Consensus of the Fragile X Clinical & Research Consortium on Clinical Practices

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 vast majority of boys and girls with FXS will become toilet-trained, it is typically delayed anywhere from one to several years later than the general population. This means 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.

Diagnosis/Recognition

Addressing toilet training is essential. 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 child care and preschool policies. Either type of case requires guidance. Medical providers should therefore actively inquire about toileting skills for both bladder and bowel during night and day, and about washing and wiping abilities and inclinations as well. It is important to define the nature of the toileting issues, as there are different causes and treatments depending on whether the child has ever been toilet trained in the past and then lost control of bowel and/or bladder (secondary encopresis and enuresis, respectively) or never acquired these skills at all (primary 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
- Indication of sensory awareness of having a full bladder or bowels

- Recognition that a diaper is wet or soiled
- 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 diet. The significant sensory issues associated with FXS can 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.
- Encourage parents to track the child's toileting schedule. It is generally easiest to start with bowel training. 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.
- Medication is not generally indicated, though the use of desmopressin 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. Encourage the use of cloth training pants so the child can tell when he is wet. Another way to accomplish this is to

consider placing a non-absorbent cloth in the child's diaper so he 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.
- 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 potty on the ground, or to use steps 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.
- Excessive language is usually lost on these children. 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 potty 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.

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 potty. This will increase his stability and make him feel safe. When using the big potty, place a block or an old phone book under his feet.

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 and will familiarize the child with the new situation and the items that go with it. 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 her to exert control in an appropriate manner. Involve her in the selection of the potty chair as well.
- Allow the child to decorate the potty with stickers, etc.
- Ensure that the child can place his 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 potty when first learning. Guides for typically developing children often recommend 10 minutes, which is probably too long for these children as most 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 at potty) 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." 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.

Common Q & A

What should I tell my patients?
The child will become toilet trained but patience is the key. The child will (usually) not be toilet averse for oppositional reasons though there may well be a behavioral component in addition to the physiological ones. It is therefore 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.

What to do with the preschool/school who are unhappy about the toileting situation?
Education of the educators is key. Generally toileting is just one of the elements of FXS that need to be explained to the teacher, aides, nurse and other school personnel. The individualized education program should incorporate toileting as one of its goals if this skill is not yet accomplished.

Additional Resources

NIH guide on bedwetting: http://www.nlm.nih.gov/medlineplus/bedwetting.html

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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. (800-688-8765 or www.fragilex.org)*