ADVOCACY ASKS



Support Fragile X research funding for Fiscal Year 2023.

Between the NIH, the CDC, and the DOD, the Federal government invests over \$40M/year in Fragile X research. This varies per year, depending on the research proposals funded following the peer review process. Additionally, the CDC's Fragile X program supports awareness among professionals, published consensus guidelines, and the FORWARD project, which collects data on individuals with Fragile X across the lifespan and makes it available to researchers. You can support this in three ways:

- Senate: Request that Fragile X be included as an authorized research area for the DOD's Peer Reviewed Medical Research Program. The Appropriations Committee authorizes the medical conditions, disorders, and diseases the program may research through report language each year. Which research is funded is decided by a peer review process. Last year's letter is attached. We will send you a copy of the FY23 letter when it is being circulated.
- Request support for Fragile X at the NIH and the CDC. The CDC has an annual line item in their budget request for \$2M for Fragile X, and the NIH funds projects as they are peer reviewed and, additionally, funds three Fragile X research centers. Rep. Joe Courtney (Maria Costigan at maria.costigan@mail.house.gov) and Rep. Chris Smith (Mary Vigil at MV.Vigil@mail.house.gov) are leading a letter. Contact either staff member for the latest draft and to cosign.

Contact Linda Sorensen with the National Fragile X Foundation, linda@fragilex.org, for suggested report language and/or to complete any appropriations request forms.

Cosponsor the STAT Act (H.R.1730/S.670).

There are over 7,000 known rare disorders and diseases, like Fragile X, and 93% have no FDA-approved treatment. The Speeding Therapy Access Today (STAT) Act seeks to enact policy reforms at the FDA to accelerate development of therapies and facilitate patient access to them. It authorizes a Rare Disease Center of Excellence and Advisory Committee, funds the development of best practices and research to support the approval of therapies to treat very small populations, and ensures public and private payer coverage align with science through a Rare Disease Therapy Access Program.

Contact any of the following staff to cosponsor or for more information: Sen. Klobuchar, ruth_mcdonald@klobuchar.senate.gov; Sen. Wicker, kirby_miller@wicker.senate.gov; Rep. Bilirakis, chris.jones@mail.house.gov; Rep. Butterfield, caitlin.vansant@mail.house.gov.

Support expanded opportunities for individuals with disabilities to participate in the workforce.

Work, even just a few hours a week, can bring joy and meaning to an individual with an intellectual/developmental disability. While there is no one single solution, there are some sound proposals. We ask that you support legislation to open doors for individuals to work, such as:

- ABLE Employment Flexibility Act (H.R. 4672). Allows employers to match contributions to an employee's ABLE account in the same manner they match 401(k)s.
- **Disability Employment Incentive Act (H.R. 3765/S. 630).** Expands tax credits and deductions for employers who hire and retain employees with disabilities.
- Work Without Worry Act (H.R. 4003/S. 2108). Ensures that if a dependent, disabled adult can work without worrying they will lose their Disabled Adult Child benefit.

Join the Congressional Fragile X Caucus (House only).

Contact Maria Costigan (maria.costigan@mail.house.gov) in Rep. Joe Courtney's office or Mary Vigil (MV.Vigil@mail.house.gov) in Rep. Chris Smith's office. The Caucus does not require any financial commitment or agreeing to any policy positions.

FEDERAL INVESTMENTS IN FRAGILE X-ASSOCIATED DISORDERS



We are incredibly thankful for all the federal government has done to support research in Fragile X-associated disorders! Without this investment, we would not be able to make strides towards effective treatments for Fragile X. Our advocates have been able to secure over \$430M+ in research funding over the past 20 years. These are just some of the amazing things federal investments are doing.

CENTERS FOR DISEASE CONTROL AND PREVENTION (CDC)

- The CDC funds the Fragile X Registry With Accessible Research Database (FORWARD), the largest resource of health, clinical, and social support information on people with Fragile X syndrome (FXS) in the United States.
- FORWARD includes over 9 years of longitudinal data accessible to researchers. 24 publications have been generated from FORWARD data with more to be released soon. Recommendations from many of these publications have been important for informing health management, identifying risk groups, gaps in care, and needs for intervention to improve quality of life for individuals with FXS.

NATIONAL INSTITUTES OF HEALTH (NIH)

- NIH updated their Strategic Plan for Research on *FMR1*-Associated Conditions in 2019. This plan shared goals for specific *FMR1*-associated conditions, including cross-disciplinarity goals that encourage innovative, impactful research in Fragile X
- NIH supports the Centers for Collaborative Research in Fragile X Program, with the common goal "of facilitating the translation of basic research findings from bench to bedside and bedside to community." This grant supports research to improve the diagnosis and treatment of Fragile X syndrome (FXS) and its related conditions, aiding premier FX Centers in increasing their impact in line with the NIH Strategic Plan for Research on *FMRI*-Associated Conditions. The current awardees are Emory University, Baylor College of Medicine, and Cincinnati Children's Hospital Medical Center. The centers will receive \$25 million over the next 5 years.
- NIH funds multiple Career Development (K) awards, some of which have gone to up and coming professionals who are completing innovative projects in Fragile X. This pipeline of and for new researchers is critical to advancements for Fragile X.

DEPARTMENT OF DEFENSE (DOD)

- Congress continues to recognize Fragile X as an authorized medical research area.
- The DOD Peer Reviewed Research Program (PRMRP) has funded around \$28M in research projects related to Fragile X syndrome. Projects funded for FY 2022 haven't been released yet.

FOOD AND DRUG ADMINISTRATION (FDA)

- FDA has granted Fast Track and Orphan Drug Designation to various drug developers in Fragile X. This suggests FDA understands the need for treatments for Fragile X and supports the quest to find treatment(s).
- NFXF hosted an Externally-led Patient-Focused Drug Development Meeting for Fragile X syndrome in 2021. The report is being released in March. This meeting is an official FDA meeting that allows patients and caregivers to give feedback on what treatments they would find most helpful. The goal is to ensure drug development matches what the patients and caregivers want. The FDA offers a limited number of PFDD meetings, so we are fortunate to have one. <u>fragilex.org/pfdd</u>



United States Senate

2021 letter as example.

WASHINGTON, DC 20510

June 23, 2021

The Honorable Jon Tester Chair Senate Appropriations Committee Subcommittee on Defense Washington DC 20510 The Honorable Richard Shelby Ranking Member Senate Appropriations Committee Subcommittee on Defense Washington DC 20510

Dear Chair Tester and Ranking Member Shelby:

As Senators committed to improving the health of children and adults living with intellectual disabilities in the United States, we respectfully request your continued commitment to sustaining federal investments in biomedical research focused on the treatment and cure of Fragile X syndrome and its related conditions.

Mutations of the Fragile X gene result in behavioral, developmental, cognitive, reproductive, and potentially life-ending neurodegenerative conditions across generations in families and impact affected individuals from cradle to grave. Fragile X syndrome and associated disorders result from a single-gene mutation, which is the most common, known inherited cause of intellectual disabilities and autism. In fact, research has shown that the Fragile X protein regulates nearly one half of the genes suspected of causing autism. Up to 100,000 Americans have Fragile X syndrome, and up to 1,500,000 Americans have a variation of the Fragile X mutation and as a result either have, or are at risk for developing, one of the conditions associated with Fragile X and passing the gene mutation to their children. The known premutation issues are Fragile X-associated primary ovarian insufficiency, which causes infertility and early menopause.

The Committee's previous support of Fragile X as one of the research areas authorized for the DOD's Peer Reviewed Medical Research Program funded some important research and has the potential to ease the burden of Fragile X and other intellectual and developmental disabilities on our military families. Military families are affected substantially by the financial and emotional costs of raising a child with intellectual and developmental disabilities, including Fragile X syndrome. This impact extends to the performance and readiness of service members and their units. Strides are being made towards effective treatments for Fragile X syndrome and other associated disorders while moving towards a cure. These treatments will help ease the burden on military families.

We are requesting that Fragile X be included as an authorized research area for the DOD's Peer Reviewed Medical Research Program for Fiscal Year 2022. While we understand the challenges the Committee faces in prioritizing requests, Fragile X has a significant impact on military families across generations in every state and district. The potential for effective treatments is within reach. We believe continued support for Fragile X research is imperative. The DOD's research has been a significant contributor over the past decade, and we hope it will continue be in the future.

We look forward to working with the Subcommittee on this important issue. Thank you for your consideration.

Sincerely,

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Debbie Stabenow United States Senator

Bill Cassidy, M.D. Bill Cassidy, M.D.

Cory A. Booker United States Senator

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Gary C. Peters

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Ben Ray Lujan United States Senator

Chris Van Hollen United States Senator

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Elizabeth Warren United States Senator

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Jacky Rosen United States Senator

Mark Kelly United States Senator

Roger Wicker

Kinsten Gillibrand

Kirsten Gillibrand United States Senator

Kyrsten Sinema United States Senator

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Richard Blumenthal United States Senator

Shurrod Brown

Sherrod Brown United States Senator

Alex Padilla

Congress of the United States Washington, DC 20515

April XX, 2022

The Honorable Rosa DeLauro Chairwoman Subcommittee on Labor, HHS, Education House Committee on Appropriations Washington, DC 20515 The Honorable Tom Cole Ranking Member Subcommittee on Labor, HHS, Education House Committee on Appropriations Washington, DC 20515

Dear Chairwoman DeLauro and Ranking Member Cole:

As Members of Congress committed to improving the health of children and adults living with intellectual disabilities in the US, we respectfully request your continued commitment to sustaining federal investments in biomedical research and public health initiatives focused on the treatment and cure of Fragile X and its related conditions.

Mutations of the Fragile X (FX) gene result in behavioral, developmental, cognitive, reproductive, and potentially life-ending neurodegenerative conditions across generations and impact affected individuals from cradle to grave. Fragile X syndrome and associated conditions result from a single-gene mutation, which is the most common, known inherited cause of intellectual disabilities and autism. In fact, research has shown that the Fragile X protein regulates nearly one half of the genes suspected of causing autism. Up to 100,000 Americans have Fragile X syndrome, and up to 1,500,000 Americans have a variation of the Fragile X mutation and as a result either have, or are at risk for, developing one of the conditions associated with Fragile X.

The Committee's previous support for the important work underway at the National Institutes of Health (NIH) and Centers for Disease Control & Prevention (CDC) is advancing research towards more effective treatments and a cure for Fragile X. For instance, the CDC funds the FORWARD project, a natural history study. FORWARD is a longitudinal database that now includes 9.5 years of historical data of individuals with Fragile X and additional data on premutation carriers and other family members. This data gives researchers a view of Fragile X over the lifetime and across generations. These are resources that are not available anywhere else and are invaluable tools for educators and clinicians.

The NIH supports Fragile X research across multiple Institutes, with the primary one being the National Institute of Child Health and Human Development. This includes three national Fragile X research centers that focus on stimulating multi-disciplinary, multi-institutional research with a goal of translating basic research into treatments. We are seeing promising treatments for many of the behaviors associated with Fragile X syndrome – including several currently in various phases of clinical trials – and research that will also inform treatments and better understanding of autism. Fragile X is the most common, known, single gene cause of autism.

To ensure the rapid translation of ongoing research into near term targeted treatments, we must continue these federal investments in the Fiscal Year 2023 Labor, Health and Human Services, and Education Appropriations bill. Specifically, we respectfully request your support for directives to:

- Expand the base of researchers and clinicians who are familiar with and trained in the Fragile X-associated disorders and promoting collaboration between basic scientists and clinicians to enable researchers to better understand phenotypes, document variations in how the disorder presents itself, identify potential biomarkers and outcome measures, and develop new interventions.
- Maintain dedicated support for CDC's national Fragile X public health program.

To this end, we respectfully request the inclusion of the following report language regarding Fragile X Syndrome:

<u>CDC</u>

Fragile X and Fragile X-Associated Disorders. -- The Committee commends CDC's efforts to identify and define the population impacted by fragile X (FX) and all conditions associated with the gene mutation with the goal of understanding the public health impact of these conditions. To help this effort, the Committee urges the National Center on Birth Defects and Developmental Disabilities (NCBDDD) to support additional strategies to promote earlier identification of children with FX, such as voluntary newborn screening. The Committee also recommends the NCBDDD work to ensure underserved populations with FX conditions are being properly diagnosed and are aware of available medical care and support.

<u>NIH</u>

Fragile X.-- The Committee notes the importance of expanding the base of researchers and clinicians who are familiar with and trained in the Fragile X-associated disorders and promoting collaboration between basic scientists and clinicians to enable researchers to better understand phenotypes, document variations in how the disorder presents itself, identify potential biomarkers and outcome measures, and develop new interventions. The Committee also commends the NIH for recognizing the ethical, legal, and social issues in premutation screening and testing and encourages to NIH to look at existing pilot studies that are looking at innovative ways to screen newborns, and to coordinate efforts and research with the CDC as they look at screening solutions for FMR1-related conditions.

While we understand the challenges the Committee faces in prioritizing requests, Fragile X has a significant impact on families across generations, on individuals throughout their lives, and on communities in every state and district. The potential for effective treatments is within reach, and continued support for Fragile X research and public health education is imperative. We look forward to working with the Subcommittee on this important issue. Thank you for your time and consideration.

Sincerely,

CHRISOTPHER H. SMITH Co-Chair, Fragile X Caucus JOE COURTNEY Co-Chair, Fragile X Caucus