# Duchenne Muscular Dystrophy

## BACKGROUND

Parent Project Muscular Dystrophy

### What is Duchenne Muscular Dystrophy?

Duchenne muscular dystrophy is a genetic disorder characterized by the progressive loss of muscle. It is a multi-systemic condition, affecting many parts of the body, which results in deterioration of the skeletal, heart, and lung muscles.

Duchenne is caused by a change in the dystrophin gene. Without dystrophin, muscles are not able to function or repair themselves properly. Becker muscular dystrophy, which typically progresses more slowly than Duchenne, occurs when dystrophin is produced, but not in the normal form or amount.

Because the dystrophin gene is found on the X-chromosome, it primarily affects males, while females are typically carriers. However, some females can manifest varying ranges of physical symptoms of Duchenne and are therefore called "manifesting carriers".

### Who is Parent Project Muscular Dystrophy?

Parent Project Muscular Dystrophy (PPMD) was founded in 1994 by President Pat Furlong, mother to two boys with Duchenne, and a dedicated group of fellow parents determined to change the future of Duchenne muscular dystrophy. At a time when families were told there was "no hope and little help," they built an organization focused on advocacy and resources to drive research, improve care, and accelerate progress toward treatments.

A rare disease, Duchenne is the most common fatal genetic disorder diagnosed in childhood, affecting approximately 1 in 5,000 male live births. What began as a grassroots movement has grown into the leading global organization in Duchenne, advancing research, shaping standards of care, influencing federal policy, and helping bring multiple therapies to approval.

Today, because of PPMD's leadership and the strength of its community, families have better access to information, care, and treatment options and the momentum continues.

# PPMD's MISSION & IMPACT

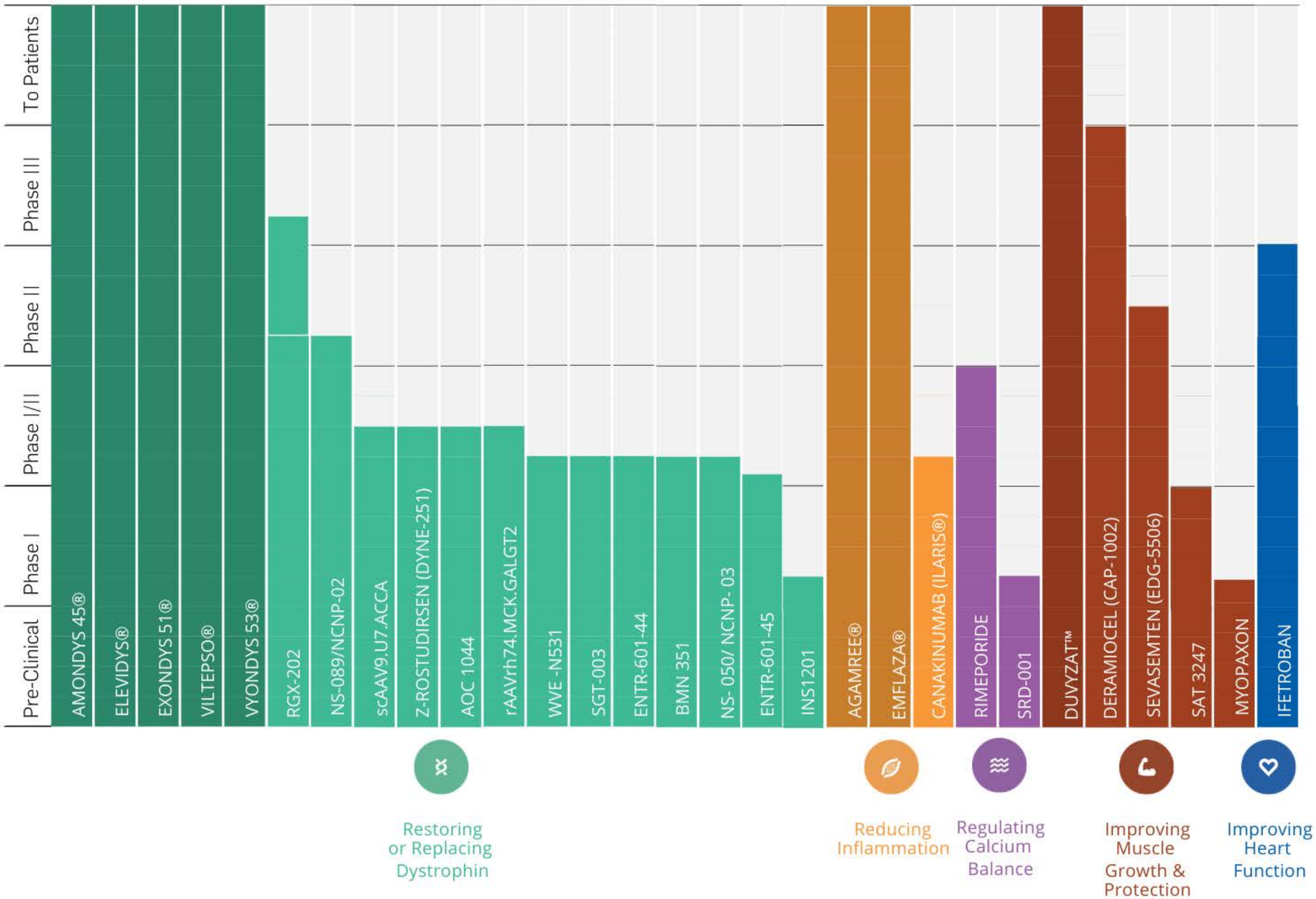
**Parent Project Muscular Dystrophy (PPMD)** fights to end Duchenne. We accelerate research, raise our voices to impact policy, demand optimal care for every single family, and strive to ensure access to approved therapies.

PPMD is proud to have played a vital role in every single victory against Duchenne since 1994 and our compassion, strength, and innovation continue to lead this community.

# DUCHENNE DRUG DEVELOPMENT PIPELINE

Parent Project Muscular Dystrophy
JOIN THE FIGHT. END DUCHENNE.

The Drug Development Pipeline is full of potential treatments that are being tested. These include therapeutic approaches that restore or replace dystrophin and those that treat Duchenne symptoms. The goal? To test combinations of these therapies to create the best "cocktail" for each patient.




March 2026

# Duchenne Muscular Dystrophy
## CARRIERS

Parent Project Muscular Dystrophy

## What Does Being a Carrier Mean?

A carrier of Duchenne muscular dystrophy (Duchenne) is typically a female (XX) with one altered copy of the dystrophin (DMD) gene, located on the X chromosome. This genetic change may be inherited from a parent or occur spontaneously (de novo variant). While many carriers do not experience symptoms, they do have an increased risk for heart disease, and can have additional physical or psychological impacts.

## What Are the Health Risks for Carriers?

- **Heart:** Female carriers are at an increased risk of developing cardiomyopathy (a weakening of the heart muscle) or arrhythmias (irregular heartbeats).
- **Muscle:** Some carriers experience difficulty with physical activities such as climbing stairs or running. They may also have muscle cramps, pain, and fatigue easily. In rare cases, muscle symptoms progress over time and are significant enough to be called muscular dystrophy.
- **Emotional/Psychological:** Carriers may experience psychological impacts related to the risk of passing on the condition or developing symptoms, and they have higher rates of learning and behavioral differences related to the genetic variant.

## Health Monitoring and Care for Carriers

To manage potential health risks, carriers should take a proactive approach:

### 1 Cardiac Monitoring

- Begin heart evaluations by early adulthood and repeat testing as recommended by a cardiologist familiar with Duchenne.
- Testing includes heart imaging (echocardiogram/MRI) and screening for arrhythmia (EKG)

### 2 Neuromuscular Care

- If muscle symptoms (weakness, cramping) develop, consult a neuromuscular specialist.
- Physical therapy or other interventions may be recommended for managing symptoms.

### 3 Family Planning Support

- Make an appointment with a genetic counselor to learn about the risk of Duchenne for other family members.

### 4 Mental and Emotional Well-being

- Connect with counseling or peer support groups to address concerns and reduce stress related to being a carrier.
- If a learning or behavioral disability is suspected, meet with a psychiatrist for an evaluation.

## Genetic Testing for Carrier Status

Genetic testing identifies variants in the dystrophin gene to determine if someone is a carrier.

**Who Should Be Tested?** Testing can be considered for all pregnant people or those planning a pregnancy, and especially for:

- Individuals with a family history of Duchenne.
- Mothers of children with Duchenne.
- Women with unexplained muscle weakness or cardiac symptoms.

Testing is a simple blood or cheek swab test and can provide crucial information for health management and family planning.

## Reproductive Considerations

Carriers have a 50% chance of passing the altered gene to their children:

- **Sons:** Inheriting the variant leads to Duchenne muscular dystrophy.
- **Daughters:** Inheriting the variant leads to health risks associated with carrier status

Options to reduce transmission risk include IVF and other means of growing a family, such as adoption.

## PPMD Support and Resources

PPMD is dedicated to providing carriers with tools, knowledge, and connections to manage their health and improve their quality of life:

- **Carrier Research:** Ongoing studies explore the unique health needs of carriers, focusing on cardiac, muscular, and emotional well-being.
- **Support Programs:** PPMD offers support groups, educational webinars, and resources tailored to carriers and their families.
- **Advocacy:** PPMD works to ensure carriers have access to appropriate healthcare, insurance coverage, and genetic counseling.

## Empowering Carriers to Take Charge of Their Health

Carriers play a crucial role in advancing the understanding of Duchenne and ensuring better outcomes for future generations. By staying informed, undergoing regular health evaluations, and seeking support, carriers can lead healthy, fulfilling lives.

# CONTACT PPMD FOR MORE INFORMATION

Together, we can ensure every carrier has the resources and care they need.

**www.parentprojectmd.org** **info@parentprojectmd.org** **1-800-FIGHTMD**

# Newborn Screening

## for Duchenne Muscular Dystrophy is now a Federal Recommendation

Parent Project Muscular Dystrophy

In December 2025, Duchenne muscular dystrophy was added to the Recommended Uniform Screening Panel (RUSP), the Department of Health and Human Services' list of conditions recommended for inclusion in every state's newborn screening program.

### Why State Action Is Needed

Although Duchenne is now recommended for universal screening, **each state must take its own steps to implement testing**, including:

- Legislative or regulatory authorization
- Laboratory readines
- Clinical referrals and care coordination
- Provider and family education

This requires **leadership, coordination, and investment**. Without state action, newborn screening for Duchenne cannot reach families.

### Why is it important that every state screens babies for Duchenne?

Duchenne is a severe, progressive muscle weakness disorder that primarily affects boys, occurring in approximately 1 in 5,000 male births.

People with Duchenne lose their ability to walk and can have serious heart and lung complications. Most people are diagnosed late, at age 4 or later, and lose years of treatment that could have slowed their progression.

Newborn screening enables diagnosis at birth, allowing:

- Reduced diagnostic delays
- Earlier treatment and care
- Improved health outcomes
- Access to early clinical trials

## NEXT STEPS

Duchenne advocates, clinicians, and public health partners will continue to work collaboratively to advance implementation efforts in states across the country.

**We look forward to further engaging with policymakers to explore pathways to bring Duchenne newborn screening to families in your state.**

The Honorable Robert Aderholt
Chair
Labor, HHS, Education,
& Related Agencies Subcommittee
Subcommittee Committee on Appropriations
Washington, DC 20515

The Honorable Rosa DeLauro
Ranking Member
Labor, HHS, Education,
& Related Agencies
Committee on Appropriations
Washington, DC 20515

The Honorable Andy Harris
Chair
Agriculture, Rural Development, FDA,
FDA, & Related Agencies Subcommittee
Subcommittee Committee on Appropriations
Washington, DC 20515

The Honorable Sanford Bishop, Jr.
Ranking Member
Agriculture, Rural Development,
& Related Agencies
Committee on Appropriations
Washington, DC 20515

The Honorable Ken Calvert
Chair
Defense Subcommittee
Committee on Appropriations
Washington, DC 20515

The Honorable Betty McCollum
Ranking Member
Defense Subcommittee
Committee on Appropriations
Washington, DC 20515

Dear Chairs Aderholt, Harris, and Calvert and Ranking Members DeLauro, Bishop, and McCollum:

Thanks to the leadership of Congress, starting with the passage of the bipartisan Muscular Dystrophy Community Assistance, Research and Education (MD CARE) Act in 2001, significant progress has been made over the quarter-century fight to end Duchenne and Becker Muscular Dystrophy (DBMD), the most common lethal genetic disorder diagnosed during childhood. As you prepare your Fiscal Year (FY) 2027 appropriations bill, we urge you to include provisions to help further these pursuits, particularly to advance scientific breakthroughs, to accelerate therapy development, to ensure consistent high-quality care across the country, and to help improve life for patients and caregivers affected by this disease.

As a result of the MD CARE Act and subsequent amendments, federal commitments to research have expanded, helping spur scientific breakthroughs to develop potential therapies. These commitments have also leveraged significant non-federal funding from academic institutions, industry, and venture investors in a true public-private partnership model. In addition to research breakthroughs, the MD CARE Act has helped capture important epidemiological data, information that has helped standardize and improve patient care and inform payer decision making.

Our FY 2027 Duchenne and Becker Muscular Dystrophy appropriations request contains language and provisions to help continue and strengthen these and other ongoing initiatives. Specifically, the request would:

- Fund the Duchenne Muscular Dystrophy Research Program within the Department of Defense (DOD) Congressionally Directed Medical Research Programs (CDMRP) at $13.5 million.

- Encourage the Food and Drug Administration (FDA) to:
    - Convene a public meeting to examine pathways for the continued use of natural history data as a control in Duchenne muscular dystrophy clinical trials.

- Direct the National Institutes of Health (NIH) to:
    - Continue advancing next-generation backbone chemistry for platform technologies in gene-targeted therapies, with a particular focus on more challenging and rare genotypes often not addressed in current gene-targeted therapy development.
    - Develop a multi-institute strategy to accelerate discovery of platform approaches to DNA- and RNA-directed therapies.

- Fund Muscular Dystrophy at $10 million and direct the Centers for Disease Control and Prevention (CDC) to:
    - Fund a cooperative agreement to understand and address the barriers to accessing care related to patient geography and other factors.
    - Expand CDC-supported surveillance systems to track outcomes of newborns with Duchenne identified through newborn screening (NBS).
    - Encourage the collection of data on these newborns through existing CDC programs.

- Direct the Health Resources and Services Administration (HRSA) to:
    - Conduct, in cooperation with CDC, an evaluation of newborn screening data collection and evidence generation for Duchenne muscular dystrophy.
    - Engage critical stakeholders (including state laboratory leadership, patient organizations, clinical organizations, public health organizations, and research organizations) in an evidence-informed process to generate national, state, and local priorities to address strengths and deficiencies identified in the evaluation.
    - In conjunction with HHS, designate a mechanism with appropriate authority, accountability, and resources to implement the resulting recommendations at both the federal and state levels, as recommended by the National Academies.

Much progress has been achieved in recent years, but more work remains to be done. The FY 2027 Duchenne MD request will focus federal energies toward the highest priority needs to accelerate the development of therapies and treatments and to improve life for all patients impacted by this disease.

Below is the specific language we are requesting:

**Department of Defense (DOD)**
*Congressionally Directed Medical Research Programs (CDMRP) Duchenne Muscular Dystrophy Research Funding:* $13.5 million (an increase from $12.5 million in FY26)

**Food and Drug Administration (FDA)**
*Duchenne and Becker Muscular Dystrophy* – While important progress has been made on Duchenne and Becker muscular dystrophy treatments, the unmet medical need remains high. Therapeutic options vary widely depending on a patient's genotype, age, and stage of disease and no available treatments are curative. While evolving treatment options and earlier diagnosis are leading to improved natural history for Duchenne in particular, this also complicates the ability to evaluate potential therapies without requiring a control group. Patients and families should be able to benefit from better treatment outcomes without the community having to revert to placebo-controlled trials for a condition that remains progressive and lethal. The Committee urges FDA to convene a public meeting to examine pathways for the continued use of natural history as a control for Duchenne muscular dystrophy trials.

**National Institutes of Health (NIH)**
*Duchenne and Becker Muscular Dystrophy* – NIH support for basic and translational research in Duchenne and Becker muscular dystrophy has enabled important advances in treating these conditions. At the same time, there continues to be significant unmet medical needs across the community. The treatment landscape varies widely depending on a patient's genotype, age, and stage of disease and no available treatments are curative. The Committee appreciates the NIH focus on funding rare disease research that benefits Duchenne patients as well as the Muscular Dystrophy Coordinating Committee's update of its Action Plan. With wider research and adoption of gene targeted therapies, the Committee recognizes a critical opportunity to advance platform technologies for targeted therapies that can be efficiently replicated across multiple rare mutations and conditions. The Committee encourages NIH to continue to advance next generation backbone chemistry for platform technologies, with a particular focus on more challenging and rare genotypes that are often not addressed in gene targeted therapy development. The Committee urges the NIH to develop a multi-institute strategy to accelerate discovery of platform approaches to DNA and RNA-directed therapies.

**Centers for Disease Control and Prevention (CDC)**
*BIRTH DEFECTS, DEVELOPMENTAL DISABILITIES, DISABILITIES, AND HEALTH*
$10 million for Muscular Dystrophy (an increase from $9 million in FY26)

*Duchenne Muscular Dystrophy* – The Committee recognizes that individuals living with Duchenne muscular dystrophy who have difficulties accessing multi-disciplinary care due to factors such as geography and health insurance coverage often are not benefiting from emerging treatment strategies. The Committee includes $1,000,000 for the CDC to enter into a cooperative agreement with an external organization with expertise in patient and caregiver engagement for Duchenne and Becker muscular dystrophy to analyze the outcomes for Duchenne and Becker muscular dystrophy treated both in and out of Certified Duchenne Care Centers, barriers to high-quality care, how to overcome them, and how to enable better health outcomes for Duchenne and Becker muscular dystrophy patients both in and out of Certified Duchenne Care Centers. Additionally, now that newborn screening for Duchenne has been added to the Recommended Uniform Screening Panel (RUSP), CDC should ensure that existing, CDC-supported surveillance systems are expanded to track outcomes of newborns with Duchenne identified through NBS and encourage such data to be collected through existing programs.

**Health Resources and Services Administration (HRSA)**
*Newborn Screening* – Even with nationwide newborn screening, critical gaps remain in our ability to systematically and longitudinally track laboratory generated, child-level, and system-level health outcomes. Efforts to date have not resulted in meaningful, coordinated evidence generation at the national level and resource gaps remain at the state level. The Committee directs HRSA, in cooperation with CDC, to conduct an evaluation of newborn screening longitudinal data collection and evidence generation. Further, the committee directs HRSA to engage critical stakeholders, including public health laboratories, patient advocacy organizations, clinical organizations, public health organizations, and research organizations to generate national, state, and local priorities to build from strengths and ameliorate deficiencies identified in the evaluation. Finally, HRSA and HHS should designate a mechanism with appropriate authority, accountability, and resources to enact the resulting recommendations on the federal and state levels, as recommended in the National Academies "Newborn Screening in the United States: A Vision for Sustaining and Advancing Excellence" (2025).

We understand the challenges the Committee faces in setting priorities within this critically important spending bill and appreciate its consideration of this request.

Sincerely,

DORIS MATSUI
Member of Congress

TROY BALDERSON
Member of Congress