



## **Sustaining NIH Research on Fragile X-associated Disorders-Justification**

*"Fragile X syndrome is an excellent model for autism. Today, just a relatively short period of time after the gene for Fragile X was isolated, clinical trials are proceeding for drugs that seek to correct the central neurochemical defect underlying the condition. I've seen some early results from these trials, and they look quite encouraging to me.*

***"Francis S. Collins, M.D., Ph.D., Director, NIH***

*"Until we can better understand the biological bases of these different forms of autism it will make it difficult to understand any of them". **Dr. Allen Guttmacher, Director of NICHD***

### **FY2018 Appropriations Request: National Institutes of Health**

**The NFXF urges Congress to sustain existing federal investment in biomedical research managed by the National Institutes of Health (NIH) and to direct NIH to maintain its investment in Fragile X research at not less than current levels.** *Each institute with Fragile x and autism portfolios should explore ways to create greater efficiency and synergy among Fragile X and autism research tracks to accelerate translational research toward a better understanding of both conditions and to shorten the time to bring effective treatments for both to market. The NIH Research Plan on Fragile X Syndrome and Associated Disorders revised in 2012 should be published and fully implemented with prioritization of the updated goals and objectives.*

**Previous NIH Support for Fragile X:** The Fragile X Research Breakthrough Act of 2000, a provision of the Children's Health Act (Title II), authorized funding for the establishment of at least three Fragile X Research Centers. In response, the National Institute of Child Health and Human Development (NICHD) funded three Centers in March 2003 in the amount of \$3.75 million "... to conduct research to improve the diagnosis and treatment of, and to find a cure for Fragile X-associated Disorders." These Centers have proven critically important to the development of effective therapeutic interventions as well as our epidemiological understanding of the condition. As a result of various public and private research efforts, effective treatment for Fragile X-associated Disorders is close to becoming a reality-the condition has been reversed in multiple animal models and new drugs are currently in human trials. However, even as the potential for treatments and a cure increase, federal funding for Fragile X-associated Disorders (FXD) research has not always kept up. For many years, federal funding remained stagnant and even dropped from FY 2005 (\$22 million) to FY 2006 (\$20 million). In FY 2007, though, thanks to continued pressure from Congress, the NIH devoted \$27 million to Fragile X research and projects, which steadily increased to \$32 million in FY2009. In FY 2010, 2011 and 2012 this amount retreated to 29 million. Now, *given the inextricable connection between the FX protein and autism, the prospect of first-ever targeted treatments for both conditions and mindful of current budgetary constraints, the NFXF urges the Office of the Director and his counterparts at each institute with Fragile X and autism portfolios to explore ways to create greater efficiency and synergy among the Fragile X and autism research tracks to accelerate translational research toward a better understanding of both conditions and to shorten the time to bring effective treatments for both conditions to market. Even greater efficiencies are within reach if NIH partners with the CDC, the Fragile X Clinical & Research Consortium and the private sector*

to get the most out of available resources by working together collaboratively on research questions and priorities of mutual interest and a unified database and registry of patients.

**Rationale/Justification for Request** In 2012 fourteen of the NIH's individual Institutes and other research entities supported nearly 200 diverse grants exploring basic science, the connection between FX and autism and the quest for targeted treatments for the closely related conditions of FXS and autism. Given this array of NIH-based Fragile X efforts, the soon to be published NIH Fragile X Syndrome and Associated Disorders Research Plan, the Therapeutics for Rare and Neglected Diseases (TRND) and National Center for Advancing Translational Sciences (NCATS) programs, and the undeniable reality that targeted drugs capable of treating the underlying neuro-biological mechanism in FXS and autism are moving toward FDA approval, a renewed effort to achieve the goals set out in the updated Research Plan, as well as, increased NIH support for the Fragile X Clinical & Research Consortium must be undertaken to maximize resources dedicated to FXD. *Moreover, given the newly discovered connections between the FX protein and autism and mindful of current budgetary constraints, the NFXF urges all NIH institutes with Fragile X and autism portfolios to explore ways to create greater efficiency and synergy among these two research tracks to accelerate translational research toward a better understanding of both conditions and to shorten the time to bring effective treatments for both conditions to market.*

**Action Requested** Congressional leaders are asked to support the NFXF's FY 2018 appropriations request and suggested appropriations report language by co-signing a group letter to the Appropriations Committee leadership. Appropriations Committee members are respectfully encouraged to include this appropriations request among their FY 2018 funding priorities.

## **Supporting CDC's National Fragile X Public Health Initiative-Justification**

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*"Fragile X Syndrome itself has much broader ramifications than we originally thought and the link with autism is a very important and fundamental discovery that we think will have very important consequences. Fragile X Syndrome has much broader public health implications than we ever thought"* **Don Bailey, Ph.D., Distinguished Fellow, RTI International**

### **FY2018 Appropriations Request: Centers for Disease Control and Prevention**

**The National Fragile X Foundation (NFXF) is seeking level funding at the FY 2012-13 level to support the dedicated National Fragile X Public Health Initiative within National Center on Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention (CDC).** This funding is requested to support existing and new activities, including the further expansion of the epidemiological and public health data collection activities of the Fragile X Clinical and Research Consortium (FXCRC), the further development and coordination of the consortium's database and registry with other overlapping efforts, a targeted outreach to typically underserved populations, and further epidemiological research, surveillance, screening and the promotion of early interventions and supports to address the significant public health impact of Fragile X-associated Disorders (FXD).

### **Previous Federal Support for Fragile X**

Since 2003, Congress has urged the CDC to create and grow a Fragile X public health program to conduct surveillance and epidemiological research on FXD as well as provide education to health care professionals and the public on FXD and other developmental disabilities. Over the last ten fiscal years, Congress has provided nearly 14 million dollars in total funding to address gaps in FXD research, screening, treatment, and awareness. As a result, the CDC has funded multiple grants in the areas of Fragile X Epidemiology and

Screening, Fragile X Cascade Testing and Genetic Counseling, the creation of a Single Gene Disorders Resource Network and in partnership with the NFXF, a Fragile X Clinical and Research Consortium (FXCRC). The FXCRC has now grown to 26 clinics across the country collecting critically needed data and providing specialty services and referrals to individuals with FXD. A greater effort is needed to make these services available to typically underserved populations and the NFXF can continue to partner with the CDC to accomplish this and believes CDC should invest its dedicated Fragile X resources in improving access to services for the underserved. The community is pleased with the CDC's progress thus far in implementing a public health program on FXD, but wants to assure that these funds are used for the purposes that Congress intended. In FY 2012 and again in 2013 a proposal to consolidate CDC NCBDDD funding accounts (including FX) was rejected. The NFXF urges Congress to continue the dedicated funding to address the unique and far reaching public health impacts of FXD.

#### **Rationale/Justification for Request**

Although it is the leading known cause of inherited cognitive impairment and the most common known genetic cause of autism, and although more than half of all individuals with FXS are on the autism spectrum, FXS is not always included in current federal developmental screening or surveillance activities. In order to increase our understanding of Fragile X, the CDC national Fragile X public health initiative should focus its efforts on increasing epidemiological research, surveillance, and screening efforts, with particular attention towards collecting epidemiological data on the incidence and prevalence of FXD. The NFXF is pleased that upon expiration of an initial three year grant to support the FXCRC that a second grant was awarded in 2011 to assist the 26 U.S. FX clinics collect epidemiological data to better define and address the far reaching public health impacts of FXD and provide a database and registry through which ongoing clinical trials of targeted new drug therapies can be advanced. The NFXF welcomes new activities by the Agency to address this disorder through initiatives such as the early identification of individuals with a FXD; genetic counseling and a database of best practices. Each of these activities will have a profound impact on the Fragile X community, and could serve as a model for a more expanded focus on the appropriate screening, diagnosis, counseling and treatment of an array of developmental disabilities. Moreover, given the newly discovered connections between the FX protein and autism and mindful of current budgetary constraints, the NFXF urges the NCBDDD Divisions with Fragile X and autism portfolios to explore ways to create greater efficiency and synergy among these two public health tracks to accelerate epidemiological data gathering toward a better understanding of both conditions and to shorten the time to bring effective treatments for both conditions to market.

#### **Action Requested**

**Congressional leaders are asked to support the NFXF's FY 2014 appropriations request for level and dedicated funding and adoption of the suggested appropriations report language by co-signing a group letter to the Appropriations Committee. Committee members are encouraged to include this appropriations request among their FY 2014 funding priorities.**