



NATIONAL **FRAGILE X** FOUNDATION
FRAGILE X CLINICAL & RESEARCH CONSORTIUM

Consensus of the Fragile X Clinical &
Research Consortium

TOILETING ISSUES IN FRAGILE X SYNDROME

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Introduction

Toilet training, which can be challenging even in the general population, is characteristically an area of stress for families with a child affected by fragile X syndrome (FXS). While the majority of boys and girls with FXS will become toilet-trained, this is typically delayed anywhere from one to multiple years later than the general population. An analysis of data from FORWARD (Fragile X Online Registry With Accessible Research Database), a natural history project collecting data from FXS patients at most of the FXCRC (Fragile X Clinical and Research Consortium) Clinics, has recently provided new information on the age of toilet training in children with FXS, and factors that impact when children with FXS will toilet train (Berry-Kravis et al. 2019, reference provided at the end of the document). This allows an understanding of which patients are most at-risk for very late toilet training and thus can guide training strategies in terms of when intensive toilet training techniques will need to be applied. For most males with FXS the various stages of toilet training will each likely take longer to master and will require specific training geared to both global characteristics of FXS and the individual needs of each child.

Age of Toilet Training in Males and Females with FXS

Data from 633 individuals with FXS enrolled in FORWARD between 2012 and 2016 (77% male) were used to generate the curves (called survival curves) shown in the Figures at the end of this document; (Figures 1 and 2 for bladder training and Figures 3 and 4 for bowel training). The figures compare females (red/pink) and males (blue/green). These curves tell us for any given age (plotted along the bottom of the chart) what the chance a male or female with FXS will be toilet trained for urination (Figure 1) or bowel (Figure 3). Overall, females train much earlier than males, and males train later with about half of males trained at age 5. There are some individuals (mainly males) with FXS who do not train at all or are trained very late (after age 10). Figures 2 and 4 show the male and female curves like Figures 1 and 3 but divide the groups into those with a co-diagnosis of autism spectrum disorder (ASD) and those without ASD. Most of the males who train very late have a co-diagnosis of ASD. For males with FXS without ASD, about 65% are trained by age 5. Additional description is provided in the text with the Figures at the end of the document.

An analysis that included the 313 individuals with FXS over age 10, showed that 39 individuals (12.4%) were not toilet trained at age 10. All but 1 of these was male. This severe delay in toilet training was associated with low or nonverbal language abilities, lower IQ, presence of ASD, and severe behavior in areas of both irritability

(being easily upset, over-reacting to things, tantrumming) and hyperactivity. Since many of these features can occur together, statistical models (multivariate and Cox proportional hazard models) were used to determine which factors are the main drivers of late toilet training. These analyses showed that lower language milestones, ASD comorbid diagnosis, and irritable behavior were the only independent predictors of severe delay in toileting. Of these, language and communication milestones were the biggest predictor of age of toilet training, such that for each higher language level of 1) small number of words or less, 2) many single words and two-word phrases, and 3) phrases of 3 words and sentences, the individuals with FXS were twice as likely to be toilet trained.

Diagnosis/Recognition

Addressing toilet training is essential for all children with FXS, especially males as a large percent will exhibit delayed toilet training, even if they do not have ASD or low language skills. Often the family will be struggling with numerous issues and just assume that delayed toilet training is expected. They may even be under the impression that acquisition of this skill is unrealistic. However, the converse is also true. Many families initiate toilet training based on the child's chronological age and expected milestones, based on typically developing children. Initiating toilet training too early is often linked to extraneous factors, primarily related to childcare and preschool policies. Either situation requires guidance. Medical providers should therefore actively consider the developmental age of the child, and inquire about toileting skills for both bladder and bowel during night and day, and also about washing and wiping abilities. It is important to define the nature of the toileting issues, as there are different causes and treatments depending on whether the child never acquired toileting skills at all (primary encopresis and enuresis, respectively), or was toilet trained in the past and then lost control of bowel and/or bladder (secondary encopresis and enuresis, respectively).

Current Treatment Guidelines

Current treatment guidelines for toilet training in children with FXS are not intrinsically different from the general population, but must take into account the increased anxiety, sensory defensiveness, and in particular, the poor sequential learning skills of this population. It is also important to impress upon families that while eventual mastery can be expected, the time scale must be expanded. Each individual's readiness must be carefully evaluated, as forcing the issue too early is likely to be highly counterproductive. Toilet training is actually a complicated

behavior that involves elements of physiological, motor, and communicative readiness.

The first step in training is to make sure that the child is ready.

Readiness Signs

- Being dry for periods of at least two hours
- Recognition that a diaper is wet or soiled
- Indication of sensory awareness of having a full bladder or bowels
- Ability to communicate about wetness or toileting need
- Motor skills to get to the bathroom, pull pants down and sit on the toilet
- Ability and willingness to follow simple one-step directions
- Ability to sit in one spot for several minutes at a time.

Discussion Points with Parents

Though the following suggestions are divided by domain it does not mean that they should be implemented in isolation. This division illustrates the complex nature of toilet training and is designed to ease discussion with parents.

Physical

It is important to evaluate for any medical causes of toileting problems. As noted above, there are different medical causes for day and/or night as well as primary and secondary enuresis and/or encopresis. Any condition that can affect a child in the general population can also affect a child with FXS; it is therefore important to conduct a general pediatric evaluation for these conditions (the discussion of which is outside the scope of this guide but might include constipation, urinary tract infections, seizures, etc.).

- Discuss the consistency of the child's stools to determine if additional fiber supplements should be added to his/her diet. The significant sensory issues associated with FXS can sometimes result in a very limited diet, thus affecting the consistency of the child's stool. If that appears to be the case, then further strategies for dietary options may be considered with a pediatric dietician and/or a therapist (e.g. occupational, behavioral) with experience in helping to increase children's willingness to explore other foods.
- Discuss the side effects medications may have on the child's bowels. For example, some antibiotics cause diarrhea and guanfacine and antipsychotics like risperidone (Risperdal) and aripiprazole (Abilify) can cause constipation in some children.
- Encourage parents to track the child's toileting schedule. It is generally easiest

to start tracking bowel movements. Once a pattern has been established, they should begin placing the child on the toilet around the time she usually has a bowel movement. (Initially the parent rather than the child will be trained!)

- Once success has been achieved with bowel movements, parents can use the same process with urination. For many children, the opposite sequence will be more successful.
- Medication is not generally indicated, though the use of desmopressin to treat nighttime urinary incontinence for specific situations, e.g. trips, may be warranted.

Sensory

- Disposable diapers are so effective they may hinder the toilet training process because they do not allow the child to feel the wetness of the diaper. Many children with FXS are successful using cloth training pants so the child can tell when he/she is wet. Another way to accomplish this is to consider placing a non-absorbent cloth in the child's diaper so he/she can be aware of feeling wet.
- Some children do not feel comfortable pooping outside of the diaper. If that is the case, place a diaper loosely around the child and allow her to poop while sitting on the toilet. Gradually place the diaper farther away from her bottom and closer to the bottom of the toilet. This type of behavior shaping allows the child to gradually feel safer while adapting to the toilet. Sometimes a hole can be cut into the diaper so the child still feels the diaper around his/her waist and that helps the child feel more comfortable making a bowel movement.
- Many children with sensory issues have gravitational insecurity, meaning they do not feel comfortable when they are up off the ground. Encourage parents to place the toilet on the ground, or to use steps or a bench that are solid to add a feeling of security.
- Eliminate or at least reduce "exotic" smells as a way to limit sensory input in the bathroom. (If weather permits and noise from outside is minimal, an open window can help.)

Language

- Encourage the use of simple, concrete and consistent language when referring to body parts as well as the toileting act itself.
- If the child is non-verbal, apply whatever type of communication system is used in the other parts of his life. The speech therapist should be alerted to

the toilet training experience so she or he can provide assistance in this area. There are many picture schedules and social stories available as visual aides to support toileting skills.

- Excessive language is not usually beneficial. Short, two-to-three-word sentences and directions are recommended.
- Since language development is often an issue, pair these words with signs. Research has shown the introduction of sign language often stimulates oral language.

Cognitive

Learning a new skill can be a slow process for children with FXS, but they do learn and make consistent progress. The key is to present the information in a way that is meaningful to the child. Individuals with FXS have strong imitation skills, and they learn by observing and copying.

Encourage parents to do the following:

- Read books to the child about toilet training, and watch toilet training videos with him. Choose books that have limited language. Sitting and listening to an entire story is difficult.
- Allow the child to see family members or other children using the toilet, and use observational remarks such as, “He is going potty” to narrate what is happening. Remember to use short sentences.
- Practice toilet training with a doll, action figure or stuffed animal, e.g. “Sponge Bob goes potty.” This allows the child to have control over the situation and to practice the sequencing of the tasks without pressure. This type of activity is also helpful for decreasing anxiety.

Motor

- In order to account for fine motor delay, encourage parents to dress the child in clothes that she can manipulate easily. Elastic waistbands work the best.
- Due to low muscle tone, balance can be difficult for children with FXS. Have parents make sure that the child’s feet can touch the floor when sitting on the toilet. This will increase his stability and make him feel safe. When using the big toilet, place a block or an old phone book under his feet.
- Wiping also requires motor skills and coordination, and can be a challenge for some children. Occupational therapists may be able to provide some additional guidance and strategies if this is an area of challenge.

Psychological

Psychological factors are probably the most difficult of the entire process. Children with FXS often experience anxiety, but due to their limited language skills, we typically see only the ensuing avoidant behavior. This can leave parents feeling frustrated and confused. It is best to try to decrease the anxiety, rather than try to guess what is causing it. Anxiety can be decreased by presenting the child with information about the situation in a way that is easily understood. Practice and repetition will familiarize the child with the new situation and the items that go with it. Medications such as selective serotonin reuptake inhibitors (SSRIs) should be used to treat underlying anxieties that are affecting the child generally, and not specifically for toileting related issues.

- Provide a picture schedule.
- Spend time talking about the toilet, looking at it, touching it and sitting on it before it is time to actually use it.
- Do not initiate this process during known times of stress, i.e. right after a move, the arrival of a new baby, or other major changes.
- Involve the child in the selection and purchase of the training pants. This allows him/her to exert control in an appropriate manner. Involve him/her in the selection of the potty chair or toilet seat as well.
- Allow the child to decorate the toilet with stickers, etc.
- Ensure that the child can place his/her feet on the ground or stable platform; this has been found to be very helpful in diminishing associated fears.
- Modify the amount of time that the child sits on the toilet when first learning. Guides for typically developing children often recommend 10 minutes. Initially, this may be too long for children with FXS as many of them will have ADHD.
- Novelty will increase attention. Encourage the placement of some books next to the toilet. These books or toys should be engaging and should be kept in the bathroom, which will keep them “new” and engaging. Use whatever is engaging for the individual child.
- Pushing a child who is not developmentally ready for toilet training may slow or disrupt the process. If the parent is becoming too frustrated, encourage him or her to stop and try again in a month or so. There is nothing wrong with a parent taking time to regroup, and it may well benefit the child.
- Providers should work with parents to carry out a careful analysis of what is happening in order to separate antecedent behavioral elements from developmental/physiological factors. This will help in creating a developmentally appropriate plan for the individual child.
- Identified behavioral elements, if present, should be addressed with appropriate therapies.

- Cognitively, training with visual cues (e.g. video modeling, likely via cartoon, rather than showing an actual child on the toilet) is often very helpful.
- The preschool/school should work with the family, as far as possible, to use the same system for training.
- A behavioral reward system based on verbal praise should be helpful, as individuals with FXS have a strong drive to please.
- Regarding isolated nocturnal enuresis, consideration may be given to the “pad and bell” alarm systems. This is a popular method for treating nocturnal enuresis in the general population. If indicated, one of the less forceful variants (e.g. a pad and buzzer, rather than bell) may be a better option. Generally this should be undertaken only after careful consideration, since sleep disturbance can be a problem in FXS and will likely be exacerbated with this method. In addition the sensory stimulus of the bell may increase anxiety and potentially worsen general behavioral problems.

Special Focus for Children At-Risk for Very Delayed Toilet Training

For children with FXS with poor language function, co-diagnosis of ASD, or irritable behavior that puts them in a higher risk group for very delayed toilet training based on the FORWARD data, additional effort and different approaches may be required from early in training. Since language is considered to be a central factor in toilet training, priming using language-heavy books or lengthy conversations should be not be done. Punishment procedures that involve language and/or extensive multi-step processes of restitution, including cleaning up and putting the soiled clothes in the laundry, may cause too much frustration and, without effective language skills, may result in a lack of associating the punishment with the toileting accident; this would, in turn, eliminate any positive effect of the punishment, and as such is also ill-advised. Scheduled sitting could be particularly effective because language could be diminished or virtually eliminated with the implementation of a picture schedule, a visual mechanism for marking time, and simple sign language for communicating success and completion. In addition, the use of video modeling or priming using picture books or social stories could be very effective.

Previous literature recommends starting with bowel training rather than bladder training; however, the data from FORWARD are not consistent with bowel training occurring first. Bladder training occurs before bowel training for many individuals with FXS, and based on the predictors identified from FORWARD, bladder training can be addressed first for children who seem predisposed to this. This suggests that the utilization of elimination schedules as a foundational component of any

toilet training approach with individuals with FXS is essential. Elimination schedules identify the patterns of elimination, which can be used to inform a scheduled sitting approach in which individuals are placed on a toilet for set amounts of time and rewarded when they void their bladder or make a bowel movement. The schedule increases the likelihood of “catching” the child at the right time so that they void when placed on the toilet. Since bowel movements come less frequently and are easier to notice and track, the likelihood of success at “catching” the child at the right time is increased (this may have been the reason for the idea that bowel training occurred first). The key is to acquire accurate data for the individual child to inform the process, the order of training, and to allow for many opportunities to reinforce success.

For children at high-risk for late training, it is important to consider accommodations and modifications to the environment that take into consideration the known behavioral phenotype of individuals with FXS. As previously discussed, modifying the bathroom environment by providing the child a stool to rest their feet on and an insert for the toilet might make some feel more stable. Decreasing the sensory stimuli such as lotions, potpourri, and candles might also be effective for those with sensory sensitivities. Technological aides for detecting beginning urination could also be considered. It will be important for the quality of life for both the individual child and the family to set appropriate expectations and to introduce alternative toilet training strategies.

Information from the FORWARD data can inform toileting practices and expectations for children with FXS child with ASD and/or poor language skills. It will be important to focus on the foundational skills for daily living related to toilet training, such as teaching the child to remove clothes, to sit on the toilet for several minutes 2 to 3 times per day, and to flush the toilet and wash hands. This might be a more prudent and appropriate strategy for parents and teachers, rather than expecting traditional successful toilet training during the preschool and early elementary school years. This does not suggest that anyone has “given up” on independent toilet training, but rather acknowledges the significant challenges for some children and creates a more appropriate plan which will benefit both the child and his/her family. The expectation should be to employ consistent work towards toilet training across multiple years to achieve gradual progress -- extra time or schedule training will probably be most effective. Individuals who are schedule trained can engage effectively in their communities with their peer groups and with the technology available can learn to use the toilet almost completely independently. Watches, phones, and iPads can be programmed with timers

to tell an individual it is time to use the restroom. A schedule training approach will require the parents to work with a behavioral specialist, trained educator, occupational therapist or a psychologist to create an individual plan. Adjusting and resetting expectations about toilet training and toileting independence for the individual is key. The focus should be on allowing the individual to live the most meaningful life in the long term and that often means setting a goal on reducing barriers to inclusion across the lifespan. It is important to note that initially this may be very difficult for parents and may trigger a grief cycle as it represents yet another loss. Acknowledging this will be crucial and counseling support may be advised.

Common Q & A

What should I tell my patients about toilet training in FXS in general?

The child will become toilet trained but patience and consideration of readiness and developmental level are key. The child will (usually) not be toilet averse for oppositional reasons though there may be an anxiety and/or behavioral component in addition to standard physiologic considerations. It is important to carefully analyze the process and break down the different elements involved in order to move forward in a planned manner, emphasizing that speed of training is less important than consolidation and assimilation of skills.

How do I help families work effectively with the preschool/school around toileting?

It is important to note that issues around toileting are often related to licensing requirements rather than the personal preferences of the teachers. Classrooms are licensed for different types of activities. Changing diapers is one activity and the classroom will be licensed by the state's Health and Human Service Department according to its ability to conduct this activity in a safe manner. This licensing includes physical structures such as child size toilets and diaper changing areas as well as training for those who will be involved in changing diapers.

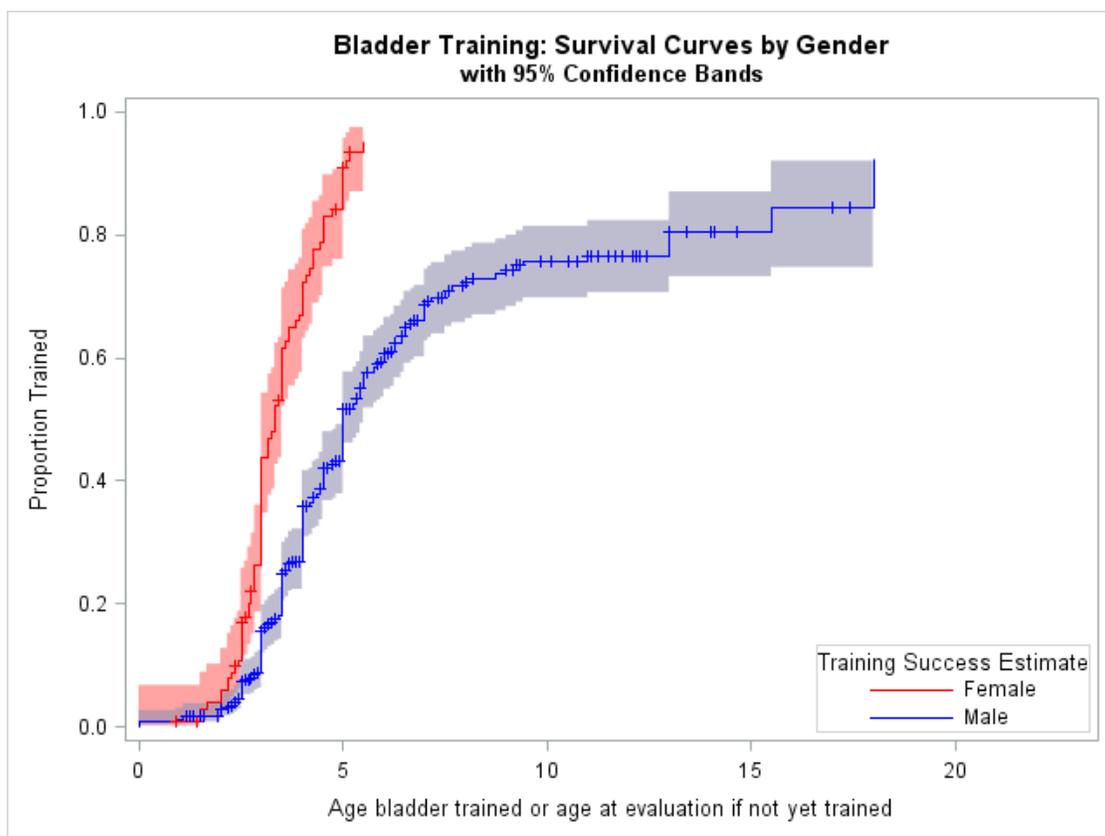
Communication and education in working with the school is key. Toileting is another one of the elements of FXS that require the entire team to work together. The individualized education program (IEP) should incorporate toileting as one of its goals if this skill is not yet accomplished. This will ensure that the child is provided with the necessary opportunities to practice toileting, and will also provide the teacher with the necessary supports needed to accomplish this task in a way that meets licensing requirements. For individuals who are on IFSPs or are attending preschool in independent settings, work with the director to understand the

limitations and opportunities within their licensing and then work on a plan that will serve the child's needs within these parameters. This can be accomplished in a variety of ways, again the key is communication and creativity.

Figures Depicting When Individuals With FXS Toilet Train

In all of the figures age is shown on the horizontal line at the bottom of the graph. This is plotted against the proportion of individuals toilet trained at a given age (eg. 0.6 would mean 6 of 10 or 60% of individuals are trained).

Figure 1 showing when males and females with FXS achieve bladder or urinary toilet training (FORWARD Project)



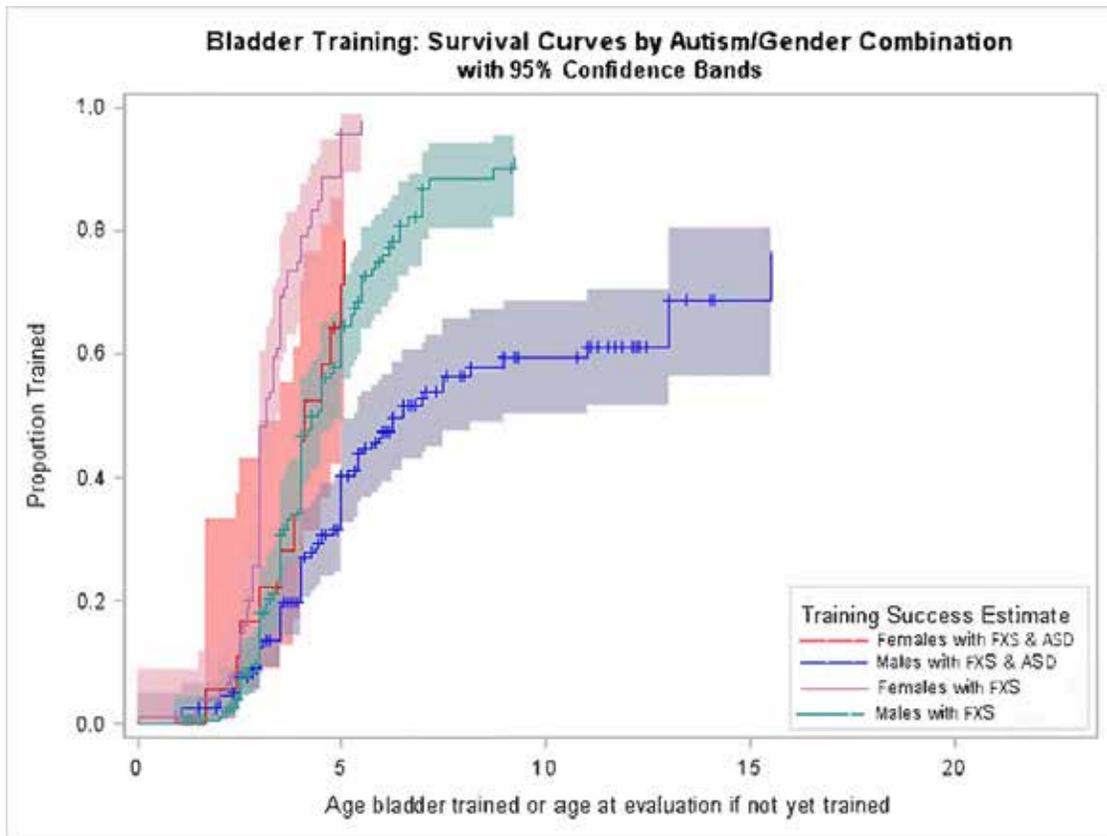
The red line shows the time when females achieve bladder toilet training. By 5 years of age, almost all (95%) females achieve training – this is not far beyond typically developing females.

The blue line shows the time when males achieve bladder toilet training. By 5 years of age, only about 40% of males achieve training. By age 10, about 75% of males

achieve training, but beyond that age, there are some males that are still having difficulty.

NOTE: These lines include individuals with FXS with and without ASD.

Figure 2 showing when males and females with FXS (with and without ASD) achieve bladder or urinary toilet training (FORWARD Project)



The purple line shows the time when females with FXS without ASD achieve bladder toilet training. By 5 years of age, almost 100% of females achieve training. ASD status in females does not make much of a difference in achieving bladder toilet training.

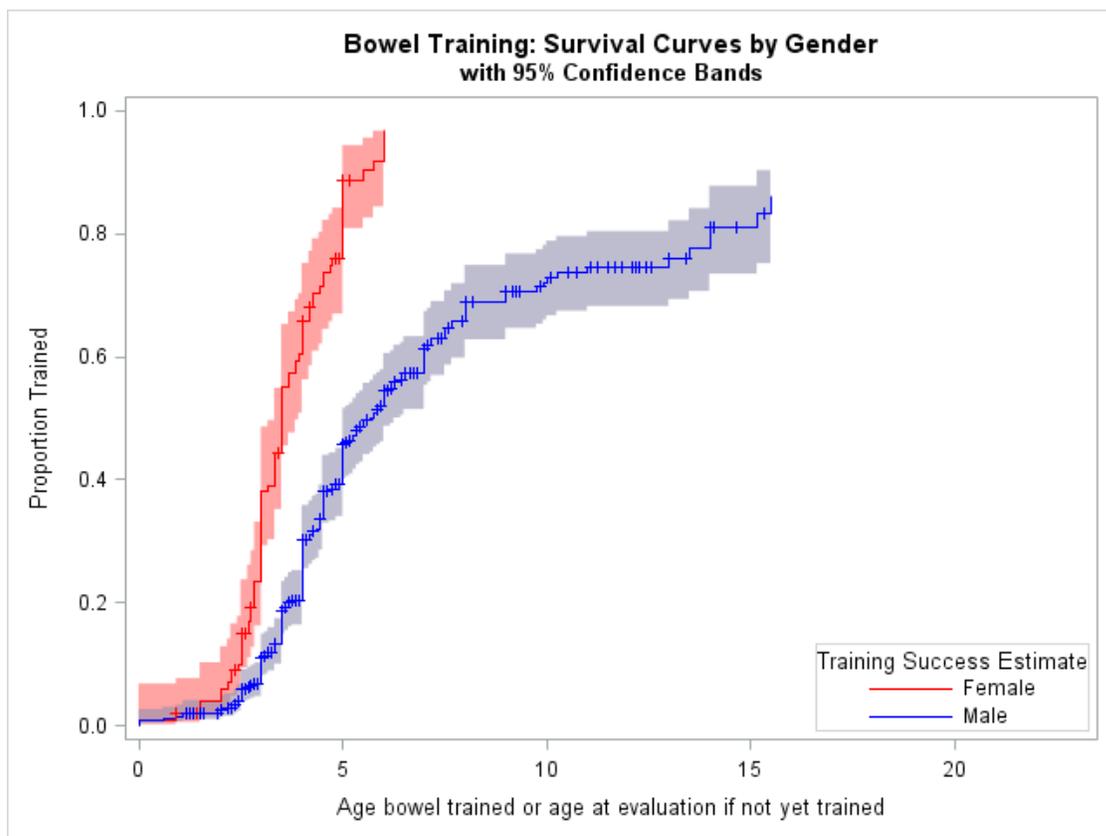
The red line shows the time when females with ASD achieve bladder toilet training. By 5 years of age, almost 70% of females with ASD achieve training –females with FXS and ASD lag slightly behind females with FXS without ASD, but the lag is not substantial. There is not enough data on females to give a good idea beyond this age, but we encourage those who have not achieved training to have further evaluation for alternative approaches and new strategies.

The green line shows the time when males with FXS without ASD achieve bladder toilet training. By 5 years of age, about 50% of males achieve training. By age 10, about 90% of males achieve training. At age 4, the time to achievement starts to diverge for males, with those without ASD achieving at a much higher rate earlier than males with ASD.

The blue line shows the time when males with FXS and ASD achieve bladder toilet training. By 5 years of age, only about 35% of these males achieve training. By age 10, about 60% of males achieve training and there is some success beyond that age, with about 70% at age 15.

NOTE: An assessment for ASD for males over 6 with bladder toilet training challenges may be helpful, because if the child has ASD, it may be helpful to modify strategies in order to achieve toileting success.

Figure 3 showing when males and females with FXS achieve bowel toilet training (FORWARD Project)

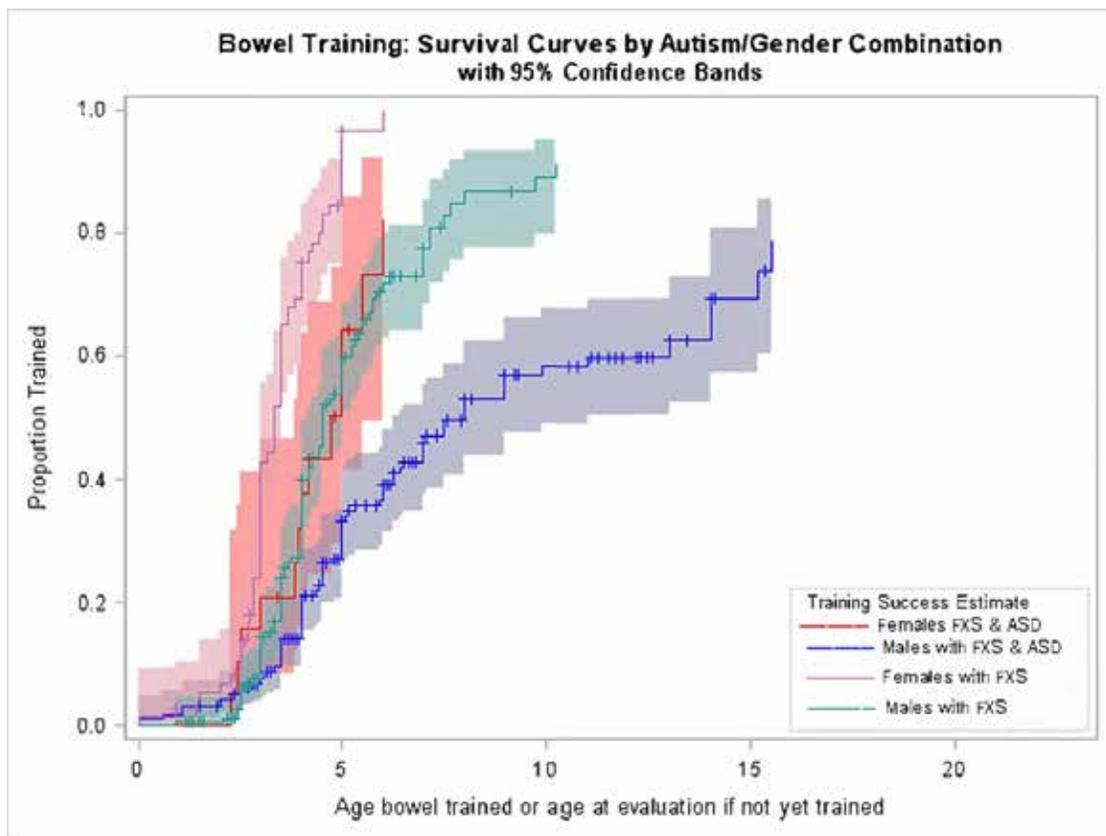


The red line shows the time when females achieve bowel toilet training. By 6 years of age, almost all (95%) females achieve training – this is about 2 years beyond typically developing children.

The blue line shows the time when males achieve bowel toilet training. By 6 years of age, only about 50% of males achieve training. By age 10, about 70% of males achieve training with a plateau of success until age 15 (80%), and beyond that age with some males that are still having difficulty.

NOTE: These lines include people with and without ASD.

Figure 4 showing when males and females with FXS (with and without ASD) achieve bowel toilet training (FORWARD Project)



The purple line shows the time when females with FXS without ASD achieve bowel toilet training. By 6 years of age, almost 100% of females achieve training. ASD status in females does not make much of a difference in time to achieving bowel toilet training.

The red line shows the time when females with FXS with a diagnosis of ASD achieve bowel toilet training. By 6 years of age, almost 75% of females with ASD achieve

training –females with FXS and ASD lag behind females without ASD, but the lag is not substantial. There is not enough data on females to give a good idea beyond this age, but we encourage those who have not achieved training to have further evaluation for alternative approaches and new strategies.

The **green** line shows the time when males with FXS alone achieve bowel toilet training. By 6 years of age, about 70% of males achieve training. By age 10, about 90% of males achieve training. At age 6, the time to achievement starts to diverge substantially for males, with those without ASD achieving at a much higher rate earlier than males with ASD.

The **blue** line shows the time when males with FXS and ASD achieve bowel toilet training. By 6 years of age, only about 35% of males achieve training. By age 10, about 55% of males achieve training and there is some success beyond that age, with about 70% at age 15.

NOTE: An assessment for ASD for males over 6 with bladder toilet training challenges may be helpful, because if the child has ASD, it may be helpful to modify strategies in order to achieve toileting success.

Additional Resources / References

1. NIH guide on bedwetting:
<http://www.nlm.nih.gov/medlineplus/bedwetting.htm>
2. Hepburn, S. (2009). Toilet Training for Children with Special Needs. Western Media Products. <http://media-products.com/catalog/toilet-training-children-with-special-needs-p-121.html>
3. Berry-Kravis E, Kidd SA, Lachiewicz AM, Choo TH, Tartaglia N, Talapatra D, Aguirre-Kolb C, Andrews H, Riley K, Toilet Training in Fragile X Syndrome, Journal of Developmental Behavioral Pediatrics, 2019 Dec;40(9):751-761
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Toilet Training the Older Child

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Author note: This guideline was originally authored by Karen Riley, PhD, Jonathan Picker, MD, PhD, and Ramzi Nasir MD, MPH. The 2019 updated document was authored by Karen Riley PhD, Elizabeth Berry-Kravis MD PhD, Sharon Kidd PhD, Ave Lachiewicz MD, Nicole Tartaglia MD, and Christina Aguirre-Kolb PhD, and was reviewed and edited by consortium members both within and external to the Fragile X Clinical & Research Consortium. It has been approved by and represents the current consensus of the consortium.

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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