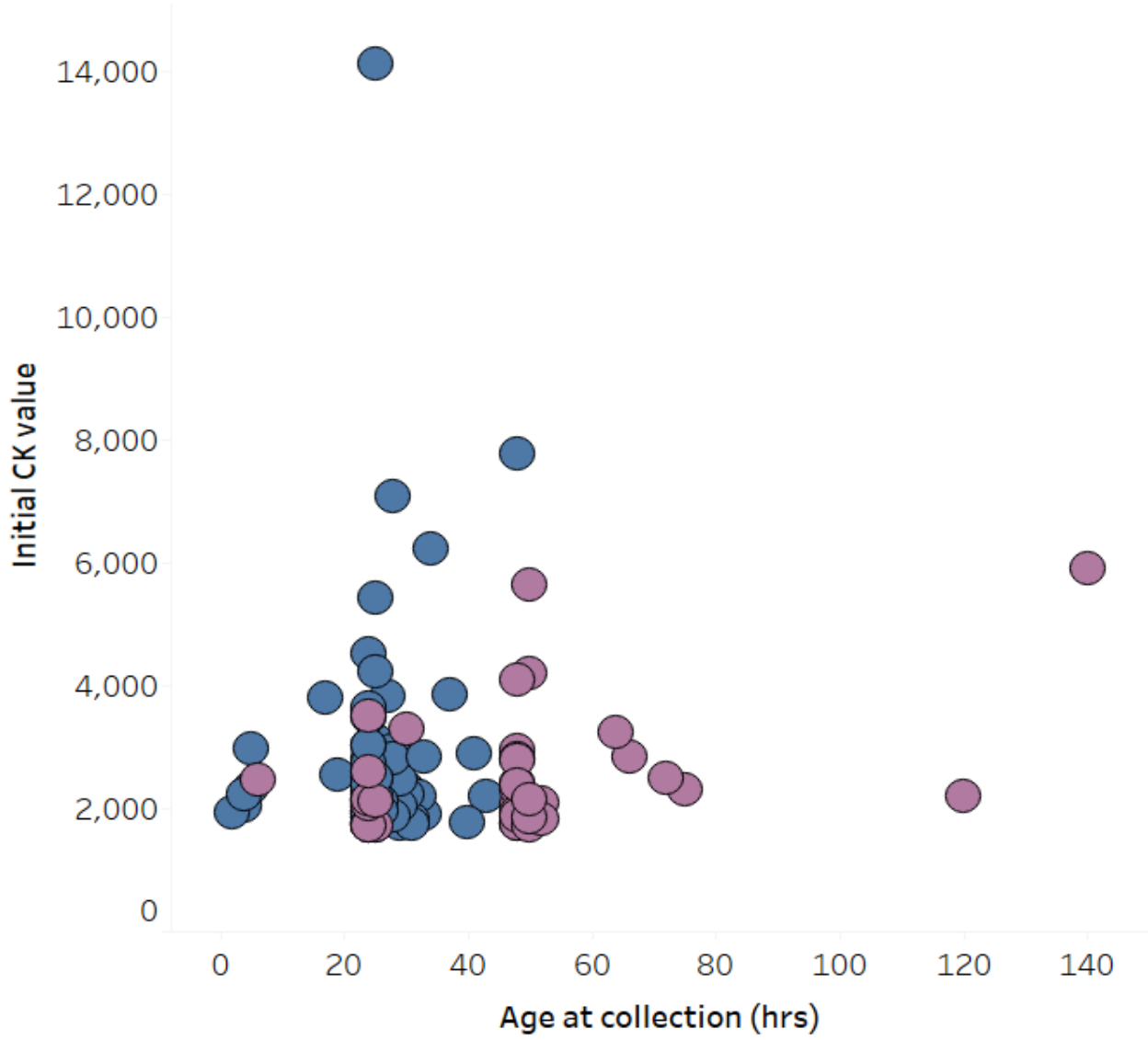
# No Infants with DMD have been Identified

- Of those with complete follow-up:
  - 134 (98%) had normal repeat CK testing
  - **3 (2%) have been referred** for diagnostic follow-up
    - 2 were referred after positive repeat screen
    - 1 was referred after abnormal clinical CK (instead of NBS)



# Birth Injury Associated with Borderline DMD Screen

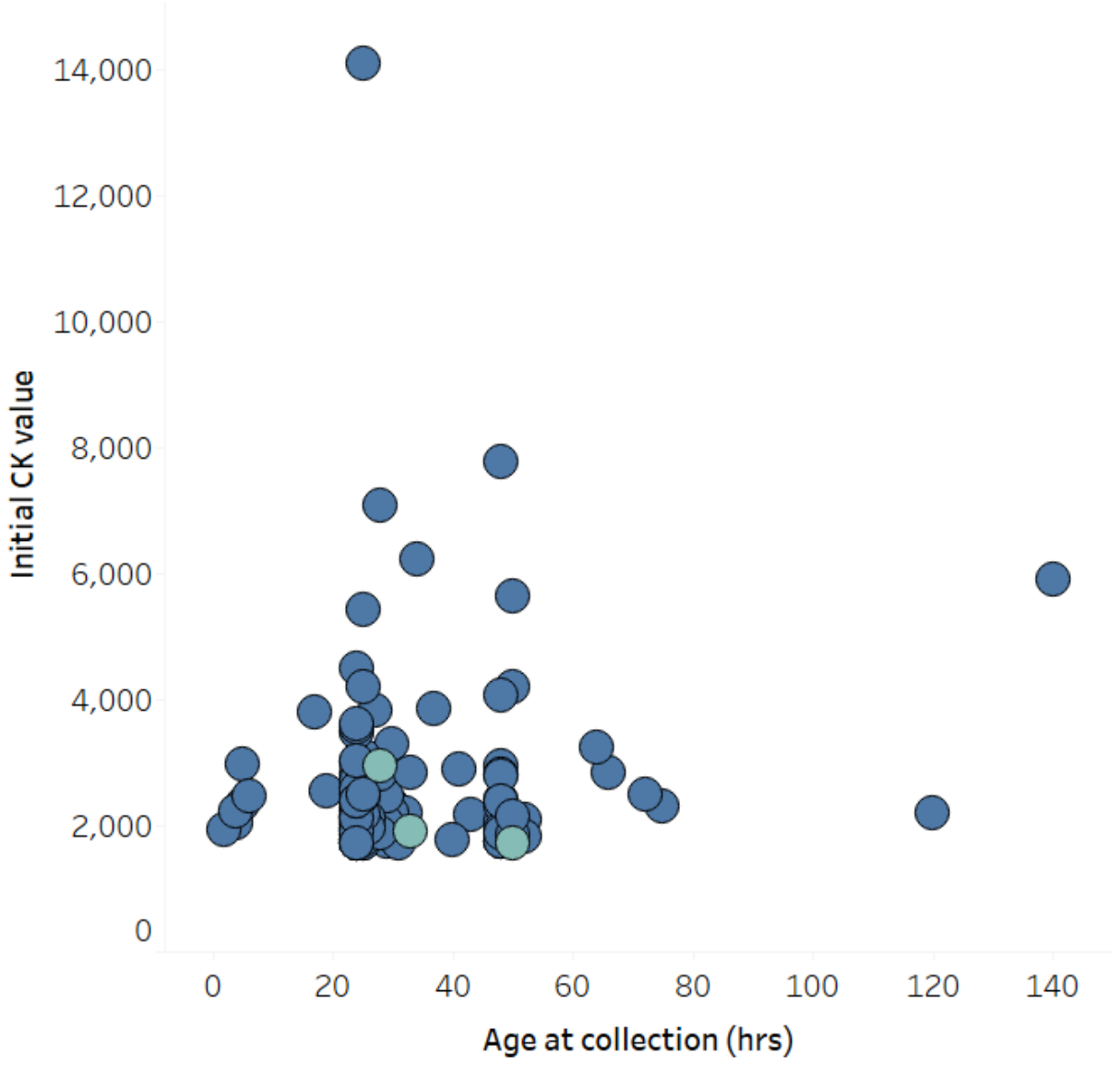
- 36 (25%) have a **birth injury** noted in NBS notification
- Most are shoulder dystocia
- Almost certainly an underestimate



# Few Low Birthweight Newborns Have a Borderline DMD Screen

- Of the newborns with a borderline DMD screen, only **3 were <2,000 grams** (LBW)
  - 2% of borderline screens are LBW
  - 0.3% of LBW newborns have borderline result

  (3% of our birth population is LBW)



# Insights

# Cost Considerations

- GSP instrument
- Kit costs
- Staff- Laboratory and/or follow-up
- Laboratory Information Management System Changes
- Molecular Testing

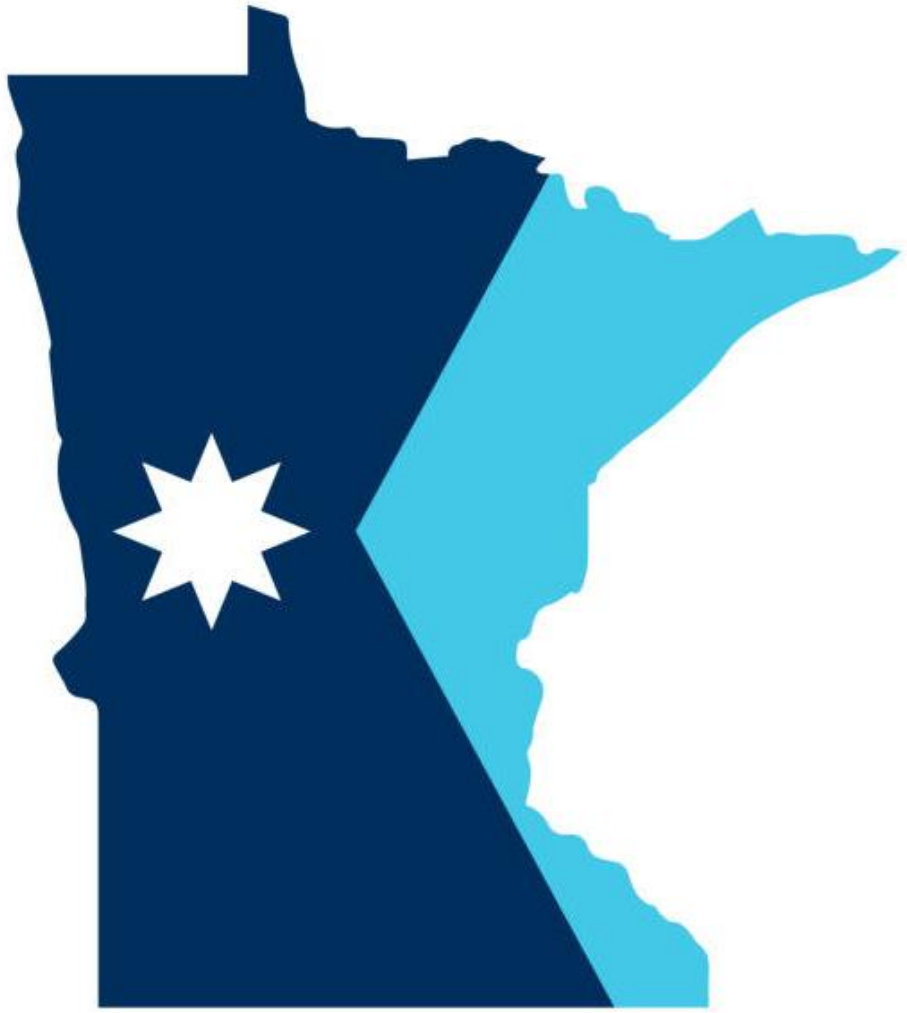
# No DMD Case = Unsettling

- One of our three positive cases has presumed presymptomatic limb girdle (unconfirmed as family is electing not to pursue additional testing at this time)
- We don’t have a case of DMD yet
  - We would have expected to have about 3 males by this time using prevalence of 1 in 5,000
  - We recently screened our first DMD case (brother with DMD)
    - Initial specimen was borderline for DMD
    - Family refusing follow-up… “has faith and thinks son will not develop disease”

# Repeat Newborn Screen Hassles

- Difficulty getting repeat NBS for rural and out of state babies – prompted shift
  - Babies born/screened in MN but live in another state – cannot get a repeat screen from their state's program if they do not screen for DMD so have to travel back to MN
  - Distance to nearest birth hospital for some more rural families is burdensome vs collecting clinical labs
  - Despite recommendation for repeat NBS, providers ordering clinical CK labs
- Clinical CK labs not so simple either
  - Some providers ordering clinical labs are misinterpreting CK-MB

DEPARTMENT OF HEALTH



# Questions?