Thank you for your participation in FORWARD, your involvement is essential to what we discover about FXS!

Due to your continued support and contribution, FORWARD is now the largest resource for clinical and demographic data of the fragile X syndrome (FXS) population in the United States. There are over 2,700 individuals enrolled into the registry (participants with FXS and their family members), over 1,000 of those participants with FXS have also enrolled into the longitudinal database.

The mission of FORWARD is, and will continue to be, the pursuit of knowledge to improve the care and quality of life for those affected by FXS. Although many scientific advancements have been made to better understand the genetics of FXS, we are only just beginning to understand how FXS affects individuals and their families.

This study collects clinical, health and quality-of-life information about individuals with FXS. In addition, pertinent information such as support and basic demographic data is gathered from family members unaffected or with a premutation via a one-time registration form. Participants with FXS are enrolled in a longitudinal study and subsequently, annual follow-up information is collected through standardized forms to monitor and document changing data. In this way, we are able to observe how FXS presents itself over a person’s lifetime in order to gain a better understanding of this condition. Attaining and documenting data, both from the clinician and parent/caregiver perspective, will lend itself to better care and services for those affected with FXS. Ultimately, it is our hope to improve their lives and the lives of their loved ones.

FORWARD also facilitates FXS research by connecting interested researchers to Fragile X clinics. In so doing, the researcher might find potential patient matches for their particular study. This service is not only expeditious for the researcher, but can also help clinics and participants become aware of available FXS studies. This benefits both researchers and the patient community.

With more knowledge, it is our hope to pave a kinder and gentler path for future generations of those with FXS.
FORWARD represents the largest data resource with both clinical and parent reported data on the FXS population in the United States. The intent is that this data can be used to further our understanding of FXS by providing a more robust description of the FXS population, developing an improved understanding of the natural history of FXS across the human lifespan, describing the impact of co-occurring conditions, and describing the impact of treatments and interventions on health outcomes and quality of life.

Preliminary estimates of participants with FXS enrolled from 9-7-2012 to 6-30-2016 suggest that:

- 77% were males and 23% were females. (Data from 897 participants total.)
- The vast majority of participants resided with parents or families (95%), with the remaining 5% not residing with family. (Data from 525 participants total.)
- Level of Intellectual Disability (ID) as reported by the clinician; 6% were reported as having no ID, 13% reported with development delay, 9% Borderline ID, 23% Mild ID, 42% Moderate ID, 7% Severe ID. (Data from 781 participants total.)

Numerous small and parent reported studies have shown that a considerable percentage of individuals with FXS also carry a diagnosis of autism spectrum disorder (ASD). Individuals with a dual diagnosis of FXS and ASD were reported to show greater cognitive and behavioral impairment.

The first large-scale study looking at ASD in FXS was conducted using FORWARD data. With this data, we discovered a high percentage of ASD in the FXS population. Findings further showed, greater behavioral impairments as well as disparities between expected and observed behavioral treatment use, of those with FXS and ASD when compared to those with FXS alone.

While looking at medical problems in the largest national cohort of patients with FXS to date, FORWARD is learning more about health issues associated with FXS, including seizures, recurrent ear infections, and GI symptoms (both constipation and loose stools).

In addition to affecting general health, medical problems can contribute to developmental and
behavioral difficulties for those with FXS. For example, ear infections can affect hearing and as a result can contribute to speech delays. Pain and discomfort may manifest as behavioral difficulties and sleep problems.

Understanding the common medical problems with FXS at varying ages becomes important to guide recommendations for medical screenings and interventions. The current FORWARD project is expanding to collect much-needed information on medical problems in adults with FXS, while continuing to collect more information in the pediatric age group.

Do Individuals with FXS Have Sensory Problems and Hyperarousal?
Primary Presenters: Ave M. Lachiewicz MD and Debra Burgess, Duke University Medical Center

It appears that sensory processing problems and hyperarousal are common in individuals with FXS through childhood and early adulthood. This study provides a clearer understanding of specific sensory problems associated with FXS, specifically, it can identify a sensitivity to particular sounds. It does not appear that interventions for sensory based problems are utilized to the degree that the problem seems to warrant. Mental health problems are commonly reported and most participants were on medications. While medical management may be valuable, some mental health problems, like anxiety, may be increased by sensory processing dysfunction and hyperarousal. Some individuals, who are not being treated for sensory processing problems and hyperarousal, may be missing the benefit that would be received from appropriate treatment.

Given the prevalence of sensory and hyperarousal issues, Fragile X clinics may find it helpful to increase their understanding and be aware of these problems. Fragile X clinics may be able to set the direction for how these conditions are addressed.

Toilet Training Milestones in Fragile X Syndrome
Primary Presenter: Elizabeth Berry-Kravis, MD, PhD, Rush University Medical Center

FORWARD data suggests that although girls with FXS in general do fairly well with toilet training, a significant fraction of males with FXS are achieving toileting milestones very late, possibly out of proportion to the level of overall developmental delay. It appears that over 20% of males with FXS are not trained at age 10. This represents a significant chronic burden for families of boys with FXS. The data suggests that toilet training strategies should be targeted for improved intervention strategies in order to accomplish toilet training earlier for FXS boys. It is our hope that this will improve the quality of life for both the boy and his family.

Preliminary analysis suggests that seizures, along with medication used for behavioral problems, are associated with delays in toilet training. Functional skills such as writing, language, cognition, autism status, and global functioning were also preliminarily found to be associated with delays in toilet training. Autism seems to impact training considerably. About 40% of males with combined FXS and autism, compared to approximately 10% of males with FXS alone, were not trained at age 10.
Please remember to complete your child’s yearly FORWARD Follow-up!

In addition to contributing to FORWARD, yearly visits to a Fragile X clinic will give Fragile X experts a better understanding of you and your family. Getting to know your child will help clinicians assess any acute issues. This can be very difficult to identify in a first meeting or when a patient has not been to clinic for some time.

What if you are unable to visit a clinic for your child’s yearly FORWARD Follow-up?

A coordinator at your clinic can send you forms to fill out at home. You can send the completed forms back to your clinic in a pre-paid envelope that they include. You may have the option to complete these forms online. Please contact your clinic for more information about these options.

FORWARD website coming soon - visit FORWARDFX.org for more information!

Visit clinics.fragilex.org for a list of Fragile X clinics.

If you have questions about visiting a clinic, you may contact Jayne Dixon Weber, the NFXF Director of Education & Support at jayne@fragilex.org.

Your contribution makes a difference!

Dr. Nicole Tartaglia,
Children’s Hospital of Colorado

Dr. Craig Erickson,
Cincinnati Children’s Hospital

Being part of FORWARD has been really important for our clinic in many ways. The consensus documents have been very useful as handouts in clinic for families. It has also helped us to have a more systematic way of following patients’ progress over the years. Part of the FORWARD protocol includes yearly evaluation of life skills – and as a doctor it really helps to have this information when seeing patients in clinic to get a broader idea of how things are going in day-to-day life.

In clinic we are often asked what will happen with my child when they get older. With the FORWARD project we are gathering the information we need to be able to let families have a better idea of what to expect. This information is also invaluable to developing best practice guidelines that will inform Fragile X care around the world. We have enjoyed participating in the FORWARD project since its inception and hope to continue with this work for many productive years into the future.