

FRAGILE X INFO SERIES: YOUR VISIT TO A FRAGILE X CLINIC - AN IN-PERSON VISIT



Overview

Fragile X is a group of conditions associated with changes in the Fragile X gene – called FMR1 and located on the X chromosome. The FMR1 gene can undergo changes, when inherited, which affects a pattern of DNA called CGG repeats. Typically, the FMR1 gene has up to 54 CGG repeats, though the range between 45 – 54 repeats, is called the intermediate or gray zone. A premutation carrier has 55–200 CGG repeats, and someone with a full mutation has more than 200 CGG repeats.

When a premutation or full mutation is present, it can result in a Fragile X-associated Disorder (FXD). These include:

- Fragile X syndrome (FXS): An inherited condition affecting intellectual, behavioral, and social development. It occurs in both males and females who have a *full mutation* of the FMR1 gene.
- Fragile X-associated tremor/ataxia syndrome (FXTAS): An adult onset (over 50 years of age) neurological condition, more common and more severe in some male *premutation carriers*. It can cause tremors, memory, and balance issues.
- Fragile X-associated primary ovarian insufficiency (FXPOI): A condition affecting ovarian function that can lead to infertility and early menopause in some female *premutation carriers*.
- Other issues may be present in premutation carriers and researchers continue to study possible associations with these and other health issues in people with Fragile X premutations.

The Fragile X Clinical and Research Consortium (FXCRC)

The Fragile X Clinical and Research Consortium (FXCRC) was created by The National Fragile X Foundation (NFXF) in response to the growing needs of families whose members have one of the Fragile X-associated Disorders (FXD). The FXCRC is composed of over 30 clinics each of which has expertise in Fragile X.

Though all clinics serve individuals with Fragile X syndrome (FXS), many also serve those or have referrals for those with fragile X-associated tremor/ataxia syndrome (FXTAS), fragile X-associated primary ovarian insufficiency (FXPOI), and other premutation carrier conditions.

Though each clinic operates independently, they often collaborate with one another, sharing resources, participating in research projects, and attending meetings of the consortium. New clinics are being added regularly across the U.S., and around the world. For a regularly updated map and clinic information, see: <https://fragilex.org/clinics/>

Where to Begin

Begin by calling the clinic coordinator to discuss your concerns about your child or family member and what you hope to get out of the visit. Ask about the following:

- What has to be done before the visit? Paperwork, etc. See Information Needed below.
- What does a typical visit entail? Some clinics conduct evaluations over two days, which requires an overnight visit. Others may require only several hours.
- In addition to seeing the clinic director, what evaluations are offered and by whom? Examples might include speech, occupational, physical, and behavioral therapy, and genetic counseling.
- What other specialists are available? For example: psychologists, audiologists, neurologists, cardiologists, ophthalmologists, etc.
- Are referrals made to local providers in your area?
- After the evaluation, will the clinic director be available to review and discuss the results? Will each specialist be available, or will the clinic director review the findings of each specialist with you?
- When will a written evaluation be available? Will it consist of separate reports by each clinic specialist or one report? Can you request that the report be sent to your providers, teachers, therapists, etc.?
- Who will be available to interact with your local providers to make sure treatment recommendations are clarified, discussed, and implemented?
- Do they have a visual schedule of the visit you can access?

Also Ask About

- What written or online materials will be available? A list of state and local resources? Handouts or articles on FXD for you to take back to school personnel, doctors, and other providers?
- Will there be a possibility to participate in research?
- What kind of insurance does the clinic accept? Also, check with your insurance to see what your responsibilities will be, financial and other requirements. (If you do not have insurance, ask if there are alternative payment plans and/or opportunities to participate in research trials).
- Do they have recommendations for lodging facilities to meet your needs regarding price, proximity to the clinic, dining opportunities, etc.? Do any of them offer discounts for those making hospital visits?

- What restaurants, grocery stores and other attractions are in the area? (You may want to make a mini vacation of the visit.)
- Is there an NFXF CSN group or local family you could talk to about the area and about your visit to the clinic? (CSN is Community Support Network – volunteer led groups/people providing local support; see: <https://fragilex.org/living-with-fragile-x/community-support/>)
- A special consideration for those with FXTAS: Make sure to inquire about hotel and transportation/wheelchair issues.
- If you need financial assistance to travel to this clinic, ask about the Fly with Me Fund.

Information Needed

Each clinic has an intake form for which you will most likely need the following:

- Information about your immediate and extended family, including diagnoses of any individuals—living or deceased—with developmental delays, behavioral disorders, neurological disorders (including late onset conditions), fertility issues, and any genetic testing that has been done. Bring photos of any family members who may have Fragile X syndrome.
- Information from baby books, scrapbooks, and journals that would note ages of developmental milestones, illnesses, medication (including any reactions), therapeutic interventions, etc.
- Medical and educational records (for those with FXS). This would include genetic testing, pediatric, medical, or psychiatric records, psychological, speech and occupational therapy evaluations, and records from school therapists and teachers.
- If your visit is for FXTAS, bring MRI reports, neurological and/or psychiatric records, and any journal of symptoms and their progression.
- If your visit is for FXPOI, bring records of medications, medical, menstrual/pregnancy history, and any questions related to medical management or reproductive issues/options. The wait to be seen at a clinic varies, but in some instances can be 3-6 months.
- If your visit is for other premutation carrier issues, bring information from other doctors that may be appropriate.

What are you wondering about?

As you prepare for your visit, begin to write down basic questions for the clinic staff that may not have been covered in your initial discussion with the clinic coordinator. Please know—and continue to remind yourself—that no question is silly or strange or unwelcome. If you are wondering about something, it is virtually certain that the very same question has occurred to many other people—and Fragile X professionals have probably heard it. Clinic staff are well-informed, trained, and compassionate, ready to answer (or find out the answer to) any question you may have. So please do ask! Typical questions include:

- Why does my child/family member do_____?
- What can or should I do in these situations?
- What can I expect from this type of therapy or medication?
- Is it wrong to_____?

Don't Forget...

- To bring favorite toys, puzzles, blankets, foods, and other comfort items for your child(ren).
- To ask for special accommodations if you feel they would help. Example: if staying in the waiting room is difficult for your child, perhaps the staff can call you on your cell phone when they are ready to see you.
- To bring books, music, laptop computers, and other enjoyable items for yourself. (Headphones in waiting rooms can come in very handy.)
- To build in some downtime during what can be a highly scheduled visit. If possible, find some time to visit a park, zoo, museum, attend a sporting event, go swimming, or anything else your family likes to do.
- To treat your clinic visit as but one step in your family's long-term education about Fragile X.

Follow Up Questions

- What if you have follow up questions after receiving the report(s).
- Will the doctor answer questions/converse by phone or email following the visit, even months later?
- How often should I expect to make an in-person visit?
- Will the doctor do telehealth visits? See: <https://fragilex.org/blog/telehealth-visits-suggestions-for-parents-on-how-to-prepare/>

About the NFXF

The National Fragile X Foundation (NFXF) was founded in 1984 to support individuals with Fragile X syndrome (FXS), their families, and the professionals who work with them. Today, it is a comprehensive resource not only for FXS, but also for the conditions of Fragile X-associated tremor/ataxia syndrome (FXTAS), Fragile X-associated primary ovarian insufficiency (FXPOI), and other premutation carrier conditions and disorders. The NFXF is dedicated to serving the entire Fragile X community to live their best lives by providing the knowledge, resources, and tools, until, and even after more effective treatments and a cure are achieved. Learn more at <https://fragilex.org/welcome>

If you have questions please reach out to us at treatment@fragilex.org or call (800) 688-8765.

