



NATIONAL FRAGILE X FOUNDATION  
FRAGILE X CLINICAL & RESEARCH CONSORTIUM

Consensus of the Fragile X Clinical &  
Research Consortium

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TRANSITION TO  
ADULT SERVICES FOR  
INDIVIDUALS WITH  
FRAGILE X SYNDROME

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## Introduction

This paper is focused on the transition from adolescence to adulthood. Because the research on this topic is limited, much of the information is anecdotal, meaning it is taken from clinical experience or provided by parents. Formal studies are referenced.

The transition from adolescence to adulthood can be challenging for many reasons. Those with Fragile X syndrome (FXS) who were diagnosed early have most likely benefitted from the Individuals with Disabilities Education Act (IDEA). IDEA provides funding for the support of those individuals in the public education systems affording educational and vocational opportunities.

Because IDEA mandates support through the age of 21, individuals with FXS often qualify for transition programs and receive support beyond graduation from high school. This type of programming is extremely important to developing as much independence as possible through the age of 21. Most states offer 'school to work' programs through their local disability organizations, but the financial support for job coaches and customized employment can be limited. Some individuals, who are more significantly impaired, may qualify for programs to address activities of daily living- all with the outcome of becoming as independent as possible. Parents may be able to access day programs that provide activities for adults with a variety of disabilities who can experience cooking, community resources like the library, concerts, local attractions, and restaurants.

## Services provided through community agencies and how to access them

There are many new issues to address as the person with FXS becomes an adult and the information below should help when planning for that transition.

In general, it is best to search for information on adult issues and services *in the state where the adult resides*. Services can vary widely from state to state and even vary within a state, so it is up to the parents or providers to find what is available and to set up the daily schedule for and/or with the person with FXS.

### *Suggested contacts:*

- The local disability organization. Hopefully, people from this organization have attended the IEP meetings the last few years at the high school. Check in with them to see how long the wait lists are, and make sure the young adult is on the correct wait list for the services that he or she will need.

- The local NFXF Community Support Network (CSN) group. Talk with parents who have adult children. <https://fragilex.org/living-with-fragile-x/community-support/>
- The Arc is the world's largest community-based organization of and for people with intellectual and developmental disabilities (I/DD). Search "Arc of (your city, county or state)" to find a local chapter to find resources for activities, advocacy, employment, and community supports.

### **Supplemental Security Income (SSI):**

Applying for SSI can take several months, involves providing documentation, and may include a medical exam, so it is good to keep that in mind when the young adult turns 18 years of age. It is important to note that it is never too late to apply for SSI. The process can be initiated by a family member, support person, or by the individual directly, at any time. For more information on SSI, visit <https://www.ssa.gov/ssi/> or call or visit the local Social Security Administration office.

### **Medicaid Waivers:**

Medicaid waivers help provide services so people can stay in their homes and in their community. The services and the wait lists vary widely from state to state, but it is important that the parents get their child on the wait list for services as early as they can. If they decide to move to a different state once the child is an adult, they may need to reapply and possibly be placed on a waiting list.

See: <http://medicaidwaiver.org/>

### **The ABLE Act:**

Eligibility for SSI and Medicaid requires that an individual's assets not exceed \$2000. The ABLE Act recognizes that there are extra costs of living with a disability and allows eligible individuals and their families to establish ABLE savings accounts that will largely not affect their eligibility for SSI, Medicaid and other public benefits.

The ABLE National Resource Center website provides the latest, most up-to-date information about ABLE accounts in every state. <https://www.ablenrc.org/> You can also sign up to receive their newsletter.

### **Guardianship and other Alternatives:**

Because the rules around guardianship and alternatives for decision-making vary from state to state, we recommend that parents research the laws and options in the state where they reside. Google: "guardianship in (your state)" or "guardianship alternatives in (your state)".

Some states require that the parents use an attorney in the process, but some don't. The local Arc is a good source of information on this topic and often offers classes where parents can learn more about this process in their state. It is important to review what guardianship means in their state and to look at all the options that may be available to the parents and their adult with FXS.

### **DMV-issued Photo Identification (ID):**

It is important to have a legal photo ID for the adult with FXS. Contact the local Division of Motor Vehicles (DMV) to find what legal papers are needed in order to get this DMV-issued ID. As an adult, it is important to have this legal ID for everything from airplane travel to going to the doctor's office.

For more information, Google: "(your state) Identification Card information"

### **Register for Selective Service:**

Almost all men ages 18-25 who are U.S. citizens or are immigrants living in the U.S., are required to register with The Selective Service. U.S. law calls for citizens to register within 30 days of turning 18. People with disabilities are required to register unless the person is confined to his home, whether his own or someone else's (including group homes) or resides in a hospital, nursing home, long-term care facility.

For more information: <https://www.usa.gov/selective-service>

### **Register to Vote:**

Many people with FXS enjoy learning about politics and exercising their right to vote. While the legal voting age in the U.S. is 18, voter registration rules are different in every state.

For more information: <https://www.usa.gov/voter-registration-age-requirements>

If the person with FXS is over 18 years of age when the DMV issues the photo ID, he or she may be offered the option to register to vote at that time.

For more information: <https://www.usa.gov/register-to-vote>

### **Transportation:**

As an adult, it is prudent to explore the transportation options in the community. If it is appropriate, it can be a great way to increase independence. While the parent can, and probably still will in many cases, provide transportation, other options to consider are walking, riding a bike, and taking public transportation.

In many cities there are also non-profit organizations that offer door to door transportation for people with disabilities, often for a nominal fee or based on a sliding scale. Contact the

local Arc to see if this option is available in the community.  
See the NFXF website for more information on transportation options.

## Living Options After School

Just as there is a continuum of school services, there are various options for living after graduation from high school or transition programs. Housing options will depend on funding and location. To find the options in your area that are/will be available with public funding, contact the local disability organization or the local ARC.

The following is a range of living services that may be considered, from least to most restrictive:

1. Completely independent
2. Shared living environment
3. Solo living environment with intermittent case manager/care provider
4. Shared living environment with intermittent case manager/care provider
5. Communal living, such as a “Camp Hill” environment
6. Group home—numbers can vary—with a responsible adult always onsite
7. Host homes in which the person with FXS assimilates into the host family who are provided the necessary resources to care for the person.
8. Residential/assisted living with larger number of residents and opportunities to explore various types of employment and increased living skills
9. State institutions/developmental centers, only considered for individuals who are a danger to themselves or others

Note: States or regions may have various terms for identical living situations. For example, one state might call a setting “prevocational training residence” and another state might refer to the same setting as “assisted living.” Therefore, consider the description rather than the term. There may be other variations of the situations listed above, but the most important consideration is to evaluate the individual’s needs when considering a living setting. For more information about housing options see the information on the NFXF website.

## Activities of Daily Living

Whether the adult is working at a paying job (competitive employment), volunteering, attending a day program, or is able to attend post-secondary education, it is important to keep them engaged in the community every day. It is essential that they leave the house every day as many will attest that agoraphobia (fear of leaving the house) is common as people with FXS age. This has been anecdotally noted more often with males. As the adult

leaves the routine and scheduling of school, it is common for them to avoid participation in community-based events.

### **Day Programs:**

To find the options in the area where the family lives, they can reach out to the local disability organization, the local Arc, or other parents who live in the area. Parents are encouraged to visit any programs they are considering, finding out what happens during the day, and they should try to visit at different times of the day, if possible, to see how the activities change throughout the day. They should ask about transportation to the program, the hours each day of the program, whether the schedule changes on a daily basis – do they do different activities each day, and how parents/providers are notified of any changes to regular daily programming.

Also note how many other people are in the program and the number of providers they have to assist the group. Do they utilize visual schedules? Are they open to learning new skills? Basically, is it an environment where you think the adult will thrive? That will be important to keep in mind if you decide to use the program.

### **Volunteering:**

Don't dismiss volunteer jobs. They could be the best thing that the person with FXS ever does. Working at the Humane Society? In a senior center? These can be very rewarding jobs. Volunteers are an important part of many organizations, and in many cases, they are invaluable!

### **Employment/Vocational Rehabilitation Services (Voc. Rehab)**

Seek employment for the person with FXS if he or she is able to work part or full time. It is not unusual for parents to find the job for their adult child, but there are also resources to help find the job - and a job coach, if needed.

Every state has a federally funded Bureau of Rehabilitation Services that helps people who have physical or mental disabilities get or keep a job. To find out if the adult with FXS qualifies for services, contact the local Vocational Rehabilitation Services for more information. The person with FXS will meet with a counselor to determine if he or she is eligible to receive services and, if they are, they will help find employment that works for the young adult.

***For many, when thinking of “work” the first thing that comes to mind is the ability to earn money. But if one thinks about the post high school needs of adults affected by FXS, it’s so much more than that!***

During the high school years, the educational environment provides opportunities for learning, social interaction, and the ability to make friends along with opportunities to gain some level of independence. But once out of the educational setting, what provides these opportunities for enjoyment, fulfillment, feedback, social development, life-skill development, and independence? Many families have found that “work” is just that setting.

A good resource for transitioning from “school” to “work” is often available right in the high school setting or through other resources provided by your state with formal programming to identify a good employment “fit” (an area of interest and ability of your child which satisfies his or her needs for success in the workplace), and placement and “coaching” in the workplace to help the new “employee” transition successfully into the work environment.

While transition resources are likely available through a formal transition program or right out of high school, sustaining successful employment is something that usually requires long-term monitoring and management as things in the workplace change over time, as will your transitioning young adult. As one approaches employment, it’s good to think through some of the most basic questions surrounding this transition.

Working gives your young adult some things that are important for all of us ... Someplace to go, something to do ... to be among other people. Work can also provide feelings of pride and satisfaction when someone is told “you did a good job! Thank you!” And the receiving of a paycheck says “I’m valued and earned something by my work” which can be the basis of a sense of pride for the person as well as of the tangible ability to buy something with their own money. Over time, the workplace can be a place that builds strong self-esteem for the young adult. But for many, the most important aspect of work is the ability to socialize with coworkers in the workplace and develop trusted friendships.

Without work, many families have observed that post high school life leaves their young adult in a more solitary setting like in a bedroom most of the day, watching television or playing games, and perhaps eating more and moving around less, contributing to a poor health profile, loneliness and low self-esteem.

That is why it is important that your young adult continue to do something when he or she leaves high school. A job, a day program or volunteering – doing “something” during the day contributes to the well-being of the person with FXS.

What does a good work environment “look like”? This is a question that does not have any single answer, but some families report characteristics which have resulted in a successful work experience for their family member. Success in the work environment often

depends on the employer's interest and willingness to learn about unique behavioral characteristics of the adult with FXS.

Each adult with FXS is an individual but there are some challenges in the environment and interactions with the employer and other employees that can best be approached with preparation and information about your son or daughter.

The following list is not all-inclusive, but for the purpose of this paper may be considered when looking for employment. The nature of the work is something that appeals to the young adult and is something that they can be "good at". This may include:

- Working with young children in a day care setting.
- Food preparation in a kitchen or restaurant.
- Working with animals/pets at a pet shelter or pet store.
- Bagging groceries or stocking shelves in a store can be doable and enjoyable job for some and provides for social interaction with customers.
- Setting the tables, chairs, and condiments at a restaurant.
- Cleaning at local business offices (after hours for some is preferred).
- Clearing tables and placing items in dishwashing area.
- Working in a warehouse or large department store doing stocking and sorting often appeals to their need to clean up or organize which is a relative strength for people who have FXS. In addition, it provides a natural occurrence of heavy lifting and active work.

If the adult has good attention to detail in repetitive tasks, perhaps a quality inspector in a printing company would be a fit, or sorting clothes at a Goodwill or clothing recycling store.

Many males with FXS do well in work environments that allow for movement and social contacts. For example, several have been successful with a job serving juice or snacks to elderly people in skilled nursing facilities because they are able to greet the residents and engage in social exchanges which are not especially complicated and short. Others tend to enjoy working with food and providing prep to chefs. Routine and repetition usually bring success such as tasks that involve sorting and stocking store items. Paper shredding and recycling are other good examples of job tasks that have proven enjoyable and meaningful.

Some females and most males with FXS do not enjoy the unpredictability of younger children. They often become agitated when a child cries or becomes verbally loud. This can also be true with animal care, although some females have enjoyed volunteer work at a humane society or even working in a pet store.



Many females have reported good work experiences at a day care or preschool, although careful consideration to the intensity of care required by infants may need to be considered.

The environment plays a big part in job success. The social component is very important. Adults with FXS tend to enjoy relationships with their supervisors and coaches more than with peers but they like “being one of the group” at work. On the other hand, a social environment that is too large or chaotic and unpredictable tends to be stressful and not a good working environment.

Being put on the spot with employers can become debilitating. Adults with FXS want to please and may feel as though they are letting their employer down if they are unable to perform certain tasks or verbally answer questions. If the job requires a fast pace or processing, it can create stress which may result in negative or avoidant behaviors. Giving adults with FXS responsibility to count change or work with money or the cash register (even for women with FXS) is also challenging and should be avoided.

## What else needs to be in place?

An information and performance feedback loop is very important. Because the workplace is outside of your direct observation, and to ensure that workplace behavior and performance stays “on track”, it is important to have a feedback loop from someone in the workplace. This could be through a job coach who gets feedback from the employee and the employer, or sometimes, the employer is comfortable and agreeable to providing feedback directly to the family/caregiver. With a good feedback loop one can identify and address any changes in the workplace or behavior to ensure ongoing success on the job. Sometimes simple priming of a certain job-related task as part of the training can make the difference between success and failure.

*A system of prompting, early in the job to help the individual with FXS initiate the next job task and sequencing subsequent tasks to complete a job, is helpful. This reduces the problem of getting stalled between job tasks due to difficulty with initiating the next step independently.*

Being able to get to work on time and home after work are obvious requirements for success in the workplace. Coming up with mobility strategies also increases the overall independence of any individual. As mentioned earlier in this paper, some families have found success using public transportation in their communities. Others have been able to support their child obtaining a driver’s license. Your community may also have services available for transportation if public transportation or self-driving are not good options.

Putting one's "best foot forward" is equally as important after your child graduates. When your child was in the educational environment, how did you make sure they could accomplish tasks? Some of these same strategies for school success may be important for workplace. Are there environmental factors in the workplace that can be modified? Is the noise level too high? Can the employer utilize more visual signage or even post a visual chart of the task sequence? Were there any medications your child took in the school setting to help with anxiety or staying "on task" that would be appropriate to continue in the workplace?

As important as work skills are for success, so are self-advocacy skills. A team member who has a trusting relationship with the person with FXS may be able to identify and teach self-advocacy skills. This assures safety in the social environment, especially if the adult is socially vulnerable. Identifying proper resources to be used proactively such as check-ins and communication about concerns are essential to establishing safe community integration.

Many families have reported that success in the workplace is an important part of their family member achieving "life success" and a feeling of fulfillment, contribution and happiness. And, like many good things in life, it can require a lot of effort on the part of you, your child and the employer. But the rewards can be great for all. Remembering that all the adults in our Fragile X community can thrive when their opinions, preferences, and needs are well supported, and these should be considered when planning or implementing program plans with and/or for them.

For more information: <https://www.pacer.org/transition/learning-center/benefits/vocational-rehab.asp>

## Post high school education

Post high school education "isn't for everyone" and probably not necessary for most. The path to post high school activities and employment wouldn't require additional formal education. Sometimes, however, further education can be useful for both a degree and training in a field of employment interest as well as a next step in independence for the student.

One family's experience for their daughter with FXS was motivated by her interest in childcare. It all began with the daughter wanting to earn an associate degree to be better prepared for this field of work and present herself as a qualified candidate. She enrolled in a two-year community college, obtained her degree and has been successful as a teacher's assistant in a day care for many years. The parents and the daughter were very happy about her earning the degree, the experience of living independently on campus in a dorm room, and the pride that went along

with the accomplishment and the feeling of preparedness for work in the field of childcare. Asked for advice for families considering a path of higher education, the family suggested:

- Make sure that the student really wants to experience higher education and is prepared for the new environment with the determination to succeed.
- Find a school with a good fit, with the right curriculum, and an environment that has adequate supports for the additional tutoring or learning needs of the student.
- Monitor success and challenges, particularly in a situation where the student will be living away from home, arrange frequent contact to make sure everything stays on track.
- Be prepared for the unexpected.

Living in a dorm with a roommate has its rewards and challenges. Sharing the room with her roommate was complicated by things such as the roommate having a boyfriend or different schedules for studying, eating, and sleeping.

College cafeterias have a tempting and abundant offering of food choices for any student and this family's experience was difficult as the daughter gained a lot of weight without the level of nutrition supervision that one has while living at home.

Some wonderful friendships with fellow students were made. Many students and education staff "looked out for" the daughter.

Often the right post high school continuing education can be found locally without the need to live away from home. This simplifies the approach, but the student still needs a high level of determination and appropriate supports in place. One family reported their experience with a daughter with FXS who wanted to earn her Certified Nursing Assistant certification (CNA) to pursue a job of working with the elderly in a nursing home. The education was readily available at the local community college, but the lack of tutoring resources and a "more patient learning environment" resulted in a failed attempt. Eventually with more research the family found a more supportive individual education program and the determined daughter was able to achieve her CNA certification.

## Adult Living

### Relationships/Marriage

Adults with FXS do marry and can procreate. It is most likely that more females with FXS will marry and have children because they are often less affected and have more opportunities to interact with others to build relationships. Females are often attracted to a mate who is strong willed or more capable, in order to be cared for. Sometimes, adults with FXS will opt not to have children due to the amount of care children require. Some

adults who have married report a platonic relationship more typical of roommates or good friends with less sexual or romantic qualities. Nevertheless, the relationships can be extremely strong and healthy with support and care for the person with FXS.

## **Exercise, Hygiene and Health**

### **Exercise/Recreation**

It is important to keep the person with FXS active. This cannot be emphasized enough. Exercise helps to maintain or lose weight, is good for building bone and muscle, and among the many other benefits, it makes people happier. Many individuals with FXS find it difficult to engage in physical activities without support and encouragement. Often, the physical deficits such as low muscle tone, lack of understanding position in space and fear of elevated surfaces may affect the likelihood of consistent exercising. Some adults have found success hiring a personal trainer or finding a friend to help maintain an exercise routine.

In order to access activities for your adult child, call the local parks and recreation department and see if they have activities for people with disabilities (ask about unified sports), knowing that some people will be able to participate in many of the regular programs. In some cases, the parks and recreation programs will provide volunteers that will enable the person with FXS to participate in regular activities. Utilize the OT on your team to consult and problem solve to ensure successful participation. There may be college students who are studying Physical Therapy, Occupational Therapy or Sports Medicine who would be interested in working with your son/daughter.

While still school aged, be sure that the IEP includes exercise as a part of everyday activities. Investigate Special Olympics in your area: Google: "Special Olympics in (your state)". Many adults with FXS (males and females) have reported enjoyment while participating in Special Olympics sports. It allows for a time to be physically active with others who may also have similar challenges. It might be good to include an exercise schedule to group home activities. Making a physical activity fun with social contacts and support will ensure better success and promote long term positive affects.

### **Hygiene**

Hygiene is often neglected in the Fragile X population due to a myriad of issues related to sensory deficits and sensitivity. Brushing hair and teeth are often a battle due to sensory overload created by touching the head or inside of the mouth.

**Dental:**

The routine for teeth brushing is not always habituated early and can result in major dental problems that may require dental maintenance using anesthesia in order to clean, repair and remove decayed teeth. Tolerating haircuts is also difficult and needs to be instilled over time by using behavioral strategies such as systematic desensitization.

**Toileting:**

Independent toileting can be delayed especially with males. This causes significant hygiene concerns due to a delay in motor planning around wiping or tolerating the feel of toilet tissue. Some males report the smell of a bowel movement to be aversive. Diet low in fiber can cause constipation which may become problematic when trying to encourage consistent bowel training and hygiene. These issues compromise successful toileting hygiene and can become triggers for behavioral outbursts with a function to avoid bowel movements and toilet hygiene. Again, it is important to consult professionals who are familiar with FXS and strategies to development independence.

**Showering and Bathing:**

Parents report difficulty with showering and bathing, again, due to the sensory overload of water hitting the body or face of the person with FXS. Motor planning to use soap and a washcloth over the entire body to ensure cleanliness and good hygiene is often problematic. Getting water on the head and then following up with hair washing is hard and the person with FXS often tries to avoid prolonged exposure to showers and hair washing. One parent developed a visual schedule heavily laminated and posted in the shower that directed her son to wash all areas of his body before ending his shower. Other parents suggest using liquid soap, low volume shower heads and music to promote tolerance and eventual acceptance.

Having good hygiene is important for social acceptance. Grooming and hygiene play an integral part in the school, community, volunteer, and work settings. The overall presentation of one who is well groomed increases the likelihood that others will offer a positive response. This is far reaching and can improve overall wellbeing and happiness. It is wise to consult an OT to help develop proactive “Fragile X way” strategies. See [fragilex.org](http://fragilex.org).

**Health**

It is often the case with FXS that attention tends to focus on the associated emotional and behavioral problems. However, there are often two important issues that may be neglected in the care of someone with FXS. Firstly, the need to consider medical illness or disease, and secondly the need for preventive care. We encourage providers to bring this information to the attention of their medical practitioners.

## Medical issues:

It is important to remember that there may be an underlying medical cause which may contribute to a person's presenting behavior. Because many individuals have altered pain perception combined with difficulty communicating that they have pain or discomfort, it can be helpful to seek an opinion from a doctor to exclude a medical cause (see Table). This is especially the case for a recent or sudden change in behavior which is not usual for that person. For example, a person may persistently complain of something being stuck in their mouth, when in fact they have a dental abscess. Abdominal pain or irritability may be caused by constipation. Sudden behavioral outbursts may rarely be a presentation of complex partial seizures or epilepsy.

**Table 1: Medical problems associated with FXS (alphabetical order)**

Cardiovascular	Mitral valve prolapse, aortic root dissection/rupture
Dental	Crowded teeth, caries, gingivitis, root abscess
Dermatological	Dry skin, eczema, striae, foot fungus
ENT	Recurrent otitis media, hearing loss
Gastrointestinal	Constipation, reflux oesophagitis chronic or intermittent gastroesophageal reflux
Neurological	Epilepsy – tonic-clonic seizures, complex-partial seizures
Nutrition	Obesity, under-nutrition
Optometric	Strabismus, visual perception defects
Orthopaedic	Pes planus, hyperextensible joints, scoliosis, asymmetrical leg length
Psychiatric	Anxiety disorders, ADHD, OCD, ASD
Urogenital	Female – polycystic ovaries, vesicoureteric reflux, renal abnormalities Male – undescended testes, hypospadias, vesicoureteric reflux, renal abnormalities

## Preventive care

A key focus of current medicine is to detect a problem early enough to defer or prevent disease. Some examples include dental checks to maintain teeth in good condition; eye checks to detect and treat glaucoma; blood pressure, cholesterol

and sugar checks to minimize the risk of stroke, heart and kidney disease and colonoscopies to prevent bowel cancer. These examples are recommended by most primary care physicians as part of a general preventive health screen in accordance with health authority guidelines for all individuals (see references). See Table 2 for a general checklist of activities to consider. For children see the AAP Guidelines <https://doi.org/10.1542/peds.2010-3500>. For adults with developmental disability in particular, see the excellent Kennedy Shriver Massachusetts University Medical Centre guidelines <https://shriver.umassmed.edu/programs/cdder/dds-preventive-health-screenings-adults-intellectual-disabilities>.

**Table 2: Care Plan checklist**

1. DNA testing, genetic counselling for information and cascade testing, especially for the females
2. Grief and supportive counselling for family
3. Hearing assessment with audiologist
4. Vision assessment with optometrist/ophthalmologist
5. Assessment with podiatrist and or a physical therapist for orthotics and orthopaedic health
6. Speech and language therapist to optimise communication strategies
7. Psychologist: assess for IQ, ADHD and ASD, behaviour management strategies
8. Occupational therapist for skills and access within workplace and day programs; sensory issues/coping and self-regulation skills, daily life skills and engagement in finding a balance in work, life, social, recreation, wellness
9. Developmental paediatrician/neurologist/psychiatrist familiar with FXS
10. Primary care physician for preventive health screen check
11. Financial aid applications for funding
12. Information on support organisations e.g. NFXF
13. Multidisciplinary assessment at Fragile X Consortium Clinic

## Finding A Doctor Who Sees Adults with FXS

Most parents report it difficult to move from their pediatrician to a doctor who specializes in adults. The NFXF has produced a webinar related to transitioning from a pediatrician to a doctor who sees adults with FXS.

During this process, it is important to guide the adult into a new environment by following the several strategies used when supporting any transition, for example, the use of visuals. The environmental changes may be familiarized by photos of the new office building, waiting room, examination rooms and staff. In addition, a meeting with the staff with an exercise of bringing cookies or other treats to the staff by the adult with FXS before the initial appointment usually pays off.

Not all physicians are comfortable treating individuals with an intellectual disability (ID), so it is critical to talk with other parent groups as well as local disability organizations to get names of those physicians who are user friendly. Many physicians do not feel comfortable treating psychiatric symptoms so it may also be important to find a psychiatrist or Nurse practitioner who treats adults with ID.

## Medications

Maladaptive behaviors in FXS are common and significantly impact vocational and social functioning. Hence, medications are sometimes necessary to decrease disruptive, aggressive or otherwise challenging behaviors and prevent dangerous consequences or dysfunction. Furthermore, medications may facilitate the individual's ability to attain optimal life skills and allow for better integration into better living options.

Psychopharmacological (medication) treatment in FXS is recommended in conjunction with additional therapeutic services, including behavioral intervention, social skills training, vocational support and relationship building.

There are common symptoms and psychiatric conditions in individuals with fragile X syndrome (FXS): anxiety, Panic Disorder, depression, other mood disorders, eating and sleep disorders. These can be treated with the same medications that are used for these issues in the general population and in individuals with other developmental disabilities (Amaria 2001, Berry-Kravis 2004, Hagerman 2009, Valdovinos 2009, Berry-Kravis 2012, Turk 2014).



**Table 3: Medication may be useful for:**

Anxiety
Sleep disturbance
Eating disorder
Depression
Mood instability: unipolar, bipolar, cyclical, erratic
Attention Deficit Hyperactivity Disorders
OCD, tics, Tourette Syndrome
Epilepsy & associated behavioural problems
Social aloofness
Aggression & violence
Self-injurious and other challenging behaviours
Early onset psychosis

There is an assumption that data from studies of medication use in autism can be applied to treatment of FXS but whether this is valid remains unclear. These medications appear to be as effective for persons with FXS as they are for people with similar symptoms or behavioral/psychiatric disorders.

**Table 4: Current Evidence-based pharmacological therapies**

ADHD	psychostimulants, clonidine & guanfacine, amoxetone
Depression, anxiety & obsessive-compulsive features	SSRIs & SNRIs
Cyclical (& not so cyclical) mood & behaviour disorders	lamotrigine, carbamazepine, sodium valproate, lithium

Sleep induction	melatonin
Sleep maintenance	clonidine
Early onset psychosis, severe challenging behaviour	risperidone, aripiprazole, other antipsychotics

There is only limited research to demonstrate the best approach to the use of medication in the FXS population, and none demonstrates conclusively that any one type of medication, or any one medication of any one type (for example, any one of the six available SSRIs) is any more effective for people with FXS than any other. These medications treat anxiety, repetitive behaviors and mood instability in anyone who exhibits them, and the best choice may depend on factors other than FXS (for example, an individual's absorption of medication from the gut, the rate of metabolism of the medication, the medication's ability to cross the "blood-brain barrier", or an individual brain's sensitivity to the medication, controlled by genes and other factors beyond FXS). Choice of a particular medication, once the need for one is established, might be guided by side effect profile of the medication (for example, can it make seizures more probable than another medication of the same class; is it more or less likely to cause constipation; will it aggravate eating or sleep difficulties; does a given medication interact favorably or unfavorably with medications the individual is already on) or history of a family member's favorable response to a medication of a given class.

It is important to remember that individuals with FXS may be more sensitive to psychotropic medications and may respond to smaller doses than the general population or may have side effects at relatively low doses. It is wise to "start low and go slow." There are genetic tests that say they will allow choice of behavioral medications. It is important to recognize that these genetic panels measure mainly liver metabolism and some measure a limited number of genetic factors that may impact brain responses to medication. The marketers of these panels and some doctors often imply that the panels are more effective at helping with medication choice than they really are. The panels can guide dosing although if starting with a low dose anyway, the panel is not likely to impact the medication strategy. There are many things about how medication work in the brain that are not screened by the panels or we just don't understand, so in the end the panel will not really guide choice of medications in FXS more than careful systematic trials of medications in clinic with careful assessment of responses and side effects. Medications that do not work should not be continued so as to avoid the FXS individual being treated with multiple medications added on to each other instead of switching one medication for another when the first is not working.

Research into medications that may be specific for FXS has (as of this writing) just begun. Identification of the effects of FXS on a particular neural signaling pathway (the glutamate receptor) led to trials of medications affecting this receptor site. Two studies yielded disappointing results; another is still underway. One value of these studies is that they led to study of the effects of the glutamate receptor (located on the nerve cell surface, where most current psychiatric medications are known to work) inside the nerve cell, where a series of chemical reactions (a “chemical cascade”) is triggered by activation of the complex glutamate receptor system. Medications operating inside nerve cells on the chemical reactions in this cascade, “rescuing” them from the effects of FXS, would be more specific to FXS, with effects possibly going beyond the relief of symptoms offered by current psychiatric medications.

So far, all of the medications known to affect the glutamate “cascade” are medications long known and used for other purposes. The first of these is the antibiotic minocycline, which was found to substitute for the missing or deficient Fragile X Protein (FMRP). Two other medications which boost and thus “rescue” the chemical cascade are lithium (Berry-Kravis, 2008), long used to treat Bipolar Disorder, and metformin (Biag 2019, Protic 2019), long used to treat Type 2 Diabetes. As of this writing, these medications have been shown to “rescue” the chemical cascade and restore more normal functioning in both fruit fly and mouse models of FXS, and human case studies with encouraging results have been published. Controlled studies in both adults and children with FXS are planned or are currently underway.

## References:

- Amaria RN, Billeisen LL, Hagerman RJ. Medication use in fragile X syndrome. *Ment Health Aspects Dev Disabil*. 2001;4:143-147.
- Berry-Kravis E, Potanos K. Psychopharmacology in fragile X syndrome – present and future. *Ment Retard Dev Disabil Res Rev*. 2004;10:42-48.
- Berry-Kravis E, Sumis A, Hervey C, Nelson M, Porges SW, Weng N, Weiler IJ, Greenough WT. Open-Label Treatment Trial of Lithium to Target the Underlying Defect in Fragile X Syndrome. *J Dev Behav Pediatr* 2008; 29:293–302.
- Berry-Kravis E, Sumis A, Hervey C, Mathur S. Clinic-based retrospective analysis of psychopharmacology for behavior in fragile X syndrome. *Int J Peds* 2012, in review.
- Biag HMB, Potter LA, Wilkin V, Afzal S, Rosvall A, Salcedo-Arellano MJ, Rajaratnam A, Manzano-Nunez R, Schneider A, Tassone F, Rivera SM, Hagerman RJ. Metformin treatment in young children with fragile X syndrome. *Mol Genet Genomic Med*. 2019; 00:e956. <https://doi.org/10.1002/mgg3.956>
- Braden, M., (2000) *Fragile, Handle With Care, More About Fragile X Syndrome – Adolescents and Adults*, 214 E Dale St #110, Colorado Springs, CO 80903
- Erickson CA, Stigler KA, Wink LK, Mullett JE, Kohn A, Posey DJ, McDougle CJ. A prospective open-label study of aripiprazole in fragile X syndrome. *Psychopharmacology*. 2011;216:85–90.
- Hagerman RJ, Jackson AW, Levitas A, et al. Oral folic acid versus placebo in the treatment of males with fragile X syndrome. *Am J Med Genet* 1986;23:241-262.
- Hagerman RJ, Murphy MA, Wittenberger MD. A controlled trial of stimulant medication in children with the fragile X syndrome. *Am J Med Genet*. 1988;30:377-392.
- Hagerman RJ, Fulton MJ, Leaman A, et al. Fluoxetine therapy in fragile X syndrome. *Dev Brain Dysfunct*. 1994;7:155-164.
- Hagerman RJ, Riddle JE, Roberts LS, et al. A survey of the efficacy of clonidine in fragile X syndrome. *Dev Brain Dysfunct*. 1995;8:336-344.
- Hagerman RJ, Hills J, Scharfner S, Lewis H. Fragile X syndrome and selective mutism. *Am J Med Genet* 1999;83 ;313-317.
- Hagerman RJ, Berry-Kravis E, Kaufmann WE, et al. Advances in the treatment of fragile X syndrome. *Pediatrics*. 2009;123:378-390.
- Ingrassia, Turk J. The use of clonidine for severe and intractable sleep problems in children with neurodevelopmental disorders: a case series. *European Child & Adolescent Psychiatry*, 2005;14:34-40.
- Kronk R, Bishop E, Raspa M, Bickel JO, Mandel DA, Bailey DB. Prevalence, nature, and correlates of sleep problems in fragile X syndrome. *SLEEP*. 2010;33:679-687.
- Protic D, Aydin EY, Tassone F, Tan M, Hagerman RJ, Schneider A. Cognitive and behavioral improvement in adults with fragile X syndrome treated with metformin-two cases. *Mol Genet Genomic Med*. 2019; 7:e745. <https://doi.org/10.1002/mgg3.745>

- Strom CM, Brusca RM, Pizzi WJ. Double-blind placebo-controlled crossover study of folinic acid (Leukovorin) for the treatment of fragile X syndrome. *Am J Med Genet.* 1992;44:676-682
- Torrioli MG, Vernacotola S, Peruzzi L, et al. A double-blind, parallel, multicenter comparison of L-acetylcarnitine with placebo on the attention deficit hyperactivity disorder in fragile X syndrome boys. *Am J Med Genet A* 2008;146:803-12.
- Turk J. Sleep disorders in children and adolescents with learning disabilities and their management. *Advances in Mental Health in Learning Disabilities* 2010;4:50-59.
- Valdovinos MG, Parsa RA, Alexander ML. Results of a nation-wide survey evaluating psychotropic medication use in fragile X syndrome. *J Dev Phys Disabil.* 2009;21:223-37.

### **Additional Citations and Resources**

- Joseph H. Hersh, Robert A. Saul and Committee on Genetics  
*Pediatrics* May 2011, 127 (5) 994-1006; DOI: <https://doi.org/10.1542/peds.2010-3500>  
Eunice Kennedy Shriver Center, University of Massachusetts Medical School: DDS Preventive Health Screenings for Adults with Intellectual Disabilities:  
<https://shriver.umassmed.edu/programs/cdder/dds-preventive-health-screenings-adults-intellectual-disabilities>  
CDC statement: <https://www.cdc.gov/ncbddd/disabilityandhealth/hcp.html>  
from CDC  
WHO statement: <https://www.who.int/news-room/fact-sheets/detail/disability-and-health>  
Canadian guidelines: [https://www.researchgate.net/publication/51126266\\_Primary\\_care\\_for\\_adults\\_with\\_Developmental\\_Disabilities\\_Canadian\\_consensus\\_guidelines](https://www.researchgate.net/publication/51126266_Primary_care_for_adults_with_Developmental_Disabilities_Canadian_consensus_guidelines)  
UK guidelines: [https://www.ndti.org.uk/uploads/files/RH\\_Health\\_Checks\\_Guide.pdf](https://www.ndti.org.uk/uploads/files/RH_Health_Checks_Guide.pdf)  
Occupational Therapy scope of practice and role with adults with IDD:  
<https://www.aota.org/About-Occupational-Therapy/Professionals/RDP/Intellectual-Disabilities.aspx>

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The Fragile X Clinical & Research Consortium was founded in 2006 and exists to improve the delivery of clinical services to families impacted by any Fragile X-associated Disorder and to develop a research infrastructure for advancing the development and implementation of new and improved treatments. Please contact the National Fragile X Foundation for more information.

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