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Mothers' perspectives on challenging behaviours in their children with fragile X syndrome

Kristen Muller^a, Nancy C. Brady^a, Steven F. Warren^a, Kandace K. Fleming^b

^aDepartment of Speech-Language-Hearing: Sciences and Disorders, The University of Kansas, Schiefelbusch Institute for Life Span Studies, Lawrence, KS 66045, USA;

^bResearch Design and Analysis Unit, The University of Kansas, Schiefelbusch Institute for Life Span Studies, Lawrence, KS 66045, USA

Abstract

Background: Individuals with intellectual and developmental disabilities are more likely to engage in problem behaviours than peers with typical development. The purpose of this study was to provide descriptive and qualitative information about problem behaviours in children with fragile X syndrome (FXS) and how families respond to these behaviours.

Method: We examined interview responses from 53 mothers of 9-year-old children with FXS.

Results: Defiance, tantrums, inattention, stereotypy, and aggression were the most frequently reported problem behaviours of children with FXS. Stereotypy, physical aggression, self-injury, and elopement were reported more often by mothers of children with dual diagnoses of FXS and autism than by mothers of children with a single diagnosis of FXS.

Conclusions: Results indicated that for many families, by 9 years of age, problem behaviours led to frustration and altered family activities.

Keywords

Fragile X syndrome; autism spectrum disorders; problem behaviour

Fragile X syndrome (FXS) is the most common inherited cause of intellectual disability (Crawford, Acuna, & Sherman, 2001) and autism spectrum disorder (ASD) (Duy & Budimirovic, 2017). It affects approximately “1 in 4000” and “1 in 6000” females (Crawford et al., 2001). The average age of diagnosis is 35–37 months for males and 42 months for females (Bailey, Raspa, Bishop, & Holiday, 2009). FXS is caused by expansions of a CGG triple repeat segment located in the FMR1 gene in the X chromosome (Kaufmann & Reiss, 1999). Genetic tests that identify the number of CGG repeats in the FMR1 gene are used to diagnose FXS and identify pre-mutations that may lead to FXS when passed to future generations. Individuals with FXS experience the full mutation of the FMR1 gene (> 200 CGG repeats) (Maddalena et al., 2001) and typically have cognitive and language delays.

CONTACT Kristen Muller kmuller2@kumc.edu, mullerkristen2@gmail.com.

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Individuals who have a premutation of the FMR1 gene (~55–200 CGG repeats) do not have FXS, but the premutation can expand when passed to future generations and cause FXS in offspring (Fernandez-Carvajal et al., 2009).

Typically, males are more severely affected than females due to the X-linked inheritance of the mutation (Crawford et al., 2001). Males with FXS usually display more severe intellectual impairment, behavioural and social impairments, and language delays than females with FXS. In addition, 30–46% of males and 10–16% of females with FXS have a comorbid diagnosis of ASD (Bailey, Raspa, Olmsted, & Holiday, 2008; Demark, Feldman, & Holden, 2003). ASD is a developmental disorder characterised by social communication deficits and restricted, repetitive behaviour or interests. The average age of ASD diagnosis is between 4 and 5 years old (Christensen et al., 2016). The purpose of this study was to provide descriptive and qualitative information about how problem behaviours impact family functions during middle childhood.

Problem behaviours

Individuals with intellectual and developmental disabilities (IDD) are more likely to engage in problem behaviours than peers with typical development (Eisenhower, Baker, & Blacher, 2005; Kiernan & Kiernan, 1994; Schroeder et al., 2014). Problem behaviour is behaviour that is undesirable and typically deviates from the legal or social norms of society (Jessor, 1982). Self-injurious behaviour (SIB), aggression, and property destruction are examples of frequently reported problem behaviours (Schroeder et al., 2014; Soke et al., 2016). Individuals who engage in problem behaviour tend to have low scores across many indicators of quality of life (Emerson et al., 2001; Rusch, Hall, & Griffin, 1986). These behaviours also correlate with high levels of parental stress (Eisenhower et al., 2005; Hastings, 2002).

According to a 2007 literature review, the most common problem behaviours exhibited by children with FXS were hyperactivity, tantrums, irritability, stereotypy, poor eye contact, SIB, and aggression (Valdovinos, 2007). These behaviours have been observed in both genders, but more often in males (e.g., Hatton et al., 2002; Kau et al., 2004; Valdovinos, 2007). Hessel et al. (2001) reported that males with FXS have social deficits, limited communication abilities, social avoidance, limited eye contact, unusual responses to sensory stimuli, stereotypic behaviour, inattention, hyperactivity, and impulsivity. Females often exhibit social withdrawal, depression, and hyperactivity; however, these symptoms are less severe than in boys with FXS (Hessel et al., 2001).

Parents of children with FXS reported that challenging behaviour such as self-injury may begin as early as 12–15 months of age (Hessel et al., 2008; Symons, Clark, Hatton, Skinner, & Bailey, 2003). Other challenging behaviour, such as repetitive and stereotypic behaviour and social inappropriateness, were reported as beginning in early childhood (between 2 and 8 years) (Kaufmann et al., 2004; Rogers, Wehner, & Hagerman, 2001). Adolescence is a period of time associated with heightened rates of challenging behaviour (Hessel et al., 2008). Parents of adolescent males with FXS often report very high prevalence rates of stereotypy, aggression, and SIB (Sansone et al., 2012; Symons, Byiers, Raspa, Bishop, & Bailey Jr,

2010). In a two-month span, 79% of parents of adolescent males with FXS reported that their son engaged in SIB; 75% reported aggression toward others; and 98% of parents reported stereotypic behaviours (Hessl et al., 2008).

As in other populations, challenging behaviours associated with FXS are likely to be the result of difficulties in communicating and the inability to control events through conventional means. Langthorne and McGill (2012) reported that children with FXS were more likely to engage in SIB, aggression, and property destruction to escape demands or obtain tangible objects (e.g., preferred toys), than to engage in these behaviours to get someone's attention. Aggression, SIB, and stereotypic behaviours have also been associated with intense sensory stimuli (e.g., at a carnival) or unexpected changes in the physical or social environment (Hessl et al., 2008).

The majority of existing literature examining challenging behaviour in individuals with FXS has focused on specific problem behaviours based primarily on data from questionnaires and checklists. For the current study, we analysed data from semi-structured interviews conducted with parents of children with FXS. These interviews included open-ended questions that evoked detailed descriptions and examples of individuals' experiences and meanings attributed to them (Reeves, Kuper, & Hodges, 2008). These semi-structured interviews allowed participants to describe unique behaviours and situations that may not be addressed in checklists or questionnaires. This is, to our knowledge, the first study using qualitative-narrative accounts from mothers, to examine problem behaviour and the impact it has had on families of children with FXS. The inductive and interpretive process of discerning themes from narrative accounts provides an authentic portrayal of mothers' experiences with challenging behaviour.

Method

Participants

Participants were 53 biological mothers of boys ($n = 42$) and girls ($n = 11$) with full mutation FXS. Families represented a sample of convenience and were recruited from paediatricians' offices, genetics clinics, and developmental clinics across the United States. This study utilised the same sample of mothers and children as Warren and Brady in their ongoing longitudinal study of the effect of parenting on children with FXS (see Brady, Warren, Fleming, Keller, & Sterling, 2014; Warren, Brady, Sterling, Fleming, & Marquis, 2010; Warren, Brady, Fleming, & Hahn, 2017). For the present study, we analysed interviews collected when the focus child was between the ages of 8 years 11 months and 10 years 1 month ($M = 9$ years and 6 months old).

Maternal characteristics

The average age of mothers at the time of the interviews analysed for the current study was 38.8 years, with a range of 25.9–47.1 years. The majority of mothers were Caucasian (91%); two were African American, and one was Latino. Fifty-one mothers were premutation carriers and two had the full fragile X gene mutation. Families reported a wide range of income: 11% reported incomes of \$30,000 or lower and 33% reported annual income of

greater than \$100,000. The families lived in 23 different states. Maternal education ranged from 9 to 20 years, with an average of 15.5 years and 61% had a college degree. Three mothers had less than a 12th-grade education. Most of the mothers worked outside the home, either part-time or full-time (61%); and 72% were married.

Child characteristics

Children in this study had varying degrees of intellectual disability and autism symptoms. The mean mental age according to the Leiter International Performance Scale (Roid & Miller, 1997) completed at the time of assessment was 3 years and 7 months for boys in the study ($SD = .99$, range = 2.06–6.02 years) and 5 years and 3 months for girls in the study ($SD = 1.2$, range = 3.03–7.11 years). Eighteen participants (16 boys and 2 girls) had a diagnosis of autism in addition to FXS. Eleven were diagnosed by a paediatrician, six by a psychologist, and one by a developmental paediatric neurologist.

Data collection and analysis

A semi-structured interview protocol was designed by the research team, whose members had extensive experience in qualitative methodology and interviewing procedures with parents of children with disabilities. During the interviews, we asked questions that elicited information about antecedents of problem behaviours, descriptions of problem behaviours, and strategies for managing problem behaviours (see Appendix). Interviewers specifically asked mothers to describe (a) their child's temperament, (b) behaviours that were challenging or stressful, (c) situations that they believed to cause challenging behaviours, and (d) strategies used to prevent or manage challenging behaviours. Interviews were conducted by trained research assistants in families' homes, with the exception of a few families who opted to be assessed at the Frank Porter Graham Child Development Institute at the University of North Carolina at Chapel Hill. Interviews typically lasted between 60 and 75 minutes. Each interview was digitally recorded and transcribed verbatim.

An inductive data analysis process, similar to Gibbs' *Grounded Theory* approach, was used to explore the data (Gibbs, 2008). The coding process consisted of three phases: (i) open coding; (ii) axial coding; and (iii) selective coding. Coders included the first two authors and four research assistants. During the open coding phase, each coder independently read the same five interview transcripts and identified responses that related to problem behaviour. Then coders met as a group and discussed similarities and differences between these responses across the interviews. Five broad categories emerged for further analysis: (a) antecedents of problem behaviours; (b) problem behaviour descriptions; (c) consequences and general strategies surrounding problem behaviours; (d) mothers' reasoning about the possible causes of problem behaviours; and (e) mothers' feelings about the behaviours.

In the axial coding phase, the coding team identified subcategories within each main category. For example, physical aggression and defiance are subcategories of problem behaviours; and demands and social situations are subcategories of antecedents. The coding team discussed and agreed upon a thematic coding scheme with definitions and examples to describe each main category and subcategory.

In the final phase, selective coding, the first author examined links between main categories, like antecedents, behaviours, and consequences, using pivot tables in Microsoft Excel. A pivot table is a data summarisation tool that can organise, sort, and total data from an Excel spreadsheet. Information from the pivot tables was shared with the coding team. We were able to infer possible functions of some of the problem behaviours by examining antecedents and consequences that mothers reported as accompanying these behaviours. We also examined mothers' feelings related to their child's behaviour. This helped us identify behaviours and situations that were of greatest concern to mothers of children with FXS, and differentiate such behaviours from those that may have occurred frequently, but were not a major issue to the mothers.

Reliability

Weekly meetings were conducted to examine coding reliability, and, if necessary, revise the codes to fit the data. Following revisions, previously coded interviews were recoded. Thirty percent of the interviews were double coded to check for reliability. Agreement was examined for behaviour and for function. Agreement for behaviour was calculated by examining the percent agreement between the primary and reliability coder for each behaviour mentioned in the interview. Agreement for function was calculated by examining the percent agreement on the antecedent and consequence associated with each behaviour that was coded. Inter-rater reliability scores (averaged across all double-coded interviews) were 87% agreement for behaviour and 78% agreement for function. Disagreements between primary and reliability coders were discussed, and a final code was derived by consensus.

Results

In order to capture the pervasiveness of specific topographies of problem behaviour, we first present descriptive data on how many mothers reported each topography of problem behaviour. We also present descriptions of potential functions of these behaviours, and describe differences across subgroups (based on gender and autism status). Following these descriptions, we present the overarching themes that together describe how mothers viewed problem behaviours within their family life.

Common problem behaviours

Table 1 lists the 20 behaviour topographies that mothers reported in their interviews, and the number and percentage of the mothers who mentioned each behavioural topography. Data are presented separately in Table 1 for all four subgroups (i.e., boys with FXS only, boys with FXS and autism, girls with FXS only, and girls with FXS and autism). Defiance and hyperactivity or inattention, or both, were the most prevalent behaviours noted in the interviews. Other frequent problem behaviours were tantrums, stereotypy, social inappropriateness, and verbal and physical aggression.

Functions

The same behavioural topography may serve different functions for different children; and one behavioural topography can also serve multiple functions for a given child. For example,

two mothers reported different functions for their child's physical aggression. One mother said her son engaged in physical aggression to escape the demand of homework. The other mother said her son would slap her when he wanted access to the television remote control (a tangible). Table 2 provides brief definitions and an example for the four most commonly coded functions: attention, tangible, escape-avoidance, and sensory.

Defiance

One of the most prevalent problem behaviours mothers mentioned was defiance, occurring for 89% of the children. Defiance was reported frequently across all four subgroups. Defiance was defined as refusing to obey authority, for example, children who verbally say "no," or ignore repeated demands. Placing a demand on the child and not allowing access to tangibles were the most frequently cited antecedents associated with defiance. This suggests that children with FXS may engage in defiant behaviour to escape demands and to access preferred items and activities.

A few mothers described defiant behaviours that may have been triggered by physical discomfort; or avoidance of certain sensory stimuli. One mother said her son refuses to brush his teeth and another mother reported that her daughter refuses to brush her hair or wear her hair in a ponytail. Individuals who are hypersensitive to sensory input may experience pain or distress from the sensory stimulation involved in activities of daily living, such as teeth and hair brushing.

Children also engaged in defiance during transitions from preferred activities or environments. One mother explained her son's behaviour when it is time to leave his grandmother's house:

He'll either just stay in the house; he'll go slam doors. He'll stand in the yard and just sit there or just stand there. Knowing he has to go outside the house, you know, knowing he has to go to the car; he'll just stand there and wait.

She described her response to this behaviour and how it makes her feel:

Finally, I have to threaten him. I'm like "R, if I have to get out of this car, I'm going to have to call your dad." My thing is I don't spank him, so I don't know what more to do for him. He doesn't listen to me as well as he listens to [his dad].

Although defiance is common in all children, some of the examples described by mothers indicated a level of behaviour that would be highly disruptive. For example, a mother described her son's refusal to leave the beach one afternoon. She prepared him for the transition by giving him a set time they were going to leave; and she alerted him five minutes, two minutes, and one minute before departure time. When it was time to leave she said, "He laughed at first like 'Oh, I'm not gonna get up.'" She reported feeling "More frustrated and upset" as time went on;

Then he started laying on the ground, and he got obsessed over people saying goodbye to him, and he started yelling "Say goodbye to A." And I explained "You know, nobody knows who you are. They're not going to say goodbye." Of course,

that didn't register; so that just got louder. I pretty much had to drag him off of the beach.

Hyperactivity/Inattention

Forty-seven of the 53 mothers (89%) in this study reported that their child was hyperactive or had a short attention span. Twenty-seven mothers said their child was on medication to help control their inattentive or hyperactive behaviour. One mother talked about how her son could not sit still even while he was eating dinner, so she would let him run around the house while he ate. She mentioned that his running around the house did not bother her unless they were at someone else's house:

I hate family get togethers if it's not at the house here because you're taking him over to a house. You're supposed to be sitting and enjoying a meal that he's not going to sit and enjoy, and then you're letting him run all over somebody else's house. God only knows what he's going to break or what he's getting into. So it's a matter of getting up every two seconds to keep an eye on him, and it's like I can't understand why my family can't understand that that is not enjoyable.

Tantrums

For this study, a tantrum was defined as an emotional outburst or meltdown that may involve crying or spitting. Tantrums were reported by 77% of mothers; and were reported more frequently by mothers of males with FXS without ASD (80%) than by mothers of children with other sub-phenotypes. One mother stated that the best strategy for dealing with her son's attention-seeking tantrums is to ignore them: "He can scream and throw a huge fit. It can be really bad. If it gets that bad, I completely ignore him because it's that attention. The more and more you say 'stop', the more he's loving that attention."

Mothers also noted that tantrums occurred when their child was over-stimulated in public: "You're at a carnival, and it's just too over-stimulating, and he can't just run amuck everywhere, then there's going to be a meltdown." This also happened at home: "If someone comes to the door, and the dog barks, it startles him. He might have a meltdown over that." Mothers discussed strategies they used to prevent tantrums, and many said they avoided loud and crowded environments that could be over-stimulating for their child.

Children also used tantrums to escape or avoid non-preferred tasks and situations. Strategies for handling escape-maintained tantrums varied from family to family. Some mothers tried to pair a reinforcing item or activity with the non-preferred activity. For example, one mother said she would give her son Fritos on the way to and from the doctor's office to prevent tantrums. Some mothers mentioned following through with the demand even during the tantrum. For example, one mother described the following situation that occurred when her daughter had a tantrum while doing math homework:

I don't let her give up. We just power through because otherwise she just knows she can act this way and give up. So I'm like "Sit down. We're going to do this. You might as well get over it".

Other mothers allowed their children to escape demands and situations because they did not want to deal with the tantrums. Many mothers talked about avoiding doing homework with their children and avoiding or leaving certain environments and situations that their children did not like. Some mothers removed their children from public situations during tantrums due to embarrassment. One mother talked about removing her son from the grocery store when he had tantrums: “You just get so mad, and it’s embarrassing sometimes if there’s people around, you know. I would just remove him from the situation and sit him in the car.”

Stereotypy

Stereotypy was defined as repetitive or ritualistic movements, vocalisations, or postures and was reported by 60% of the mothers. Some examples of stereotypic behaviour include rocking back and forth, hand flapping, spinning, and tapping or patting an object repeatedly. Most (89%) mothers of children with comorbid diagnoses of FXS and ASD reported that their child engaged in stereotypic behaviour, while less than half (46%) of mothers of children with a single diagnosis of FXS reported stereotypic behaviour.

Most (79%) of the mothers who spoke about the function of their child’s stereotypy said their child engaged in stereotypy for sensory reasons. Some mothers said their child engaged in stereotypy to seek out additional sensory input: “Behaviour-wise the biggest issue I think is the cards, the tactile stimulation stuff. That he always has to have something small in his hands that he’s carrying around, or playing with, or sleeping with even.” Other mothers noticed stereotypic behaviour when their child was over-stimulated and may have wanted to tune out input from their surroundings.

Verbal stereotypy.—Verbal stereotypy refers to repetitive verbal behaviour or echolalia. This includes repeating words or phrases heard from another individual or a TV show, repetitively asking specific questions, or perseverating on one conversation topic. One-third of the mothers described verbal stereotypy in their child, and the majority of these mothers had sons with a diagnosis of FXS without ASD. Many of these mothers mentioned that their child perseverated on daily schedules or engaged in idiosyncratic and repetitive behaviour during social situations. One mother explained that her son had a routine set of questions he asked every person when he greeted them; and another mother stated that after meeting a new person, her son would imitate her or his voice and mannerisms. Mothers also talked about their children perseverating on routine-oriented questions. For example, one mother recounted,

Oftentimes it’s either “Where are we going? What are we doing? Where is somebody? What are we having for dinner? What’s for dinner? What’s for lunch?” He knows the answer, but he’ll still ask the question. Those are the things that he fixates on.

Twenty-five mothers described how they felt about their child’s stereotypic behaviours. About half of these mothers said that stereotypy was a major challenge or concern. One described her son’s repetitive questions as “an irritant.” She went on to say,

It's just frustrating for us because it's the same thing over and over. It's kind of like the kid in the car who says "Are we almost there yet? Are we almost there yet? Are we almost there yet?" Except he does it all the time. All the time.

Another mother talked about her frustration with her son's perseverative vocalisations: "I usually get really irritated because that sound just grates on my nerves. Just the constant 'ee.' I can't stand that noise."

Thirty-nine percent of mothers further stated that their child's stereotypic behaviour was particularly frustrating or embarrassing when exhibited in public. A mother described feeling embarrassed when her daughter's stereotypic behaviours attracted attention from strangers. She explained: "There's a sound that she makes that's a steady like 'ah.' And that is probably the one thing I wish she could stop because that really gets attention [when we] take her out. People find it unusual." She went on to talk about her daughter's stereotypic body movements:

For instance in my husband's Subaru the windows were clear, and then I noticed at a stoplight that people would stare at her cause she'd be shaking her head or something, and I didn't like it so we got our windows tinted.

Mothers said they altered their schedule and avoided certain environments and situations because of their child's stereotypic behaviour. As one mother described for her son,

He likes to lay upside down [and flap his hands]. That's just not – you know, it's challenging. We can't take him to church; we can't take him to parties. I mean, if there's a lot of people, you can't have a child – a big kid laying on the floor. It's dangerous. Like they're going to step on him or he's going to trip someone. I mean he's kicking his legs and hands in the air. It's like, you can't do that.

Another mother said that she grew up very religious, but she did not take her son to church because someone asked her to leave once.

He was loud, and someone asked us to leave, and I thought it was the rudest thing ever the way they did it, and I've never stepped foot in the church with him again. We just don't go because that stuck with me. I know not every place is going to be like that, but I think I just get like the anxiety and the nerves and the defensiveness just even thinking of walking into a church with him. I just can't do it.

Although stereotypic behaviours were challenging for most, two mothers stated that their child's stereotypy was not a major concern. As one mother explained,

The stimming out in public, I don't really find it stressful, but I want him to learn that it's not appropriate behavior out in public to "eeee" or run around in circles or something. But it's not horrible. It's kind of a minor thing, really I think.

Verbal aggression

Thirty mothers (57%) described instances of their children engaging in verbal aggression, which was defined as yelling, screaming, using inappropriate or vulgar language, or "name calling." Like verbal stereotypy, verbal aggression was most commonly reported by mothers with sons with FXS without ASD; 61% of mothers with sons with only FXS reported verbal

aggression. Eight of the 18 mothers (44%) who discussed specific antecedents of verbal aggression, noted verbal aggression occurring after their child was denied access to a desired item or activity. For example, a mother described a trip to Walmart with her son:

He had two Barbies for a birthday gift for my niece, and he saw something he wanted. And so, because I said no, he threw the Barbies in the aisle, and then threw himself down on the ground and started cussing at me, and I said, "If you don't get up I'm going to go get the manager." And he said "Go get the f-ing manager then."

She said this behaviour is very difficult to manage, especially in public.

Mothers described multiple consequences for verbal aggression. Twelve mothers said they would put their child in time-out after they engaged in verbal aggression. Some mothers tried to avoid dealing with it by avoiding certain environments and situations. One mother explained that she no longer took her son to the park, because when they would go he often got mad at other children and would scream at them.

Another mother refused to do homework with her son because of his verbal aggression. She said,

They used to send home homework until I sent it all back, and said "This is too painful." It was just too hard on everybody. M didn't want to do it. So it turned into a screaming match for half an hour until I said "What am I doing?" So I sent the folder back to school, and I said "Don't ever send me another homework because I'm not a special ed. teacher, and it's just turning into a screaming match, and I don't want to have the four or five hours that he's awake and [at] home spent this way."

Physical aggression

Twenty-nine of the 53 mothers interviewed (55%) reported that their child engaged in physical aggression, defined as hitting, kicking, pushing, biting, grabbing, pulling, scratching, spitting at, pinching, or throwing an object at another individual. Physical aggression was noted in each subgroup, but was reported most often by mothers of children with FXS + ASD. Children with FXS often engaged in physical aggression to escape demands, seek attention, and/or to attain tangibles. For example, one mother explained her son's physical aggression by saying,

If you try to stop him from doing something he wants to do, he gets mad. If you try to make him do something he doesn't want to do, he gets mad. So he's going to slap. He's going to hit. I tell him no and then I get slapped. If I'm trying to take him to the grocery store or Walmart he'll blow an absolute fit. He'll slap; he'll hit; he'll kick if he can.

Of the 29 mothers who reported that their child engaged in physical aggression, 13 (45%) provided a rationale as to why their child engaged in these behaviours. Seven mothers believed their child used physical aggression as a means to communicate. One mother explained: "He can't say 'I want this.' You know, he just can't verbalize it. So he's going to slap. He's going to hit. He's going to just bite the hell out of himself." Another mother

talked about how her child communicated verbally, but that his speech was often unintelligible: “If we can’t understand what he’s saying, he’ll continue to try. He communicates by pinching too when he’s mad or hitting. I mean that’s his communication.” Two mothers explained that their sons would push their siblings and other children when they got too close or invaded their personal space. These mothers tried to teach their sons to say, “step back” and “move” instead of pushing.

Despite recognising the potential communicative intent of physical aggression, mothers indicated that dealing with these behaviours on a daily basis was extremely difficult. As one described, “It’s been challenging to say the least. You need that break at some point. The hitting you know, I am really tired of being slapped.” Another mother also talked about needing a break when her son was physically aggressive towards her: “I usually need a time-out. Like if he’s pulling my hair and crap, I’m like I’m best to be away from you too.” Some mothers worried that the physical aggression would intensify as their child got older and stronger.

Social inappropriateness

Social inappropriateness includes social deficits such as being unaware of personal space, the inability to change conversation topic or tone based on the communication partner, and restricted or unusual social interactions. Over half of the mothers in this study (58%) reported that their child exhibited social inappropriateness.

Of the ten mothers who provided a rationale for their child’s social inappropriateness, four said the behaviour was due to communication breakdowns or the inability to fully express himself or herself. One mother said that her son did not know how to initiate play with his peers:

He doesn’t know how to say, “Hey, do you want to play four square?” or something, or if he does ask he does it in such a way they don’t know he’s asking. He’ll say, “You’re it.” Or “Tag. You’re it.” When nobody knows what’s going on he’ll just run up and “Tag. You’re it.” And that’s kind of his way I think of saying “I want to play a game. Come play with me.”

Of the nine mothers who discussed their feelings about their child’s socially inappropriate behaviour, four said it made them feel frustrated or embarrassed. One mother said she was frustrated that her son would go up and touch strangers in the grocery store. Another mother described an encounter where her daughter walked up to a woman in the grocery store and said, “How come you got a mustache?” The mother said she was “mortified” and felt like she was “going to die” of embarrassment.

Sleep problems

Thirty percent of the mothers described their child as having sleep problems, with the highest prevalence being in the subgroup of boys with FXS and ASD. Some mothers thought that their child had difficulty sleeping because of medications they were taking. One mother of a boy with FXS + ASD said that even when her son was extremely tired, he still had a difficult time sleeping.

If he's overly tired, or if we've gone out of town for the day and came back, he might stay up until 1:00 or 2:00 am. He can't go to sleep, and it's hard for us. We'll be so tired. We've been somewhere, and he'll just be wide awake at 2:00 or 3:00 in the morning. I'm like "How can you be wide awake?"

Self-Injury and elopement

Similar to sleep problems, self-injury and elopement were more commonly reported by mothers of children with dual diagnoses of FXS and ASD. Self-injury was reported by 25% of mothers in this study, but within the subgroup of 16 boys with FXS and ASD, 56% mothers described self-injury as a problem. Mothers reported that their children engaged in self-injury when people could not understand them, or when they did not get something they wanted.

Elopement was defined as running away or wandering off unsupervised. Of the 15 mothers who reported elopement, the majority (67%) had children with FXS and ASD. One mother said she put alarms on the front and back doors of her house because there were several occasions when she was asleep and her son left the house to wander through the neighbourhood. Another mother, whose son also had FXS and ASD, said that when her son was asked to do something he did not want to do, he would either "fight or flight," meaning he would either run away or become physically aggressive. She elaborated, "There's been times he's ran out onto the street. He'll get a swat on the bottom for that because it's a safety hazard."

Mothers viewed elopement and self-injury as major challenges and concerns. One mother of a boy with FXS and ASD said she always worried when she took her son out in public places because he would take off running if he saw something he wanted. She also said they had to put a chain on the front door of their house "just to make sure he doesn't take off when he decides that he wants to take off." However, this same mother said that her son's SIB worried her the most. She said he would "smash his head against the wall or the floor." When he did this she said she would restrain him so that he could not hurt himself anymore.

Discussion

Our data indicate that multiple different topographies of problem behaviour are prevalent in children with FXS at 9 years of age. Mothers in this study frequently reported behaviours such as defiance, tantrums, inattention, stereotypy, aggression, and social inappropriateness in their children, and described these as having a major negative impact on family life. Mothers had diverse approaches to dealing with problem behaviour; however, most mothers reported altering their daily routine in some way because of their child's problem behaviour. Most 9-year-old children may engage in problem behaviour on occasion, but the prevalence described in this study, as well as the impact on families, suggest that children with FXS exhibit more frequent or intense problem behaviour than what is typical for this age group.

Dual diagnoses of FXS + ASD

Not surprisingly, some behaviours were more commonly reported for children with comorbid diagnoses of FXS and ASD than for children with a single FXS diagnosis. Our

findings are consistent with previous research indicating that individuals with dual FXS + ASD diagnoses exhibit higher levels of stereotypic behaviour than individuals with a single diagnosis of FXS (Newman, Leader, Chen, & Mannion, 2015; Smith, Barker, Seltzer, Abbeduto, & Greenberg, 2012), a single diagnosis of ASD (McDuffie, Thurman, Hagerman, & Abbeduto, 2015; Smith et al., 2012), or individuals with general intellectual disabilities (Sansone et al., 2012).

Previous research regarding physical aggression and FXS suggests that physical aggression is more commonly seen in males with FXS than in females with FXS (Valdovinos, 2007). Yet, both of the mothers of girls with FXS + ASD reported that their daughters engaged in physical aggression. However, gender-specific findings should be examined with caution due to the very small number of females in our sample.

Sleep issues, self-injury, and elopement were also more common in individuals with FXS + ASD than in individuals with just FXS. These findings complement Symons et al. (2010)'s study that showed individuals with FXS who engage in self-injury often have co-occurring conditions such as sleep issues and diagnoses of ASD. The prevalence of self-injury is higher for individuals with ASD than for individuals with other IDD; and research has shown that comorbid ASD increases the likelihood of self-injury in individuals with IDD (McClintock, Hall, & Oliver, 2003; Soke et al., 2016).

Verbal stereotypy and verbal aggression were more commonly reported by mothers of children with FXS without ASD, specifically mothers of males with FXS. This could be due to the verbal nature of these behaviours. Individuals with comorbid diagnoses of FXS + ASD tend to have less sophisticated language skills than individuals with just FXS (Smith et al., 2012), so individuals with FXS + ASD may be less likely to engage in verbal stereotypy and verbal aggression because they have limited verbal abilities.

Impact on families

The majority of mothers in this study described one or more of their child's problem behaviours as a major challenge or concern. Mothers mentioned avoiding public places such as grocery stores and movie theatres, and altering their schedules because they did not want their child to cause a scene or engage in problem behaviour in public. Mothers also talked about avoiding certain demand situations, like homework, because they did not want to struggle with problem behaviours in the home.

Future directions

Based on mothers' reports of the frequency and magnitude of problem behaviours in the pre-adolescent children in this study, there is a need for further research focused on interventions that reduce the severity and frequency of problem behaviour in children with FXS. In addition to outwardly focused challenging behaviours like physical aggression, the degree of annoyance and disruption described in response to stereotypic behaviours is noteworthy. Further research on how to decrease perseverative speech and other behaviours would likely benefit many families.

It is important for clinicians and practitioners to recommend interventions aimed at preventing problem behaviour as early as possible for families of individuals with FXS. Interventions that teach adaptive and prosocial skills and reduce or prevent problem behaviour, such as positive behaviour support (PBS) and early intensive behaviour interventions (EIBIs), have been shown to be effective with children with ASD and other developmental disabilities and would be likely to benefit children with FXS as well (Carr et al., 2002; LeBlanc & Gillis, 2012). Clinicians who are knowledgeable about the impact problem behaviour has on families may be more likely to recommend these interventions as a proactive measure to families with young children with FXS. It is important to mention that a diagnostic label should not denote limited expectations for a child, but should indicate the need for individualised support that addresses behavioural issues and should contribute to stress reduction for families.

In addition to research on decreasing and preventing problem behaviour, the data presented here suggest the need for further interventions aimed at helping families cope with the challenges of raising a child with FXS. For example, strategies such as mindfulness meditation have been reported to be effective in reducing stress in parents of children with autism (Dykens, Fisher, Taylor, Lambert, & Miodrag, 2014). Parent support groups can also offer an outlet where family members may help each other by listening and offering suggestions based on personal experiences. Practitioners can help families of children with FXS by recognising the frequency of problem behaviour and understanding the families' stresses and coping mechanisms. Practitioners who are aware of the challenges families face will likely be better able to communicate and collaborate with families to form effective treatment plans.

In conclusion, the results of this qualitative study elucidate the pervasiveness of problem behaviour in children with FXS, and the substantial impact it has on families. There is a need for more research on supporting family members and treating problem behaviour in children with FXS. Effective treatments are likely to combine the behavioural approaches described above with pharmacological treatments aimed at treating conditions such as anxiety or sleep disorders that contribute to problem behaviour. Reducing or preventing problem behaviour appears to be of paramount importance for improving family functioning and long-term outcomes for children with FXS.

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Appendix

Semi-Structured Interview Questions about Problem Behaviour

1. You've brought up some interesting topics that I'd like to ask you more about. [Ask any follow-up questions about specific topics]:
 - a. Child's personality or temperament
 - b. Behaviour around strangers
 - c. Behaviour in new situations or settings
 - d. Interactions with other children
 - e. Things that make the child angry or frustrated, and how mother responds in these situations
 - f. Focus and attention at home and school
2. Does (child) sometimes behave in ways that you find challenging or stressful? If so, what are these behaviours?
3. How do you respond when this happens [ask for each behaviour mentioned]? What are the most effective strategies you use for dealing with this behaviour?
4. Are there any strategies that don't work at all for (child)?
5. Are there other behaviours or situations that call for disciplining the child?
6. How do you discipline him/her for these?
7. What do you find to be the most effective discipline strategy?

References

- Bailey DB, Raspa M, Bishop E, & Holiday D (2009). No change in the age of diagnosis for fragile X syndrome: Findings from a national parent survey. *Pediatrics*, 124(2), 527–533. [PubMed: 19581269]
- Bailey DB, Raspa M, Olmsted M, & Holiday DB (2008). Co-occurring conditions associated with FMR1 gene variations: Findings from a national parent survey. *American Journal of Medical Genetics Part A*, 146(16), 2060–2069.
- Brady N, Warren S, Fleming K, Keller J, & Sterling A (2014). Effect of sustained maternal responsivity on later vocabulary development in children with fragile X syndrome. *Journal of Speech Language and Hearing Research*, 57, 212–226.
- Carr EG, Dunlap G, Horner RH, Koegel RL, Turnbull A, Sailor W, ... Fox L (2002). Positive behavior support. Evolution of an applied science. *Journal of Positive Behavior Interventions*, 4(1), 4–16. Copyright (2002) by PRO-ED, Inc. Adapted with permission.
- Christensen DL, Baio J, Braun KV, Bilder D, Charles J, Constantino JN, ... Yeargin-Allsopp M (2016). Prevalence and characteristics of autism spectrum disorder among children aged 8 years – autism and developmental disabilities monitoring network, 11 sites, United States, 2012. *Morbidity and Mortality Weekly Report Surveillance Summaries*, 65(3), 1–23.
- Crawford DC, Acuna JM, & Sherman SL (2001). FMR1 and the fragile X syndrome: Human genome epidemiology review. *Genetics in Medicine*, 3, 359–371. [PubMed: 11545690]
- Demark JL, Feldman MA, & Holden JJA (2003). Behavioral relationship between autism and fragile X syndrome. *American Journal on Mental Retardation*, 108(5), 314–326. doi: 10.1352/0895-8017(2003)108<314:BRBAAF>2.0.CO;2 [PubMed: 12901707]
- Duy PQ, & Budimirovic DB (2017). Fragile X syndrome: Lessons learned from the most translated neurodevelopmental disorder in clinical trials. *Translational Neuroscience*, 8(1), 7–8. [PubMed: 28400977]

- Dykens EM, Fisher MH, Taylor JL, Lambert W, & Miodrag N (2014). Reducing distress in mothers of children with autism and other disabilities: A randomized trial. *Pediatrics*, 134, e454–e463. [PubMed: 25049350]
- Eisenhower A, Baker B, & Blacher J (2005). Preschool children with intellectual disability: Syndrome specificity, behaviour problems, and maternal well-being. *Journal of Intellectual Disability Research*, 49(9), 657–671. [PubMed: 16108983]
- Emerson E, Kierman C, Alborz A, Reeves D, Mason H, Swarbrick R, ... Hatton C (2001). The prevalence of challenging behaviors: A total population study. *Research in Developmental Disabilities*, 22(1), 77–93. [PubMed: 11263632]
- Fernandez-Carvajal I, Posadas BL, Pan R, Raske C, Hagerman PJ, & Tassone F (2009). Expansion of an FMR1 grey-zone allele to a full mutation in two generations. *The Journal of Molecular Diagnostics*, 11(4), 306–310. [PubMed: 19525339]
- Gibbs GR (2008). *Analyzing qualitative data*. London: Sage.
- Hastings R (2002). Parental stress and behaviour problems of children with developmental disability. *Journal of Intellectual and Developmental Disability*, 27, 149–160.
- Hatton DD, Hooper SR, Bailey DB, Skinner ML, Sullivan KM, & Wheeler A (2002). Problem behavior in boys with fragile X syndrome. *American Journal of Medical Genetics Part A*, 108(2), 105–116.
- Hessl D, Dyer-Friedman J, Glaser B, Wisbeck J, Barajas G, Taylor A, & Reiss A (2001). The influence of environmental and genetic factors on behavior problems and autistic symptoms in boys and girls with fragile X syndrome. *Pediatrics*, 108(5), e88. [PubMed: 11694672]
- Hessl D, Tassone F, Cordeiro L, Koldewyn K, McCormick C, Green C, ... Hagerman R (2008). Brief report: Aggression and stereotypic behavior in males with fragile X syndrome – moderating secondary genes in a “single gene” disorder. *Journal of Autism and Developmental Disorders*, 38, 184–189. [PubMed: 17340199]
- Jessor R (1982). Problem behavior and developmental transition in adolescence. *Journal of School Health*, 52(5), 295–300. [PubMed: 6919706]
- Kau AS, Tierney E, Bukelis I, Stump MH, Kates WR, Trescher WH, & Kaufmann WE (2004). Social behavior profile in young males with fragile X syndrome: Characteristics and specificity. *American Journal of Medical Genetics Part A*, 126(1), 9–17.
- Kaufmann WE, & Reiss AL (1999). Molecular and cellular genetics of fragile X syndrome. *American Journal of Medical Genetics Part A*, 88(1), 11–24.
- Kaufmann WE, Cortell R, Kau AS, Bukelis I, Tierney E, Gray RM, ... Stanard P (2004). Autism spectrum disorder in fragile X syndrome: Communication, social interaction, and specific behaviors. *American Journal of Medical Genetics Part A*, 129(3), 225–234.
- Kiernan C, & Kiernan D (1994). Challenging behaviour in schools for pupils with severe learning difficulties. *Mental Handicap Research*, 7(3), 177–201.
- Langthorne P, & McGill P (2012). An indirect examination of the function of problem behavior associated with fragile X syndrome and smith-magenis syndrome. *Journal of Autism and Developmental Disorders*, 42, 201–209. [PubMed: 21442360]
- LeBlanc LA, & Gillis JM (2012). Behavioral interventions for children with autism spectrum disorders. *Pediatric Clinics of North America*, 59(1), 147–164. [PubMed: 22284799]
- Maddalena A, Richards CS, McGinniss MJ, Brothman A, Desnick RJ, Grier RE, ... Watson M (2001). Technical standards and guidelines for fragile X: The first of a series of disease-specific supplements to the standards and guidelines for clinical genetics laboratories of the American College of Medical Genetics. *Genetics in Medicine*, 3(3), 200–205. [PubMed: 11388762]
- McClintock K, Hall S, & Oliver C (2003). Risk markers associated with challenging behaviours in people with intellectual disabilities: A meta-analytic study. *Journal of Intellectual Disability Research*, 47(6), 405–416. [PubMed: 12919191]
- McDuffie A, Thurman AJ, Hagerman RJ, & Abbeduto L (2015). Symptoms of autism in males with fragile X syndrome: A comparison to nonsyndromic ASD using current ADI-R scores. *Journal of Autism and Developmental Disorders*, 45(7), 1925–1937. [PubMed: 24414079]

- Newman I, Leader G, Chen JL, & Mannion A (2015). An analysis of challenging behavior, comorbid psychopathology, and attention-deficit/hyperactivity disorder in fragile X syndrome. *Research in Developmental Disabilities*, 38, 7–17. [PubMed: 25543996]
- Reeves S, Kuper A, & Hodges BD (2008). Qualitative research methodologies: Ethnography. *BMJ: British Medical Journal*, 337, a1020. [PubMed: 18687725]
- Rogers SJ, Wehner EA, & Hagerman R (2001). The behavioral phenotype in fragile X: Symptoms of autism in very young children with fragile X syndrome, idiopathic autism, and other developmental disorders. *Journal of developmental & behavioral pediatrics*, 22(6), 409–417. [PubMed: 11773805]
- Roid GH, & Miller LJ (1997). *Leiter International Performance Scale-Revised (Leiter-R) manual*. Wood Dale, IL: Stoelting.
- Rusch R, Hall J, & Griffin H (1986). Abuse-provoking characteristics of institutionalized mentally retarded individuals. *American Journal of Mental Deficiency*, 90(6), 618–624. [PubMed: 3717218]
- Sansone SM, Widaman KF, Hall SS, Reiss AL, Lightbody A, Kaufmann WE, ... Hessl D (2012). Psychometric study of the aberrant behavior checklist in fragile X syndrome and implications for targeted treatment. *Journal of Autism and Developmental Disorders*, 42(7), 1377–1392. [PubMed: 21972117]
- Schroeder SR, Marquis JG, Reese RM, Richman DM, Mayo-Ortega L, Oyama-Ganiko R, ... Lawrence L (2014). Risk factors for self-injury, aggression, and stereotyped behavior among young children at risk for intellectual and developmental disabilities. *American Journal on Intellectual and Developmental Disabilities*, 119(4), 351–370. [PubMed: 25007299]
- Smith L, Barker E, Seltzer M, Abbeduto M, & Greenberg J (2012). Behavioral phenotype of fragile X syndrome in adolescence and adulthood. *American Journal on Intellectual and Developmental Disabilities*, 117(1), 1–17. [PubMed: 22264109]
- Soke GN, Rosenberg SA, Hamman RF, Fingerlin T, Robinson C, Carpenter L, ... DiGuseppi C (2016). Brief report: Prevalence of self-injurious behaviors among children with autism spectrum disorder—a population-based study. *Journal of Autism and Developmental Disorders*, 46(11), 3607–3614. [PubMed: 27565654]
- Symons FJ, Byiers BJ, Raspa M, Bishop E, & Bailey DB Jr (2010). Self-injurious behavior and fragile X syndrome: Findings from the national fragile X survey. *American Journal on Intellectual and Developmental Disabilities*, 115 (6), 473–481. [PubMed: 20946000]
- Symons FJ, Clark RD, Hatton DD, Skinner M, & Bailey DB (2003). Self-injurious behavior in young boys with fragile X syndrome. *American Journal of Medical Genetics Part A*, 118(2), 115–121.
- Valdovinos M (2007). Brief review of current research in FXS: Implications for treatment with psychotropic medication. *Research in Developmental Disabilities*, 28, 539–545. [PubMed: 17034990]
- Warren S, Brady N, Sterling A, Fleming K, & Marquis J (2010). Maternal responsivity predicts language development in young children with fragile X syndrome. *American Journal on Intellectual and Developmental Disabilities*, 115, 54–75. [PubMed: 20025359]
- Warren SF, Brady N, Fleming KK, & Hahn LJ (2017). The longitudinal effects of parenting on adaptive behavior in children with fragile X syndrome. *Journal of Autism and Developmental Disorders*, 47(3), 768–784. doi:10.1007/s10803-016-2999-7 [PubMed: 28074357]

Table 1.

Occurrence of problem behaviour by gender and ASD diagnosis.

Behavioural Topography	Occurrence in males with FXS without ASD (n = 26)	Occurrence in males with FXS and ASD (n = 16)	Occurrence in females with FXS without ASD (n = 9)	Occurrence in females with FXS and ASD (n = 2)
Defiance	23 (88%)	14 (87%)	9 (100%)	1 (50%)
Hyperactivity/Inattention	26 (100%)	14 (87%)	7 (78%)	0 (0%)
Tantrum	21 (80%)	12 (75%)	6 (67%)	2 (100%)
Stereotypy	13 (50%)	14 (87%)	3 (33%)	2 (100%)
Social Inappropriateness	17 (65%)	9 (56%)	4 (44%)	1 (50%)
Verbal Aggression	16 (61%)	9 (56%)	4 (44%)	1 (50%)
Physical Aggression	14 (53%)	11 (68%)	2 (22%)	2 (100%)
Anxiety	14 (53%)	6 (37%)	4 (44%)	1 (50%)
Lack of Personal Safety	9 (34%)	6 (37%)	3 (33%)	1 (50%)
Verbal Stereotypy	14 (53%)	3 (18%)	1 (11%)	0 (0%)
Incontinence	7 (26%)	8 (50%)	0 (0%)	1 (50%)
Sleep Issues	2 (7%)	10 (62%)	3 (33%)	1 (50%)
Elopement	5 (19%)	9 (56%)	0 (0%)	1 (50%)
Clingy	6 (23%)	2 (12%)	4 (44%)	1 (50%)
Self-Injury	3 (11%)	9 (56%)	0 (0%)	1 (50%)
Property Destruction	6 (23%)	6 (37%)	0 (0%)	0 (0%)
Overeating	2 (7%)	4 (25%)	0 (0%)	0 (0%)
Stealing	1 (3%)	4 (25%)	0 (0%)	0 (0%)
Vomiting	1 (3%)	2 (12%)	0 (0%)	0 (0%)
Undressing	2 (7%)	1 (6%)	0 (0%)	0 (0%)

Table 2.

Functions of problem behaviour.

Function	Description
Attention	Child engages in behaviour to get attention from adult or peers (e.g., "He'll come up and scratch me, and he's trying to get me to react.")
Tangible	Child engages in behaviour to gain access to a toy or activity (e.g., "She wanted a specific toy from the store. I told her we couldn't get it today, and she sat there and cried and started throwing a tantrum.")
Escape/ Avoidance	Child engages in behaviour to escape demands or avoid social attention (e.g., "If I tell him to do his homework he'll start hitting the walls and kicking stuff.")
Sensory	Child engages in behaviour to access a pleasing sensation, or the child engages in behaviour to remove a sensation that is displeasing (e.g., "If it's a real congested setting and he gets hyper-aroused, he'll do a lot of hand biting, a lot of leg-hitting.")

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