

Duchenne Muscular Dystrophy was added to the Ohio Newborn Screening Program beginning April 22, 2024.

The Ohio Department of Health (ODH) is the first state health department to screen for Duchenne Muscular Dystrophy (DMD), a rare genetic muscle disease which is the most common form of muscular dystrophy, through their Newborn Screening Program.

Starting on Monday, April 22, Ohio screens all newborns for the enzyme muscle creatine kinase (CK-MM) which, if elevated, may be an indication of DMD.

DMD is an x-linked disorder that causes progressive muscle degeneration and weakness. DMD primarily affect males, but some female carriers may also develop symptoms. While DMD symptoms may appear as early as four months of age, symptoms may not be obvious until two to three years of age.

Symptoms usually start with developmental delays, particularly motor delays, or difficulty when standing, walking/abnormal gait, going up and down stairs, or getting up from sitting on the floor. While there is no cure, treatments include medications, supportive therapies, and gene therapy.

Screening Flow

CK-MM is screened on every newborn dried blood spot specimen received by the ODH Newborn Screening Program. The CK-MM cutoff levels will be interpreted based on age at collection **and** a minimum weight > 2,000 grams (4.4 pounds). When both criteria are met, the tables below reflect each cutoff level and result interpretation. CK-MM levels less than the cutoff for each age at collection are considered within range and having a low risk of DMD.

<u>Age at Collection</u>	<u>Cutoff (ng/mL)</u>	<u>Interpretation</u>
0 – < 48 hours	< 1,990	Within Range
48 – < 72 hours	< 1,430	Within Range
72 – < 168 hours	< 571	Within Range
≥ 168 hours	< 571	Within Range

<u>Age at Collection</u>	<u>Cutoff (ng/mL)</u>	<u>Interpretation</u>
0 – < 48 hours	> 1 990	Inconclusive
<u>Age at Collection</u>	<u>Cutoff (ng/mL)</u>	<u>Interpretation</u>
≥ 168 hours	≥ 571	Elevated

There are reasons, other than muscle disease, why a CK-MM level may be high during the newborn period. Some reasons include birth factors, gestational age, and birth weight.

Notification of Screening Results

The primary care provider (PCP), as indicated on the newborn screening card, will receive all newborn screening results. If a CK-MM level is inconclusive or elevated, the PCP will receive an *Action Required – Abnormal Newborn Screening* letter that contains recommended follow-up action steps. A DMD informational sheet will also be included with elevated CK-MM screening results which can be shared with the parent(s) or legal guardian.

The ODH public health laboratory will call the PCP when an elevated CK-MM screening result is noted and then will call the regional specialty team selected by the PCP.

When a PCP receives an *Action Required – Abnormal Newborn Screening* letter, the PCP should communicate screening results to the parent(s) or legal guardian and indicate that additional testing is needed to determine if their infant has a genetic muscle condition or if the increased CK-MM level has returned to “within range”.

Inconclusive & Elevated Result Follow-Up

ODH is requesting the PCP repeat testing either by ordering a second newborn screening panel or a blood test to specifically measure the total CK level for all inconclusive CK-MM screening results.

ODH recommends this repeat testing be conducted once an infant is 14 days of age or older **and** weighs at least 2,000 grams (4.4 pounds).

This second test will determine if levels have returned to “within range” or remain elevated resulting in the need for further testing and a referral to a DMD specialist. It is important to follow up on all inconclusive and elevated CK-MM results to determine if it is a persistent level that requires further evaluation.

Questions

The Ohio Department of Health is committed to the health and well-being of all Ohio infants and the addition of DMD to our newborn screening program continues this commitment. Early diagnosis and treatment for DMD can slow the progression of muscle weakness and improve overall health.

The ODH public health laboratory appreciates your expertise and thanks you for caring for Ohioans infants. Should you have questions, please feel free to call us at 1-888-634-5227 (ODH-LABS) option 1 or email odhlabs@odh.ohio.gov.