

# FAST FACTS ABOUT NEWBORN SCREENING FOR DUCHENNE MUSCULAR DYSTROPHY



## WHAT IS DUCHENNE?

Duchenne muscular dystrophy is the most common pediatric muscular dystrophy. It causes progressive muscle loss, cardiorespiratory failure, and ultimately premature death. Duchenne is X-linked and affects about one out of every 5,000 males at birth worldwide.

Duchenne is caused by a variant in the dystrophin gene, resulting in absent or nonfunctional dystrophin protein. Without functional dystrophin protein, muscles become damaged and are unable to repair themselves properly. This genetic disorder can be passed from parent to child, or it can result from a random spontaneous genetic mutation. In fact, most cases occur in families with no previous history of Duchenne.

Duchenne progresses differently for every person, but it ultimately results in loss of muscle function. Natural history data based on treatment initiated at the typical age of diagnosis shows that the average age of loss of ambulation is 12 years. Duchenne is universally life-limiting, with an average life expectancy of mid-20s to 30s.

“Despite massive advances in therapy, the average age of diagnosis for Duchenne has not changed in 30 years. If we are to have any hope of getting current treatment... and curing children born with this disease in the future, we need universal newborn screening. There is simply no other way.”

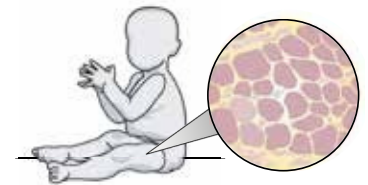
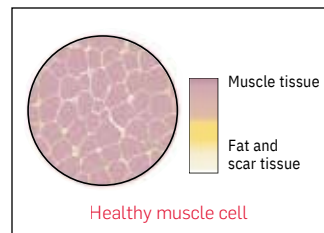
**- Mother of two children with Duchenne**

## WHY SCREEN FOR DUCHENNE AT BIRTH?

Approximately 360 babies with Duchenne are born each year in the United States, and evidence of muscle damage is present in infancy. Without newborn screening, families typically notice symptoms of Duchenne in their child by age 2, but most of those families will not receive a diagnosis until age 5.<sup>[1]</sup> This delay is even longer in families who are marginalized by race or income inequality.<sup>[2][3]</sup> By age 5, children with Duchenne have significant and irreversible muscle damage.

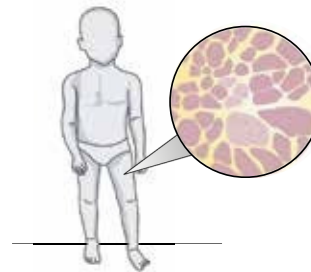
There are now eight FDA-approved treatments specific to Duchenne, four of which are approved for use in infancy. Newborn screening can eliminate the delay in diagnosis and provide families the opportunity for treatment years earlier, when children have more functional muscle tissue remaining.

### Muscle degeneration begins well before the average age of diagnosis



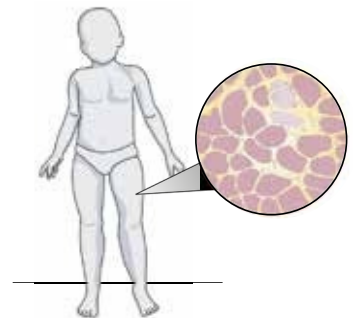
At 1 year, muscle degeneration is already evident.

Typical Duchenne muscle degeneration, as seen in muscle biopsy of 1-year-old Duchenne patient



At 3 years, the average age when parents begin noticing symptoms, muscle degeneration is more pronounced.

Typical Duchenne muscle degeneration, as seen in muscle biopsy of 3-year-old Duchenne patient



By age 5, the average age of Duchenne diagnosis, muscle degeneration is significant.

Typical Duchenne muscle degeneration, as seen in muscle biopsy of 5-year-old Duchenne patient

Illustrations based on characterization of DMD muscle phenotype, a Hematoxylin & Eosin (H&E) staining of Control (Ctr) and DMD muscles at different time points as published Cardone N, et al. Acta Neuropathologica Communications. (2023)11:167.

## HOW IS NBS FOR DUCHENNE PERFORMED?

The first-tier screen for Duchenne evaluates CK-MM, the muscle isoform (muscle version) of a marker called creatine kinase. CK-MM is a biomarker of muscle damage and is highly elevated in newborns and children with Duchenne. If a baby's CK-MM is high and stays high, confirmatory testing—DNA sequencing of the dystrophin gene, DMD—is required. The presence of a pathogenic (disease-causing) variant in DMD confirms the diagnosis. In some states, both the screening and confirmatory test can be performed on the standard NBS dried blood spots, with no additional blood draw required.

## IS THERE AN FDA-AUTHORIZED NBS TEST FOR DUCHENNE?

Yes. The FDA authorized the first Duchenne NBS test, manufactured by Revvity, in 2019.<sup>[4]</sup>

## CAN DUCHENNE NBS BE EASILY INTEGRATED INTO EXISTING NBS?

Yes. Duchenne NBS based on CK-MM uses the same dried blood spots currently collected and the Genetic Screening Processor technology already available in most NBS laboratories.

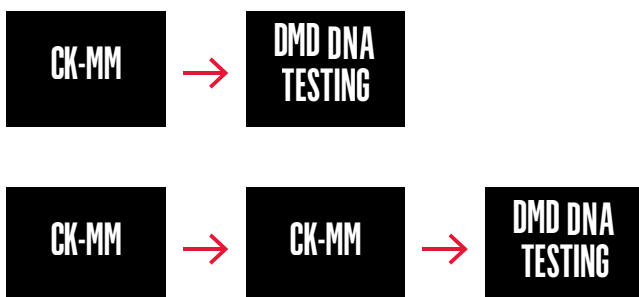
## HOW ACCURATE IS DUCHENNE NBS?

Accuracy and the false positive rate will depend on the screening algorithm and cut-offs chosen. When confirmatory DNA testing is included in the algorithm, the false positive rate is essentially zero.

## IS A SECOND-TIER OR CONFIRMATORY TEST REQUIRED FOR DUCHENNE NBS?

CK-MM cannot diagnose Duchenne, so second-tier or confirmatory testing is required. There are multiple potential algorithms for Duchenne NBS. A preferred algorithm in states with genetic testing capabilities or who are currently using a genetic testing laboratory for NBS for other conditions would include a confirmatory test, specifically DNA analysis of the DMD gene. Confirmatory testing for Duchenne could be performed on the dried blood spot.

### Possible Duchenne NBS Algorithms



## WHY NOT JUST SCREEN BOYS FOR DUCHENNE INSTEAD OF EVERY BABY?

NBS programs do not account for determination of sex, as it would add another step to the testing procedure and increase overall costs. Sex determination is a complex screening process and could increase false test results. In fact, of all the NBS programs across the world, not a single one includes DNA-based sex-specific screening.

Male-only testing also risks missing positive female carriers. A subset of female carriers will have Duchenne-like symptoms. By one estimate, one in every 4088 girls is a carrier of a pathogenic DMD variant who could be helped by early diagnosis.<sup>[5]</sup> NBS may be able to identify these girls who will benefit from intervention and may be eligible for therapies. All female carriers are at risk for cardiomyopathy, and cardiac screening is recommended for all adult female carriers.

## WHAT IS THE COST TO INCLUDE DUCHENNE ON THE NBS PANEL?

The cost will depend upon the algorithm and cut-offs chosen. Each state will determine how to approach NBS for Duchenne and whether it will include genetic testing in its process. When confirmatory genetic testing is performed for the small fraction of screen-positive babies, it is estimated that it would cost approximately \$8 per-infant-tested to screen for Duchenne with labor and referral costs <sup>[6]</sup>.

## IS DUCHENNE ON THE FEDERAL RECOMMEND UNIFORM SCREENING PANEL (RUSP)?

The RUSP is the federal recommendation for which conditions all babies in the United States should be screened for at birth. Duchenne is on the RUSP.

Parent Project Muscular Dystrophy led the nomination of Duchenne to the RUSP in 2022 in collaboration with its co-nominator, the Muscular Dystrophy Association. In 2025, less than a month from making a decision, the Health Resources and Services Administration (HRSA) committee that reviews condition nominations and makes recommendations to the Secretary of the Department of Health and Human Services (HHS) was dissolved. PPMD and other advocates worked with HRSA and HHS to help the department complete and publish the Duchenne evidence review. A public workshop was held to review the evidence with input from the lead of the evidence review, Duchenne experts, and members of the dissolved committee. HRSA published a request for public comment regarding the addition of Duchenne to the RUSP, and all of these pieces of evidence were taken into consideration as HRSA considered whether to recommend Duchenne's addition to the HHS Secretary.

In December 2025, HHS reported that HRSA recommended adding Duchenne to the RUSP and the Health Secretary formally approved its addition on December 16, 2025.<sup>[7]</sup>

## WHICH STATES SCREEN FOR DUCHENNE?

Since 2024, two states - Ohio and Minnesota - have begun newborn screening for Duchenne. Several other states have approved legislation to begin screening in the next few years. There are also many states actively reviewing the possibility of including Duchenne on their NBS panels through their NBS advisory committees and/or legislative bodies.

## WHY DO STATES NEED TO ADD DUCHENNE TO NBS NOW?

The RUSP is only a recommendation for which conditions all states should include in their programs, and many states do not currently screen for every condition on the RUSP. Although some states have RUSP-alignment laws that define a timeline in which they must review or add a new RUSP condition, most states have an undefined timeline to review Duchenne for addition, ensure that the necessary resources are available, and work toward implementation.

Whether RUSP-aligned or not, it typically takes several years for a state to add a new condition to their program and begin screening. It is vital that states consider the addition of Duchenne now, before more of our children's time and muscle is lost.

## WHAT IS THE IMPACT OF A DELAYED DUCHENNE DIAGNOSIS?

The average delay between a family identifying symptoms and a Duchenne diagnosis is 2.5 years. This diagnostic odyssey results in significant economic and emotional impacts for the family which continue long-term, as delays in diagnosis result in higher annualized medical costs on a per patient basis.<sup>[8]</sup>

### DELAYED DIAGNOSIS DRAMATICALLY INCREASES COSTS FOR FAMILIES<sup>[8]</sup>

- \$211,229 total medical cost and productivity loss per family
- 20 lost days of work for caregivers per year
- 17 out-of-state trips for medical care

## WHAT ARE THE BENEFITS OF DIAGNOSIS THROUGH NBS?

An expert group of Duchenne clinicians developed and published clinical guidance for care following a newborn screening diagnosis.<sup>[9]</sup> After NBS, families consult with pediatric neuromuscular care teams to discuss the most appropriate treatment course for each child. They have the potential to initiate therapies such as exon skipping, gene therapy, and steroids years earlier than they would if diagnosed through traditional pathways. They may be able to enroll in clinical trials.

Establishing care in infancy also enables referrals to early intervention services such as speech, behavioral, physical, and occupational therapy. Diagnosis through NBS has benefits for family members as well, including identifying at-risk relatives, like carrier mothers who are at risk for heart disease and siblings who may be carriers or affected. Many families also report using this information in future family planning.

**QUESTIONS? CONTACT [ADVOCACY@PARENTPROJECTMD.ORG](mailto:ADVOCACY@PARENTPROJECTMD.ORG)**

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## WHAT TREATMENTS ARE AVAILABLE FOR DUCHENNE?

There are currently eight FDA-approved therapies for Duchenne muscular dystrophy, with more than 40 additional potential therapies in development. The approved therapies slow progression and enable children with Duchenne to have more years of walking, using their hands, breathing well, and spending time with their families.

DUCHENNE TREATMENT	% ELIGIBLE FOR TREATMENT	ESTABLISHED BENEFITS OF TREATMENT
<b>EXON SKIPPING THERAPIES</b>		
Amondys 45: all ages Exondys 51: all ages Viltepso: all ages Vyondys 53: all ages	30%	Prolonged ambulation, slower decline in lung function, increased lifespan <sup>[10, 11, 12]</sup>
<b>CORTICOSTEROIDS</b>		
Agamree: age 2 and up Emflaza (deflazacort): age 2 and up Prednisone: all ages	100%	Prolonged ambulation and upper limb use, slower decline in lung function, decreased scoliosis, increased lifespan <sup>[13,14]</sup>
<b>GENE THERAPY</b>		
Elevidys: age 4 and up	94%	Gains or stabilization on functional testing <sup>[15]</sup>
<b>SMALL MOLECULE INHIBITOR</b>		
Duvyzat: age 6 and up	100%	Activates muscle repair mechanisms to increase muscle fiber regeneration, reduce inflammation, and reduce fibrosis <sup>[16, 17]</sup>

## REFERENCES

1. Ciafaloni et al. J Pediatr. 2009;155:380-385.
2. Counterman et al. Muscle Nerve. 2020;61(1):36-43.
3. Mann et al. Neuroepidemiology. 2023;57(2):90-99.
4. <https://www.fda.gov/news-events/press-announcements/fda-authorizes-first-test-aid-newborn-screening-duchenne-muscular-dystrophy>
5. Han et al. BioMed Res Int. 2020;8396429.
6. Parad, et al. Int. J. Neonatal Screen. 2021;7, 77.
7. [www.federalregister.gov/documents/2025/12/22/2025-23573/addition-of-duchenne-muscular-dystrophy-to-the-recommended-uniform-screening-panel](https://www.federalregister.gov/documents/2025/12/22/2025-23573/addition-of-duchenne-muscular-dystrophy-to-the-recommended-uniform-screening-panel)
8. EveryLife Foundation. The Cost of Delayed Diagnosis in Rare Disease: A Health Economic Study. 2023.
9. Lorentzos et al. Neuromuscular Disorders. 2024;45:104467.
10. Mitelman et al. J Neuromuscul Dis. 2022;9(1):39-52.
11. Ilff et al. Presentation at 2022 World Muscle Society
12. Grabich et al. Poster at 2023 MDA scientific conference
13. McDonald et al. Lancet. 2018;391(10119):451-461.
14. Raudenbush et al. Spine. 2016;41(17):E1030-E1038.
15. Mendell et al. Front Cell Dev Biol. 2023;11:1167762.
16. Mercuri et al. Lancet Neurology. 2024; 23(4): 393-403.
17. Bettica et al. Neuromuscular Disorders. 2016; 26(1): 643-649.