Help End Duchenne Muscular Dystrophy

By Supporting Research, Care Considerations, and Therapy Development

Sign the FY24 Duchenne MD Appropriations Letter on Quill

Dear Colleague –

Duchenne muscular dystrophy and the milder form, Becker muscular dystrophy, are rare X-linked recessive diseases belonging to a group of conditions known as dystrophinopathies. Duchenne and Becker are caused by mutations in the dystrophin gene leading to the absence or reduced production of the dystrophin protein, a protein that is key to stabilization of the muscle cell membranes. This lack of dystrophin in muscle leads to progressive muscle weakness and loss over time causing premature death. Affecting 1 of every 5,000 boys, Duchenne is typically diagnosed during the first few years of life. A muscle wasting disorder, Duchenne gradually robs children of their ability to walk by their teenage years. Over time, their muscles weaken further to the point of paralysis, with most patients living only into their late 20s.

Although there are now five FDA-approved therapies that may help slow its progression, there is currently no cure for Duchenne or Becker. However, there is reason for hope, due in large part to the support and research funding Congress has provided:

- More than 35 potential therapies are in various stages of clinical testing.
- The life expectancy of the average Duchenne patient has increased by about 10 years over the past 10 years, driven in large part by development and dissemination of Care Standards.

Now is the time to continue building upon these successes and move closer to achieving the goal of ending Duchenne and Becker by supporting research, public health, and therapy development initiatives. We invite you to help keep this momentum going by signing the FY23 Duchenne Muscular Dystrophy appropriations sign-on letter. This year, we are requesting language to:

- Increase CDC's Muscular Dystrophy Program funding from $7.5 million to $8 million.
- Increase funding for the Duchenne Muscular Dystrophy Research Program within DOD's Congressionally Directed Medical Research Programs (CDMRP) from $10 million to $15 million.
- Evaluate the impact of the CDC Care Considerations on patient outcomes.
- Urge flexibility in the Advisory Committee on Heritable Disorders in Newborns and Children's review of pediatric and rare diseases given data constraints.
- Report to Congress about the scientific accomplishments of research conducted by the Wellstone Muscular Dystrophy Research Network Centers of Excellence program established in 2003

- Encourage FDA to make every effort to incorporate all relevant patient experience data across its regulatory obligations.

The full request is below. We urge you to co-sign this letter to advance these priorities and bring us closer to the day of ending Duchenne.

Please sign this letter on Quill. If you have any questions, please contact Jackie Weinrich (jackie.weinrich@mail.house.gov) with Rep. Matsui or Megan Porter (megan.porter@mail.house.gov) with Rep. Balderson.

Sincerely,

DORIS MATSUI
Member of Congress

TROY BALDERSON
Member of Congress

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

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

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

The Honorable Sanford Bishop, Jr.
Ranking Member
Agriculture, Rural Development, FDA,
& Related Agencies Subcommittee
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 Chairmen Aderholt, Harris, and Calvert and Ranking Members DeLauro, Bishop, and McCollum:

Thanks in large part to the leadership of Congress starting with the passage of the Muscular Dystrophy Community Assistance, Research and Education (MD CARE) Act in 2001, significant progress has been made over the past 20 years in the fight to end Duchenne Muscular Dystrophy (Duchenne MD), the most common lethal genetic disorder diagnosed during childhood. We are writing to urge that, as you prepare your Fiscal Year 2024 Appropriations bill, you 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 Fiscal Year 2024 Duchenne MD appropriations request contains language and provisions to help continue and strengthen these and other ongoing initiatives. Specifically, the request would:

- Increase funding for CDC's Muscular Dystrophy Program from $7.5 million to $8 million.

- Increase funding for the Duchenne Muscular Dystrophy Research Program within DOD's Congressionally Directed Medical Research Programs (CDMRP) from $10 million to $15 million.

- Direct the CDC to:
    - Evaluate the impact of incorporating the Care Considerations, as identified in CDC's recent report to the Committee, into the standards of care for DMD patients in terms of patient outcomes.
    - Collaborate with stakeholders on an initiative to integrate the Care Considerations into electronic health records to improve care, understand disease outcomes, and model disease progression.
- Urge the Health Resources and Services Administration (HRSA) to:
    - Expedite consideration of Duchenne Muscular Dystrophy for the Recommended Uniform Screening Panel for newborn screening, incorporate the patient community voice in the review, and develop appropriate flexible criteria given the challenges associated with collecting data for rare and pediatric diseases.
- Directs the National Institutes of Health (NIH) to:
    - Provide a public report to Congress on the key scientific accomplishments to date of the Wellstone Muscular Dystrophy Research Network Centers of Excellence and use this information to update the NIH website content regarding the program.
- Encourage the Food and Drug Administration (FDA) to:
    - Make every effort to incorporate all relevant patient experience data, including from patient advocacy organizations, across its regulatory obligations.

Much progress has been achieved in recent years, but much more work remains to be done. The FY 2024 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:

Centers for Disease Control and Prevention

BIRTH DEFECTS, DEVELOPMENTAL DISABILITIES, DISABILITIES, AND HEALTH
$8M for Muscular Dystrophy (increase from FY23 enacted funding of $7.5M)

Duchenne and Becker Muscular Dystrophy. – The Committee includes an increase of $500,000 to enhance Muscular Dystrophy research and disease surveillance initiatives, including the evaluation of the impact of incorporating the Care Considerations, as identified in the recent report to the Committee, into the standards of care for DMD patients in terms of patient outcomes. In addition, the Committee encourages CDC to collaborate with stakeholders on an initiative to integrate the Care Considerations into electronic health records to improve care, understand disease outcomes, and model disease progression.

Health Resources and Services Administration

Newborn screening for Duchenne Muscular Dystrophy: The Committee is aware that at its February 2023 meeting, the Advisory Committee on Heritable Disorders in Newborns and Children voted not to advance the nomination of Duchenne Muscular Dystrophy for Evidence-based review for newborn screening. The Committee recognizes the challenges in collecting data for rare diseases, particularly in young children. Duchenne muscular dystrophy typically isn't diagnosed until age five despite the efforts of CDC and others to move the age of diagnosis earlier. As a result, it is difficult to meet the newborn screening criteria despite strong potential for benefit. The Committee recommends expediting consideration of Duchenne Muscular Dystrophy for the Recommended Uniform Screening Panel for newborn screening, incorporating the patient community voice in the ACHDNC review, and developing appropriate flexible criteria.

National Institutes of Health

OFFICE OF THE DIRECTOR
Duchenne and Becker Muscular Dystrophy – The Committee supports the research conducted by the Wellstone Muscular Dystrophy Research Network Centers of Excellence program established in 2003. The Committee directs NIH to provide a report to Congress and the public on the key scientific accomplishments of the Centers to date and their current activities. The NIH also should use this information to update its website content regarding the program.

Food and Drug Administration

OFFICE OF THE COMMISSIONER
Center for Drugs Evaluation and Research
Patient Experience data— The Committee supports the development of patient experience data to inform clinical research design and regulatory reviews under the patient-focused drug development process. Robust patient perspective insights have been generated by the Duchenne Muscular Dystrophy and other patient communities to ensure FDA has the benefit of this information for critical decisions including on potential gene therapies for this serious condition. The Committee encourages the FDA to make every effort to incorporate all relevant patient experience data, including from patient advocacy organizations, across its regulatory obligations.

Department of Defense (DOD)

Congressionally Directed Medical Research Program (CDMRP) Duchenne Muscular Dystrophy Research Funding: $15M (increase from FY23 enacted funding of $10M)

Sincerely,

DORIS MATSUI
Member of Congress

TROY BALDERSON
Member of Congress

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 is less severe than Duchenne, occurs when dystrophin is manufactured, 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".

PPMD's MISSION

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.

OUR IMPACT

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.

1012 14th Street NW Suite 500 Washington, DC 20005
t 800.714.5437 • 201.944.9985 **f** 201.944.9987 **e** info@ParentProjectMD.org • ParentProjectMD.org

Ensure Patient Perspectives Are Included in FDA Benefit-Risk Assessments: Cosponsor the S. 526/H.R. 1092 the Better Empowerment Now to Enhance Framework and Improve Treatments (BENEFIT) Act

Overview

Congress and the Food and Drug Administration (FDA) have made considerable progress in driving forward policies and procedures to ensure the patient perspective is considered by FDA reviewers evaluating candidate drugs and other medical products. As a result of numerous provisions of both the Prescription Drug User Fee Act (PDUFA) authorization of 2012 (known as FDASIA) and the 21st Century Cures Act passed into law in 2016, the FDA now has programs and policies in place to evaluate the benefits and risks of potential therapies and to gather and assess the patient perspectives.

But while much progress has been made, some significant gaps remain. One such gap is the lack of a requirement in law that the FDA include patient experience or patient-focused drug development (PFDD) data as part of its risk-benefit framework. Examples of patient experience data include:

- *Patient reported outcomes* (how a drug impacts activities of daily living ie: whether they can feed themselves, be independent etc.)
- *Patient testimonials* (qualitative data/patient stories of "living with")
- *Patient preference data* (how much risk patients are willing to take)
- *Natural History Data* (the natural progression of the disease without intervention)

The agency's signature tool for evaluating risk-benefit of a drug does not currently explicitly include data from the patient perspective that could be critical to informing the agency's evaluation and, ultimately, decision on whether or not to approve a product.

The BENEFIT Act

To address this gap, Senators Roger Wicker (R-MS) and Amy Klobuchar (D-MN) and Representatives Doris Matsui (D-CA) and Brad Wenstrup (R-OH) have introduced **S. 526/H.R. 1092** the Better Empowerment Now to Enhance Framework and Improve Treatments (BENEFIT) Act.

Currently, FDA indicates whether it receives submitted patient experience data – including information developed by a product sponsor or a third party such as a patient advocacy organization or academic institution – but not whether or how it was used in the review process. **This legislation will amend the Food, Drug and Cosmetic Act (FDCA) to require that FDA include in the risk-benefit framework a description of how submitted patient experience data and information were considered.** This action will enhance transparency and accountability, sending an important signal to all stakeholders that patient experience data will be incorporated into the agency's review process, encouraging such entities to continue developing and refining scientifically rigorous and meaningful tools and data.

Conclusion

The nascent field of patient engagement in drug development continues to flourish thanks to a continued interest and focus by Congress. The BENEFIT Act will continue this evolution by filling a sizeable gap by ensuring such data is fully considered as part of the FDA's risk-benefit assessment. **Advance patient engagement by cosponsoring the BENEFIT Act today.**

Senate: Support S. 526 the BENEFIT Act (Wicker-Klobuchar)
Contact: sally_thompson@wicker.senate.gov or Ruth_McDonald@klobuchar.senate.gov
House: Support H.R. 1092 the BENEFIT Act (Matsui-Wenstrup)
Contact jackie.weinrich@mail.house.gov (Matsui) or kelsi.wilson@mail.house.gov (Wenstrup)

~117H4472

.....

(Original Signature of Member)

118TH CONGRESS
1ST SESSION

H. R. 1092

To strengthen the use of patient-experience data within the benefit-risk framework for approval of new drugs.

IN THE HOUSE OF REPRESENTATIVES

Ms. MATSUI introduced the following bill; which was referred to the Committee on _____

A BILL

To strengthen the use of patient-experience data within the benefit-risk framework for approval of new drugs.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE.

This Act may be cited as the "Better Empowerment Now to Enhance Framework and Improve Treatments Act of 2023" or the "BENEFIT Act of 2023".

SEC. 2. STRENGTHENING THE USE OF PATIENT-EXPERIENCE DATA WITHIN RISK-BENEFIT FRAMEWORK.

Section 569C of the Federal Food, Drug, and Cosmetic Act (21 U.S.C. 360bbb–8c) is amended—

(1) in subsection (a)(1)—

(A) in subparagraph (A), by striking ''; and'' and inserting a semicolon;

(B) in subparagraph (B), by striking the period and inserting ''; and''; and

(C) by adding at the end the following:

''(C) as part of the risk-benefit assessment framework in the new drug approval process described in section 505(d), considering patient experience data submitted by the medical product sponsor or another party.''; and

(2) in subsection (b)(1), by inserting '', including a description of how such data and information were considered in the risk-benefit assessment described in section 505(d)'' before the period at the end.

The Honorable Roger Wicker
U.S. Senate
555 Dirksen Senate Office Building
Washington, DC 20510

The Honorable Doris Matsui
House of Representatives
2311 Rayburn House Office Building
Washington, DC 20515

The Honorable Amy Klobuchar
U.S. Senate
425 Dirksen Senate Building
Washington, DC 20510

The Honorable Brad Wenstrup
House of Representatives
2419 Rayburn House Office Building
Washington, DC 20515

RE: Support for the BENEFIT Act of 2023

Dear Senators Wicker and Klobuchar and Representatives Matsui and Wenstrup:

Thank you for your tireless efforts to encourage development of and expand access to treatments and cures for patients, including those with rare diseases. On behalf of the undersigned patient advocacy organizations, we write in strong support of your legislation, the Better Empowerment Now to Enhance Framework and Improve Treatments (BENEFIT) Act of 2023.

As you know, the 21st Century Cures Act (P.L. 114-255) includes sections 3001 and 3002, the Patient-Focused Impact Assessment (PFIA), which has accelerated the field of patient-focused drug development (PFDD). FDA now has a number of programs and policies in place to gather and assess patient perspectives within the regulatory review process, and patient advocacy organizations have been deeply engaged with the FDA over the past several years to develop PFDD tools that produce scientifically valid patient experience information. Tremendous progress has been made over the past decade since the fifth Prescription Drug User Fee Act (PDUFA) was authorized, including with PFIA and other provisions of 21st Century Cures. Now is the time to take the next step in moving patient perspectives and experience forward by enacting the BENEFIT Act.

The BENEFIT Act would require FDA to include in the benefit-risk assessment framework of a new drug application how patient experience data was considered in the review process. Currently, FDA includes patient experience data in reviews, but does not indicate how such data impacted the drug approval. Providing this information to the public, and patient communities making significant investments in developing PFDD, builds on transparency from PFIA and will accelerate PFDD strategies more broadly.

The field of patient engagement in drug development continues to flourish thanks to the continued interest and focus by Congress. The BENEFIT Act will build upon this foundation and fill a gap by appropriately disclosing how this data is considered as part of FDA review of new therapies. The BENEFIT Act initially passed the Senate in 2017 but further action was deferred as the 21st Century Cures was being implemented.

Now is the time to take this critical step in building the PFDD environment by passing the BENEFIT Act. The Cures 2.0 Act recognizes this as well by including a parallel provision to the BENEFIT Act. Thank you again for your leadership and we look forward to working with you to enact this legislation this Congress.

Sincerely,

AliveAndKickn
Alpha-1 Foundation
Alport Syndrome Foundation
ALS Association
American Brain Coalition
American Kidney Fund
Ara Parseghian Medical Research Fund
Barth Syndrome Foundation
Beyond Celiac
Coalition Duchenne
Congenital Hyperinsulinism International
CSNK2A1 Foundation
Cure CMD
Cure HHT
Cure Sanfilippo Foundation
Cure SMA
CureDuchenne
CureSHANK
Dravet Syndrome Foundation
EveryLife Foundation for Rare Diseases
FND Hope
FORCE: Facing Our Risk of Cancer Empowered
Foundation for Angelman Syndrome Therapeutics (FAST)
Foundation for Prader-Willi Research
Genetic Alliance
Hermansky-Pudlak Syndrome Network
Hope For Marian
International Pemphigus Pemphigoid Foundation
International WAGR Syndrome Association, IWSA
Jett Foundation
Kindness Over Muscular Dystrophy
Klippel-Trenaunay (K-T) Support Group
Little Hercules Foundation
Lupus Foundation of America
MLD Foundation
Mucolipidosis Type IV
National Ataxia Foundation
National Health Council

National Kidney Foundation
National MPS Society
National MS Society
NBIA Disorders Association
Organic Acidemia Association
Parent Project Muscular Dystrophy
Phelan-McDermid Syndrome Foundation
PXE International
RASopathies Network
RUNX1 Research Program
Ryan’s Quest
Sophie's Neighborhood
Stickler Involved People
Sudden Arrhythmia Death Syndromes (SADS) Foundation
Susan G. Komen
SYNGAP1 Foundation
The Global Foundation for Peroxisomal Disorders
TSC Alliance
United Mitochondrial Disease Foundation
Usher 1F Collaborative
WISKOTT ALDRICH FOUNDATION
Zack Heger Foundation

What we are trying to accomplish

Current matrix

Proposed change

Figure 1: FDA Benefit-Risk Framework

Decision Factor	Evidence and Uncertainties	Conclusions and Reasons
Analysis of Condition		
Current Treatment Options		
Benefit		
Risk		
Risk Management		
Benefit-Risk Summary Assessment		

New section

Decision Factor	Evidence and Uncertainties	Conclusions and Reasons
Analysis of Condition		
Current Treatment Options		
Patient Experience Data		
Benefit		
Risk		
Risk Management		
Benefit-Risk Summary Assessment		