The Expressed Emotion of Mothers of Children with Fragile X Syndrome

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THE EXPRESSED EMOTION OF MOTHERS
OF CHILDREN WITH FRAGILE X SYNDROME

A Dissertation
Presented to
the Morgridge College of Education
University of Denver

In Partial Fulfillment
of the Requirements for the Degree
Doctor of Philosophy

by
Jeanine M. Coleman
March 2010
Advisor: Dr. Karen Riley
Abstract

A review of the literature revealed that mothers with children with fragile X syndrome (FXS) face many challenges, including managing their child’s behavior which is a defining characteristic of children with FXS (Epstein, Riley, & Sobesky, 2002). Parents and professionals have become increasingly aware through research and everyday experiences that children with FXS are often overly sensitive to multiple stimuli (Hagerman, 1999; Hagerman & Hagerman, 2002; Miller et al., 1999). This overstimulation often leads to behavioral challenges that are characterized by tantrums, angry outbursts, and other forms of aggression. Additionally, mothers of children with FXS have their own personal challenges that come with carrying the premutation of the FMR1 gene. Many factors associated with this maternal genetic status include a heightened risk for premature ovarian insufficiency (POI), compromised self-concepts, and the possibility of giving birth to another child with FXS (Sherman, 2002). The purpose of this study is to explore the relationship between the expressed emotion of mothers of children with FXS and the behavioral and sensory characteristics of their children. Expressed Emotion is a construct of the familial emotional climate in which a parent (or close relative) expresses emotional involvement, hostility and/or criticism about their child (McCarty & Weisz, 2002). The primary way to measure expressed emotion is the Five Minute Speech Sample (FMSS) (Magana-Amato, 1993) which is coded through a non-scripted monologue by one of the parents. Adapted from a longer
interview process, the Camberwell Family Interview (CFI), FMSS is coded for content and tone of the parents’ speech sample for criticisms, emotional over-involvement, relationship status, and positive expressions (Wamboldt, O'Connor, Wamboldt, Gavin, & Klinnert, 2000). The FMSS provides a novel way of looking at the mother-child dyad in the FXS community and provide information to guide intervention for new mothers with young children with FXS. Without pathologizing the mothers of children with FXS, the findings of this study provides insight into the relationship between the expressed emotion of mothers and the behavioral and sensory characteristics of children with FXS that could lead to valuable intervention strategies developed by parents and professionals.
Acknowledgements

First, I would like to thank my advisor, Dr. Karen Riley for her support and encouragement throughout my doctoral program. She has been there to guide and mentor me through the entire process with understanding and thoughtful advice. Dr. Riley is responsible for introducing me to the families and children that have fragile X syndrome (FXS) and the researchers and therapists that work with these families. It has been a wonderful experience learning and growing from the families and professionals involved in FXS research. I would also like to thank other influential professors that have guided my work in child development and early childhood special education. Drs. Gloria Miller and Ginger Maloney have given me much needed support in times when I needed it most during the dissertation process. They were always been there to encourage me to continue and do my best work. A special thank you to Dr. Toni Linder who encouraged me to enter the doctoral program and taught me about Transdisciplinary Play-Based Assessment and intervention and constantly challenged my learning through questioning and guidance. She presented many opportunities to expand my experience and knowledge of the field.

Lastly, I would like to express my deepest gratitude and love to my husband Dr. Darrin Hicks and our daughter Grady for their continued support and encouragement. Thank you, Darrin for being there to talk through my thoughts, challenge me, and taking care of me and Grady. Grady, thank you for always smiling, asking how things were going, and giving me a big hug after a hard day. I could not have done it without the both of you.
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Chapter One: Introduction and Literature Review

Mothers experience a unique, complex, and incomparable relationship with their children from the moment they realize they are pregnant. Mothers’ expectations and hopes for their unborn children are intense and omnipresent. For mothers who give birth to children with special needs those expectations and hopes are often disrupted when their babies are born. Approximately 120,000 (one in thirty-three) babies are born with birth defects in the United States every year (March of Dimes, 2008). Both biological and environmental risk factors contribute to birth defects and congenital disabilities; although in many instances, specific causes may be unknown. Physicians can identify and begin treatment for some disabilities and disorders immediately after birth, limiting the occurrence of secondary disabilities (McDevitt & Ormrod, 2007). Less obvious disorders related to behavior, feeding, or self-regulation may go undiagnosed, initially resulting in stress for families as they struggle to understand and meet the unique needs of their child. These disorders can manifest in issues with behavioral ramifications that impact the family relationships and routines. In particular, children with fragile X syndrome (FXS), an inherited genetic disorder, are often undiagnosed for several years after birth, although subtle indicators appear much earlier (Hagerman, 2006b). These indicators cluster around challenging behaviors associated with hyperactivity, aggression, language delays, and sensory processing problems that cause additional stress for families.
(Bailey Jr, Roberts, Mirrett, & Hatton, 2001; Baranek et al., 2008; Dyer-Friedman et al., 2002).

The demands of caring for a typically developing child can be stressful under the best of circumstances, but parenting a child with significant intellectual disabilities such as FXS, can lead to excessive worry and unrealistic expectations (Weiss, Sullivan, & Diamond, 2003). Additionally, levels of support for children and adults with FXS range from intermittent to extensive, depending on the severity of the disability and the community in which they live, compounding family stress. This stress has a direct impact on mothers’ abilities to express warmth and care for their children, regardless of their feelings of love for their children (Bailey, Skinner, & Sparkman, 2003; Boger, Tompson, Pavlis, Briggs-Gowan, & Carter, 2008; C. Johnston et al., 2003). Maternal warmth and maternal stress are, in fact, two sides of the same coin each having a profound effect on children’s experiences. Maternal warmth, a protective factor, is predictive of increased social competence and secure attachment (Jennings et al., 2008). Maternal stress, on the other hand, is a risk factor that has been linked to behavior problems, inadequate fetal brain development, and childhood obesity (G. Brown, J. Birley, & J. Wing, 1972; Davies, 2004; J. P. Shonkoff & Phillips, 2000). Maternal stress can have significant influence on the emotional relationship between a mother and her child. The interaction between maternal stress and lack of maternal warmth is fundamental to the innovative construct of emotional family climate, called expressed emotion (Vostanis & Leff, 1995). Combining the factors of emotional family climate and parenting a child with FXS is what led to the purpose of this dissertation.
Operational Definitions

The two primary operational definitions for this dissertation include expressed emotion and fragile X syndrome (FXS). Operational definitions are needed to explain the complexities of these two factors. Expressed emotion will be defined first, followed by FXS.

Expressed emotion.

Expressed emotion is a complex construct that focuses on parents’ verbal and nonverbal expressions about their children and the emotional quality of the relationship (G. Brown et al., 1972; Calam & Peters, 2006; McCarty & Weisz, 2002b; St. Jonn-Seed & Weiss, 2002). This construct was first used with families with adult children with mental health disorders. Expression of criticisms, excessive worry, and dissatisfaction of family members’ behavior are primary elements of expressed emotion (Magana-Amato, 1993; McCarty & Weisz, 2002b). Brown, Birley, and Wing (1972) conducted a seminal study and identified several affective variables that became important components of expressed emotion. The authors argued that these components, which were associated with symptomatic relapse of psychiatric disorders, were based on six components based on relatives’ expressed thoughts and feelings about a patient with schizophrenia: 1) critical comments; 2) hostility; 3) dissatisfaction; 4) warmth; 5) emotional over-involvement; and 6) an overall index of the relative’s expressed emotion. The affective variables, warmth and emotional over-involvement, were positively associated, although emotional over-involvement was found to show a curvilinear relationship to criticism and hostility. This implies that the relatives who were rated high or low on emotional over-involvement also expressed the most criticisms or hostility toward the patient, supporting
the idea that expressed emotion can be either a risk factor for families or a protective factor. The salient issue of expressed emotion is the balance of the affective variables (G. Brown & Rutter, 1966). For the purposes of this dissertation, “expressed emotion” will represent the construct, while the abbreviation, “EE” will represent the ratings from the measurement of expressed emotion.

**Fragile X syndrome (FXS).**

As mentioned above, FXS is the most common cause of inherited intellectual disabilities. This claim has been supported by a body of research that has been conducted over the past 25 years (K. Cornish, Turk, & Hagerman, 2008). Hagerman (2006) states, “Fragile X refers to a family of medical and developmental problems related to an expansion of the trinucleotide repeats (CGG) on the front end of the fragile X mental retardations 1 (*FMR1*) gene. This gene is located at the bottom end of the X chromosome at Xq27.3. The *FMR1* gene was sequenced in 1991 by an international consortium (Verkerk et al., 1991)” (p. 2). More recently, the understanding of FXS has evolved and is viewed as a “continuum of gene effects” resulting in a spectrum of disorders (McConkie-Rosell et al., 2005).

FXS manifests in many behavioral, learning, and sensory challenges for the diagnosed individual and his/her family. One of the most difficult challenges to address with children with FXS is aggression, which often times stems from anxiety and sensory overstimulation (K. Cornish et al., 2008; Dixon Weber, 2000). Aggressive behaviors such as hitting, biting, pushing, and self-injurious behaviors may result from exposure to unfamiliar or unpredictable experiences (Dixon Weber, 2000; Hatton et al., 2002; Symons, Clark, Hatton, Skinner, & Bailey, 2003). There are also a number of health
challenges related to FXS that include seizures, connective tissue dysplasia, vision, and cardiac problems (Hagerman, 2006a). More detail about FXS and expressed emotion will be addressed in the literature review.

The primary purpose of this dissertation is to explore the association between the expressed emotion of mothers of children with FXS and the potential impact on their child’s behavior. The following sections will lay the foundation for the research problem, the significance of this dissertation research, and further definition of expressed emotion and fragile X syndrome.

**Statement of Problem & Significance of Study**

Families with children with fragile X syndrome (FXS) experience severe maternal and family stressors (Hagerman, Rivera, & Hagerman, 2008). Stress, emanating from information and resource needs, arises because there is limited public awareness of FXS, and diagnosis may be delayed until the child is 3-4 years old (Bailey, 2004; K. Cornish et al., 2008; Guralnick, 1998; R. J. Hagerman, 2002; Hagerman, 2006d; Hagerman et al., 2009a; Skinner, Sparkman, & Bailey, 2003). Prior to diagnosis, families may have experienced unsettling behaviors in their children, which may have led to developmental concerns and misdiagnoses. The time between the initial concerns and the eventual FXS diagnosis is extremely stressful for families because of causes and effective treatment options may be unknown. Formal diagnosis may generate new stressors related to family history and family planning (Bailey, 2004; Bailey, Skinner, Hatton, & Roberts, 2000; Bailey et al., 2003).

Children with FXS often have many developmental and behavioral challenges that affect the family dynamics. Parents may experience interpersonal distress, in
addition to the lack of information and resources, due to the challenging behaviors of their children (K. Cornish et al., 2008; Epstein, Riley, & Sobesky, 2002a; Hagerman, 2006a; Hatton et al., 2002). Significant health conditions also create considerable emotional and financial family stress (Dixon Weber, 2000; R. J. Hagerman, 2002). Additionally, parents of children with FXS routinely report that their children have behavior problems, such as excessive tantrums and feeding and sleeping difficulties, beginning in the toddler and preschool years (Hagerman, 2006e; Hatton et al., 2002).

Although sustained, consistent, and comprehensive interventions produce positive outcomes for children with FXS and their families, the road to realizing the benefits is a long and emotional one for all involved (Guralnick, 1998; Hagerman et al., 2009b; C. Johnston et al., 2003). Coping with the FXS diagnosis and coordinating services, compounds stress for parents and may contribute to a lack of confidence in their ability to care for their children. Guralnick (1998) reports, “The long-term well-being of the child and family is associated with the family’s ability to maintain a sense of mastery and control over decision-making and to do so with reasonable competence and confidence” (p. 22). The interaction of these variables greatly influences the emotional family climate, which is the basic tenet of the construct, expressed emotion (Calam & Peters, 2006).

While much is known about the effects of expressed emotion on a portion of the population with mental health disorders (Baker, Heller, & Henker, 2000; Brennan, Brocque, & Hammen, 2003; G. Brown et al., 1972; Daley, Sonuga-Barke, & Thompson, 2003; Peris & Baker, 2000), research has yet to explore the nature of expressed emotion with mothers of children with FXS. This lack of research results in a void of information that could inform parents of children with FXS, researchers in the field of expressed
emotion, as well as teachers, therapists, and other providers working with families with children with FXS. It is important to explore how mothers of children with FXS handle the stressors and emotional aspects of raising their children, as well as how they express their emotions about their children. This information can lead to important interventions and modifications that can help parents balance their expressed emotion and parenting behaviors.

Mothers of children with FXS have many stressors including raising children with significant intellectual disabilities, the emotional burden of passing on the gene mutation to their children, and a predisposition to psychological issues such as social anxiety and depression (Hagerman et al., 2008; McConkie-Rosell et al., 2005). This information, coupled with the research on expressed emotion led to research questions about the quality of relationships between mothers and their children with FXS and subsequent influences on the children’s behaviors. For these reasons, mothers of children with FXS are an important group to study in relation to expressed emotion and child outcomes.

**Purpose of Study**

The purpose of this study is to explore the relationship between the expressed emotion of mothers of children with FXS and the behavioral and sensory characteristics of their children. The determination of a cause/effect relationship between the factors is not the purpose of this study; rather, it is the establishment of a relationship between the mothers’ expressed emotion and the children’s behavioral and sensory challenges, while controlling for confounding variables, such as age, gender, and comorbid diagnoses. The existence of a relationship between these two factors can lead to intervention strategies for mothers and their children with FXS. These findings may also foster discussions
regarding early identification of FXS given that most children with FXS are not
diagnosed until 3-4 years of age (Bailey et al., 2000; Bailey et al., 2003; Guralnick,
1998). Contrary to the experiences of families with children with FXS, families with
children with Down syndrome do not experience a delay in diagnosis due to prenatal
screenings and noticeable dysmorphic characteristic at birth, resulting in less stress and
more support services in early stages for these families (Poehlmann, Clements, Abbeduto,
& Farsad, 2005).

**Research Questions**

1. What is the relationship between the expressed emotion (EE) of mothers of
   children with FXS and their perception of their children’s behavioral
   challenges?
   
   a. Expressed emotion will be measured by the Five Minute Speech
      Sample (FMSS) completed by the mother (Magana-Amato, 1993).
   
   b. Perception of children’s behavior will be measured by the Behavior
      Assessment System for Children (BASC) completed by the mother
      (Kamphaus & Reynolds, 1998).

2. What is the relationship between the expressed emotion of mothers of children
   with FXS and their perception of their children’s sensory processing?
   
   a. The perception of the children’s sensory processing will be measured
      by the Sensory Profile (Dunn, 1999) completed by the mother.

**Literature Review**

The literature review for this dissertation was organized into three sections. The
first section describes a profile of children with fragile X syndrome beginning with the
etiology of FXS, followed by the physical and behavioral phenotype. The second section describes the characteristics and challenges of mothers of children with fragile X syndrome. The third section describes the variables that influence the relationship between mothers and their children with fragile X syndrome, primarily through expressed emotion.

A profile of children with fragile X syndrome.

Etiology of fragile X syndrome.

Fragile X syndrome (FXS) is the leading cause of inherited intellectual disabilities and a complex genetic disorder that affects many children and families (R. J. Hagerman, 2002; Hagerman, 2006a; Hagerman et al., 2008). The inheritance aspect of FXS is salient to this discussion because of the multigenerational influences on families that include children, parents, and grandparents. Fragile X syndrome disorders are caused by a fragile X mental retardation 1 gene (FMR1) mutation, which is passed on through generations, and expressed as a full mutation, premutation, or intermediate/gray zone overlapping the normal and premutation (Grigsby et al., 2008; Hagerman et al., 2008; McConkie-Rosell et al., 2005). Disorders deriving from the FMR1 mutation affecting those with the premutation and full mutation; include “fragile X syndrome, fragile X-associated tremor/ataxia syndrome, fragile X-associated premature ovarian failure (now called premature ovarian insufficiency), and psychiatric problems including Autism, Autism spectrum disorders (ASD) associated with both the premutation and full mutation” (Hagerman et al., 2008, p. 1). A majority of males with the full mutation have intellectual disabilities (i.e. mental retardation), while learning disabilities, social anxiety,
and selective mutism are primary effects in females with the full mutation (Hagerman, 2006a; McConkie-Rosell et al., 2005).

The full mutation typically results in the methylation of the gene causing a lack of essential protein to be produced, which causes FXS (Anido, Carlson, Taft, & Sherman, 2005; Hagerman, 2006a). “The methylation decreases the production of messenger RNA (mRNA), a copy of the gene that is normally converted into the protein product of the gene, the FMRI protein (FMRP)” (p. 2). It is the lack of essential protein in the brain (FMRP) that creates challenges and disrupts cognitive, language, and physical functioning required in everyday life (R. J. Hagerman, 2002; Hagerman et al., 2008; Irwin, Galvez, Weiler, Beckel-Mitchener, & Greenough, 2002; Sherman, 2002).

Beginning with a historical perspective, Martin and Bell (1943) first identified the FXS phenotype by noticing a differentiated form of mental retardation (Hagerman, 1999a; Martin & Bell, 1943; Sherman, 2002). These two scientists recognized a lineage of family members with several males who had severe intellectual disabilities and a fewer number of females with mild intellectual disabilities. The Martin-Bell phenotype, as it was previously called, was distinguished by the dysmorphic characteristics that are now familiar to FXS and the cytogenetic marker identified in 1982 by Richards and Webb (Sherman, 2002).

Martin and Bell (1948) paved the way for other scientists to explore the complexities of a FXS diagnosis. In 1991, scientists identified the gene responsible for FXS, and a trinucleotide gene expansion sequence, Cytosine, Guanine, Guanine, (CGG) (W. T. Brown, 2002; P. J. Hagerman, 2002; Sherman, 2002; Verkerk et al., 1991). Within the general population, individuals typically have a CGG repeat sequence ranging
from approximately 6-50, and no evidence of the \textit{FMR1} gene mutation (McConkie-Rosell et al., 2005). Individuals with a repeat size of approximately 45-60 are in the intermediate or gray zone which captures the “overlap junction between the normal and premutation ranges” (McConkie-Rosell et al., 2005). Individuals with 55-200 repeats are carriers of the \textit{FMR1} gene mutation, can be female or male, and have historically thought to be unaffected (W. T. Brown, 2002; McConkie-Rosell et al., 2005). Finally, additional studies suggest that individuals with 200 or more CGG repeats have the full mutation of FXS and typically display intellectual disabilities. The \textit{FMR1} gene mutation expands through the female carrier, typically resulting in a full mutation of their offspring, but does not expand through males. Figure 1 illustrates the variability of the CGG repeat sequence across the population (Hagerman, 2006a; Hagerman et al., 2008).

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{variability_of_cgg_rep.png}
\caption{Variability of CGG Repeat Sequence}
\end{figure}

Fragile X syndrome affects males and females differently, although the full mutation is associated with both genders (McConkie-Rosell et al., 2005). Individuals with a full mutation do not produce the FMRP required for typical intellectual functioning. For males, who have an X and Y chromosomes, “the cognitive phenotype is characterized by a spectrum of features including developmental delay in the young child, mental retardation from mild to severe, borderline IQ, and learning disabilities” (McConkie-Roell et al., 2005, p. 253). Females affected by the FXS full mutation may experience less severe symptoms than their counterparts, typified by learning disabilities.
and social anxiety (Hagerman et al., 2008). This difference between males and females occurs because females have two X chromosomes that can produce FMRP (Abrams, 2007; Hagerman & Hagerman, 2008; Sherman, 2002). Within each cell, females have two X chromosomes, however, only one X is randomly activated to produce FMRP. “The activation ratio is the ratio of cells with the premutation X turned on to those with the normal X turned on. If you have a low activation ratio that means that you have more of the premutation Xs turned on and thus fewer of the normal Xs activated” (Abrams, 2007, p. 16). In addition, Hagerman (2006a) reports that “females with a favorable activation ratio (the majority of their cells have the normal X as the active X) will have a higher IQ” (p.3).

Fragile X syndrome accounts for 2-3% of all forms of mental retardation and 30% of all forms of X-linked mental retardation. Although epidemiological studies vary by location (Loesch et al., 2003b), research shows that FXS has a prevalence of 1 in 3600 males and approximately 1 in 4000-6000 females (Bailey et al., 2000; Bailey et al., 2003; P. J. Hagerman, 2002; Hagerman, 1999b, 2006b, 2006c; Hagerman & Hagerman, 2002; Hagerman et al., 2008; Meyer & Batshaw, 2002; Mirrett, Bailey, Roberts, & Hatton, 2004; Sherman, 2002; Weber, 2000a). The prevalence of the premutation (55-200 CGG repeats) is greater, affecting one per 130-260 females and one per 300-800 males (Hagerman et al., 2008).

**Physical profile of children with FXS.**

Common characteristics associated with FXS include a diverse range of physical features that develop as the child ages. Generally, the distinctive physical characteristics refer to males with FXS because studies have documented more pronounced physical
features in males than females (Hagerman, 2006a). Physical features include a long face, prominent ears, velvet-like skin, macroorchidism (large testicles), hyperextensible finger joints, a long narrow face, and hypotonia. (Hagerman et al., 2008) Some physical features, such as macroorchidism and prominent ears, may not appear until the onset of puberty (R. J. Hagerman, 2002; Hagerman et al., 2008).

The physical features of FXS are often unrecognizable at birth; therefore, as a result many children are not diagnosed with FXS until they are well into toddlerhood (24-36 months). This is often after many months of parental worry and concern as developmental delays or concerns emerge during the toddler years (Bailey, 2004). Retrospective and detailed research on those diagnosed in infancy have found that infants with FXS have greater head circumference and hypotonia (Baranek et al., 2005; Hagerman, 2006a). “Most children with FXS do not have dysmorphic features that are considered abnormal or unusual as do children with Down syndrome, so they are usually not recognized as having a syndrome by physical features alone” (Hagerman, 2006, p. 8). Research on the physical and behavioral phenotype of infants and toddlers with FXS is limited. Due to the delayed diagnosis and hidden dysmorphic features, infants, and toddlers with FXS appear to develop along an appropriate trajectory until major developmental milestones are not met, such as walking and talking (Bailey, Hatton, Tassone, Skinner, & Taylor, 2001a; Bailey Jr, 2004). Delays become more apparent during the preschool years when motor delays, language delays, and undesirable behaviors are more evident. Because of delayed diagnosis, children with FXS miss valuable intervention time that could greatly enhance their development and family support. Children with Down syndrome, on the other hand can start receiving early
intervention services from birth due to early diagnosis and prominent dysmorphic features (Bailey, Roberts, Mirrett, & Hatton, 2001; Meyer & Batshaw, 2002).

Physically, children with FXS experience a variety of problems related to “connective tissue dysplasia associated with abnormal elastin fibers” (Hagerman, 2006, p. 8). These problems may include joint dislocations, hernias, and recurrent otitis media as a result of the lack of FMRP due to the FMR1 gene mutation. Early gross motor skills, such as sitting, standing, and walking, initially appear to develop on a typical trajectory for children with FXS, although delays become more apparent as children grow (Baranek et al., 2005; P. J. Hagerman, 2002). Factors affecting gross motor development include low muscle tone, sensory processing dysfunction, and hyper-extensible joints. These factors are a direct result of FXS and may contribute to a lack of opportunities to interact with peers in group sports or outside play. Fine motor delays, are also significant in children with FXS, particularly as they reach school age and more fine and visual motor skills are required for reading and writing. The combination of FXS related delays and lack of opportunities to practice such skills increase the likelihood of significant long-term delays. Decreased motor development has an impact on cognitive development as well, because children who are not able to move their bodies effectively in their environment cannot learn from their environment, which stimulates brain development (Ayres, 2005; Bruner, 2009; Guralnick, 1998; Linder, 2008; J. P. Shonkoff & Phillips, 2000).

Health problems for children with FXS include seizures, gastrointestinal disorders, cardiac problems (mitral valves prolapse), and strabismus (Hagerman, 2006a; Hagerman et al., 2008). Seizures are a significant concern for children with FXS because
20% of boys with FXS are diagnosed with seizures, although the severity is reduced with age and medication (Berry-Kravis, 2002). Infants with FXS are particularly affected by gastroesophageal reflux, although treatment strategies such as holding the infant upright after feedings and targeted medication can alleviate symptoms (Hagerman, 2006a). Health disparities for any child can have a significant impact on their development, but for children with FXS, health problems compound behavior and sensory challenges (Bruner, 2009; Currie, 2005; Vernon-Feagans & Manlove, 2005). For example, when a child with FXS has recurrent otitis media, they have pain with limited hearing, which may affect their language development, behavior, and overall readiness for learning.

**Developmental and behavioral profile of children with FXS.**

Generally, children with FXS struggle with a range of developmental and behavioral challenges and these challenges impact the development in other domains such as speech and language, sensory motor, and cognition (R. J. Hagerman, 2002; Hagerman et al., 2008). Beginning in early childhood, Hagerman (2006a) reported that infants with FXS display typical infant behaviors such as eye contact and appropriate social interaction; although some suggest hypo-responsiveness and hypersensitivity to auditory stimuli manifested in exaggerated startle responses (Hagerman, 2006d). Miller and colleagues (1999) argued that children with FXS might be physiologically predisposed to sensory sensitivity, which may lead to problems with self-regulation. Parents of infants with FXS may observe concerning behaviors, that include tactile defensiveness, limited eye contact, and self-injurious behaviors (hand biting and head banging) (Bailey, Hatton, Tassone, Skinner, & Taylor, 2001b; Hagerman, 2006b; Hagerman & Hagerman, 2002; Kogan et al., 2004; Miller et al., 1999). Social,
emotional, and behavioral disorders, such as Attention Deficit/Hyperactivity Disorder (ADHD), social anxiety, and Autism have been found to be comorbid with FXS (Epstein, Riley, & Sobesky, 2002b; Hagerman, 2006b; Hagerman et al., 2008; Hatton et al., 2002; Sullivan, Hooper, & Hatton, 2007). In addition, sensory processing disorders are prevalent including tactile defensiveness that may lead to impulsive behaviors and aggression.

While it is best to maintain a holistic view of children, recognizing salient characteristics dominated by development domains (i.e. cognition, social-emotional, language, sensory motor) it is necessary to adequately understand and ultimately address children’s individualized developmental concerns. Therefore, the following sections discuss the specific characteristic of children with FXS within the context of aforementioned developmental domains.

**Cognitive development in children with FXS.**

Fragile X syndrome is the leading cause of inherited mental retardation; therefore, delays in memory, executive functioning, and abstract reasoning are challenging for children and adults with FXS. Sherman (2002) reports, “the lack of the gene product FMRP, an RNA-binding protein, is responsible for the mental retardation” (p. 138). Research has shown a positive relationship between an individual’s amount of FMRP and executive functioning skills (Hagerman et al., 2008; Loesch et al., 2003a).

Cognitive assessments or IQ measures typically identify overall delays and confounding effects within other developmental domains for individuals affected by FXS. Learning disabilities that accompany cognitive delays, involve deficits in visual processing, auditory processing, fine motor, and sequencing (Braden, 2002; Lewis,
Abbeduto, Murphy, Richmond, Giles, Bruno, & Schroeder, 2006). Most cognitive delays are apparent in children with FXS by the time they reach school age; but for infants, toddlers, and preschoolers, delays in cognition are apparent in their play skills, acquisition of language, problem-solving, and symbolic play. There is limited research on the play skills and cognitive development of young children with FXS, but there is a general consensus that IQ measures are developmentally inappropriate for young children (Bailey Jr, Roberts et al., 2001; Linder, 2008). However, the IQ assessment of children with FXS is a well-researched area in the field and the IQ level of children with FXS is an important factor that contributes to the behavioral phenotype (Grigsby et al., 2008; Loesch et al., 2003b).

The IQ of children with FXS varies depending on gender, production of FMRP and level of intervention (Grigsby et al., 2008). Hagerman (2008) reported that “Approximately 85% of males and 25% of females with the full mutation have an IQ level < 70. Females with FXS more typically present with learning disabilities, and approximately 40% have a borderline IQ (70-85), although 30% have an IQ in the normal range (>85). For individuals with a normal or borderline IQ, the presenting features are emotional and behavioral problems; particularly involving shyness, social anxiety and mood instability. Hyperanxiety disorder, social phobia, selective mutism, and/or ADHD are seen commonly and are part of the behavioral phenotype in those with FXS, whether they have intellectual impairment or not” (p. 3).

Research on middle school children with FXS indicate that IQ scores and adaptive behaviors show a steady increase in middle childhood. IQ scores start to decline during adolescence, while adaptive behaviors remained relatively stable (Dykens et al., 1989;
Dykens et al., 1996). The decline is not due to a regression of skills, but rather a widening gap in abilities due to the delayed development of executive functioning skills. Adolescent children with FXS continue to learn and show progress, albeit at a slower rate (Bennetto & Pennington, 2002). Additional research with older males with FXS points to inconsistent patterns in adaptive behavior and IQ scores remaining within the range of mental retardation. Dykens and colleagues (1996) reported adaptive skills to be relative strengths for adult males with FXS, although functional communication remained a challenge.

**Behavioral characteristics of children with FXS.**

Identifying the source of behavioral outbursts is complicated because there are many factors that can contribute to behavioral reactions. Parents and professionals have become increasingly aware through research and everyday experiences that children with FXS are often overly sensitive to stimuli. This sensitivity coupled with developmental delays in language and cognition, often leads to tantrums, angry outbursts, and other forms of aggressive and self-injurious behavior (Hagerman, 1999b; Hagerman & Hagerman, 2002; Miller et al., 1999). Aggressive behaviors include hitting or biting others, as well as self-injurious behaviors such as hand biting and head banging (Dixon Weber, 2000; Hatton et al., 2002; Symons et al., 2003). Hatton and colleagues (2002) found that parents commonly reported the existence of problem behaviors such as withdrawal, depression, anxiety, delinquency, and aggression in children with FXS. Not surprisingly, there was a significant difference between problem behaviors for children with FXS and their typical peers. Communicating feelings and emotions is difficult for all children, although children or adults with an intellectual disability, such as FXS, often
have more difficulty expressing anxiety or worry due to their decreased ability to use functional communication. Furthermore, caregivers may misinterpret behaviors and implement inappropriate interventions that may escalate aggressive behaviors since they are unaware of the behaviors associated with FXS (Hatton et al., 2002).

Typically, children with FXS have problems with anxiety and behavior during times of transition, separations from familiar adults, and adapting to new situations. A recent study of anxiety in children with FXS found that teachers and parents identified observable behaviors of anxiety that later predicted the diagnosis of anxiety disorders (Sullivan et al., 2007). The specific “behavioural equivalent of anxiety” may include avoidance behaviors such as arguing, defiance, disengagement, whining, and obsessive-compulsive behaviors (p. 62). The authors enrolled 43 children aged 6-14 years with the full mutation of FXS. Teachers and parents completed the Child Behavior Checklist and Teacher’s Report Form (Achenbach, 1991). Salient findings include the significant difference between the ratings of teachers and parents. It appeared that teachers identified behavioral equivalents more accurately than parents. This finding is noteworthy because it suggests that parents perceive their children’s behavior differently than teachers, and therefore may implement interventions that are inconsistent with the school setting, which can be ultimately ineffective.

Other behavioral implications for children with FXS include diagnoses of attention deficit hyperactivity disorder (ADHD) and sensory processing dysfunction. ADHD is typically comorbid with 70% of the FXS population (Hagerman et al., 2008). Children with FXS have problems with impulsivity, distractibility, and inattentiveness,
which are also characteristics of sensory processing disorders, and require a different type of intervention, such as medication versus sensory integration therapy.

*Sensory processing in children with FXS.*

Sensory processing dysfunction is typical for children with FXS and key to identifying appropriate treatment, especially in relation to sensory seeking and sensory avoidance behaviors (Scharfenaker, O'Connor, Stackhouse, Braden, & Gray, 2002a). Sensory integration defined as “the organization of sensations for use” (Ayres, 2005, p. 5). More specifically, sensory integration is a process in which the brain organizes and makes meaning of the various sensory stimuli in the environment (i.e. visual, auditory, tactile, movement). Sensory integration or processing dysfunction, on the other hand, is the brain’s malfunction to organize and process the sensory stimuli, which impacts learning and other coping skills (Ayres, 2005). There are variations of sensory processing dysfunction including over-stimulation and under-stimulation. Sensory seeking is a form of under-stimulation whereas an individual has an innate need to seek out sensory stimuli for comfort or coping under times of stress. Sensory avoidance is a form of over-stimulation and refers to an individual’s need to avoid stimuli, which may feel uncomfortable or negative. Tactile defensiveness (over-stimulation) is a “sensory integrative dysfunction in which tactile sensations cause excessive emotional reactions, hyperactivity, or other behavioral problems” (Ayres, 2005, p. 202). For children with FXS, tactile defensiveness may lead to lack of exposure of various sensory stimuli, thus limiting fine motor development. For example, if a child avoids materials such as playdough or shaving cream, they will not get the input into their hands that aid the fine motor development.
Children with FXS also have difficulty processing vestibular and proprioceptive input, which may lead to motor planning problems that are movements organized in the brain first and then executed by a series of actions (Ayres, 2005). The vestibular system is “the system that responds to the position of the head in relation to gravity and accelerated or decelerated movement” and proprioceptive “input tells the brain when and how the muscles are contracting or stretching, and when and how the joints are bending, extending, or being pulled or compressed” (Ayres, 2005, p. 201-202). Children with FXS find this uneasiness of the body in motion intolerable at times and often have behavioral outbursts. The behavioral outbursts may start to interfere with social interactions with peers and adults and may result in socialization deficits. Finally, auditory and visual sensitivity is the inability to filter out the sights and sounds, and for children with FXS, this may lead to gaze aversion or lack of eye contact. Again, these stimuli can affect social relationships and academic performance.

**Social and emotional development of children with FXS.**

Children and adults with FXS (without an Autism diagnosis) are inherently social beings, meaning that they want to engage in social interactions with others. The skills that are required to be social are the same skills that are most problematic, such as sustained eye contact and verbal communication. Maintaining eye contact is an area of extreme difficulty for children with FXS, not because of social unwillingness, but because the mere act of looking someone in the eye is overwhelming at a sensory level. Their lack of sensory integration skills and inability to process multiple sensory stimuli, such as direct eye contact, often leads to inappropriate social responses. For example, Scharfenaker and colleagues (2002) report, “Sensory defensiveness may lead to
decreased interactions with caregivers and other children, peer, and adults, resulting in a disruption of the natural development of social interaction and language” (p. 378). Girls and boys with FXS differ regarding severity of delays in social and emotional development. The main concerns for girls involve shyness, ADHD, social withdrawal, anxiety, depression, and low self-esteem (Hagerman, 2006a).

**Language development in children with FXS.**

A development delay in one domain, such as language, can greatly impact on other domains, such as cognition for young children (Davies, 2004). Often times, a delay in language development is the first sign of difficulty that parents notice before a diagnosis is made (Lachiewicz & Mirrett, 2000). Young boys with FXS typically develop their first words at three years of age, 2-3 word combination come later at approximately four years of age. This is a 24-36 month delay from typical language development. Girls with FXS develop language earlier than their male counterparts, although delays are still apparent (Mirrett et al., 2004). Most children with FXS learn to use functional speech and language to meet their needs. Issues such as poor oral motor planning, tangential speech, echolalia, verbal dyspraxia, and dysfluency are cause for concern, but can be improved through appropriate speech and language therapy (Lewis, Abbeduto, Murphy, Richmond, Giles, Bruno, Schroeder et al., 2006).

**FXS and comorbid diagnoses.**

Due to the genetic complexity of FXS, it is highly comorbid with other disorders that affect children’s development, primarily their behavior and social-emotional skills. The comorbidity with Autism Spectrum Disorder is of particular concern (Hagerman et al., 2008). Fragile X syndrome and Autism have been consistently connected in studies
and researchers have reported that children with FXS and Autism have considerable cognitive, receptive language, and theory of mind impairments compared to children with only FXS only, although age may be a factor (Hagerman, 2006c; Lewis, Abbeduto, Murphy, Richmond, Giles, Bruno, & Schroeder, 2006). Hagerman and colleagues (2008) report that “approximately 30% of children with FXS have clinically-defined Autism, based on formal DSM-IV criteria and an additional find 20% pervasive developmental disorder, not otherwise specified (PDDNOS); thus Autism and the broader Autism spectrum disorders (ASD) are part of the behavioral phenotype of FXS” (p. 4). The impact on parents who have children with a dual diagnosis of FXS and Autism was reported to be particularly difficult compared to parents with children who have a singular diagnosis of FXS or Down Syndrome (Lewis, Abbeduto, Murphy, Richmond, Giles, Bruno, Schroeder et al., 2006). Another study found that the behavioral differences between children with FXS and children with both FXS and Autism resulted in more significant problems with social behavior as well as overall development for the latter group (Kau et al., 2004).

FXS is comorbid with other diagnoses in addition to Autism, such as Attention Deficit Hyperactivity Disorder (ADHD), mood instability, and social anxiety (Hagerman et al., 2008). Table 1 is from the work of Hagerman, Rivera, and Hagerman (2008, p. 3) and identified characteristics of children and individuals with the FXS full mutation and premutation.
# Table 1

**Fragile X Involvement**

<table>
<thead>
<tr>
<th>Full mutation</th>
<th>Premutation in Children</th>
<th>Premutation in Adults</th>
</tr>
</thead>
<tbody>
<tr>
<td>ADHD</td>
<td>ADHD</td>
<td>Anxiety</td>
</tr>
<tr>
<td>Autism Spectrum Disorders</td>
<td>Autism Spectrum Disorders</td>
<td>Depression</td>
</tr>
<tr>
<td>Flat feet</td>
<td>Hyperextensible finger joints</td>
<td>POF</td>
</tr>
<tr>
<td>Hand biting</td>
<td>Prominent ears</td>
<td>FXTAS</td>
</tr>
<tr>
<td>Hand flapping</td>
<td>Shyness</td>
<td>-Tremor</td>
</tr>
<tr>
<td>High arched palate</td>
<td>Social Anxiety</td>
<td>-Ataxia</td>
</tr>
<tr>
<td>Hyperextensible finger joints</td>
<td></td>
<td>-Neuropathy</td>
</tr>
<tr>
<td>Long face</td>
<td></td>
<td>-Muscle pain</td>
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<tr>
<td>Macroorchidism</td>
<td></td>
<td>-Hypothyroidism</td>
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<tr>
<td>Mitral valve prolapse</td>
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<td>-Cognitive decline</td>
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<tr>
<td>Mood instability</td>
<td></td>
<td>-Anxiety</td>
</tr>
<tr>
<td>Perseverative speech</td>
<td></td>
<td>-Depression</td>
</tr>
<tr>
<td>Poor eye contact</td>
<td></td>
<td>-Apathy</td>
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<tr>
<td>Prominent ears</td>
<td></td>
<td>-Dysinhibition</td>
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<tr>
<td>Shyness</td>
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<td>Social anxiety</td>
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<tr>
<td>Tantrums</td>
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</table>

(Hagerman et al., 2008, p. 3)
Mothers of children with FXS.

A review of the literature revealed that mothers of children with FXS face many challenges. One primary challenge is managing their children’s difficult behavior, which is a defining characteristic of children, particularly males with FXS. Epstein and colleagues (2002) reported that it is often difficult to resolve the issue of determining if the behavior challenges for children with FXS are a direct result of the FXS mutation or a secondary effect of the syndrome. Emotional and behavior problems can also be a result of a combination of environmental and biological factors. Primary environmental factors may include positive or negative interactions with family members, peers, school personnel, and community members. The physical environment often encompassing overstimulating visual, auditory, and uncomfortable proximity to other people also influences the behavioral reactions of children with FXS. In addition, transitions, changes in schedules, and the unpredictability of everyday life can also have an impact on the functioning of children with FXS. These environmental factors can have a profound effect on an individual’s ability to adapt to various stimuli in an ever-changing world. Parents can in no way control all of the environmental factors that may lead to the disruptive behavior of their children with FXS. This causes additional stress and anxiety to mothers.

Mothers of children with FXS, have their own challenges that come with carrying the premutation of the FMR1 gene in addition to managing their child’s daily functioning. Factors associated with this maternal genetic status include a heightened risk for primary ovarian insufficiency (POI), social-emotional effects, and the possibility of giving birth to another child with FXS (Grigsby et al., 2008; Sherman, 2002). McConkie-Rosell and
colleagues (2005) cite “psychological issues such as denial, anxiety, anger, grief, survivor
and parental guilt, shame, blame, depression, inability to cope, damage to self-esteem,
changed relationship with family of origin, and change in sense of identity” (p. 265).
Hagerman (2002) also reported that women who carry the $FMR1$ premutation gene, do
not typically exhibit any significant cognitive deficits, although many experience shyness
or anxiety (P. J. Hagerman, 2002). Lewis and colleagues compared the psychological
well-being of mothers of children with FXS with and without the comorbid diagnosis of
Autism and mothers of children with Down syndrome (Lewis, Abbeduto, Murphy,
Richmond, Giles, Bruno, Schroeder et al., 2006). Their research revealed that mothers
with sons with FXS (with and without Autism) reported more difficulty adapting to the
challenges in their family and negative explanatory styles regarding their child’s future
than mothers of children with Down syndrome. The authors speculated that the high
level of behavior problems within the FXS population could be a contributing factor, but
this factor was not measured or explained in the study. These findings suggest that the
emotional well-being of mothers, particularly those regarding family relationships, may
influence the development of their children (Abbeduto et al., 2004; Abrams, 2008; Bailey
et al., 2000)

There has been limited research exploring the dyadic relationships between
mothers and their children with FXS. Some of the studies that have been published
involve the mother’s psychological well-being (Lewis, Abbeduto, Murphy, Richmond,
Giles, Bruno, Schroeder et al., 2006), women’s attitudes toward FXS screening (Anido et
al., 2005), family experiences (Bailey et al., 2000; Bailey et al., 2003; Poehlmann et al.,
2005), and maternal stress (C. Johnston et al., 2003). However, the interaction between
the behavioral characteristics of the child and the overall experiences and characteristics of the mother has received limited attention. Lewis and colleagues (2006) reported in their conclusions that this relationship should be further explored.

**The impact of the delay in FXS diagnosis.**

Families, particularly mothers of children with disabilities, experience high levels of stress when parenting a child with a disability due to the disproportionate caretaking activities in comparison to families with children who are developing in a typical manner (C. Johnston et al., 2003). The FXS literature reports that mothers of children with FXS often experience increased levels of stress, feelings of isolation, and frustration more than other groups of mothers with children with disabilities, such as Down syndrome and Autism (Dixon Weber, 2000; C. Johnston et al., 2003; Lewis, Abbeduto, Murphy, Richmond, Giles, Bruno, Schroeder et al., 2006; Poehlmann et al., 2005). A number of factors may contribute to the struggle with the emotional well-being of mothers of children with FXS. The first factor may be due to the delayed diagnosis of FXS. As previously stated, the average age for a FXS diagnosis is three or four years of age, often after several misdiagnoses, resulting in a lack of or inappropriate early intervention. Many mothers appear to have worries about their child’s development long before confirmation of the FXS diagnosis and maintain persistent advocacy efforts until an accurate diagnosis is achieved. This delay in diagnosis or misdiagnosis may result in a loss of valuable intervention programs for the child and family (Bailey, 2004). It is not only possible, but also probable, for a mother to give birth to an infant with FXS without the knowledge that she is a carrier for the *FMR1* gene mutation. Furthermore, the mother continues to care for her child with the intuition that something is wrong and has limited
support with everyday challenges. A delayed diagnosis can lead to feelings of isolation and incompetence, especially if the young child has a difficult temperament, which in turn reinforces the mother’s feelings of hopelessness and frustration (Dixon Weber, 2000). If this perpetual cycle continues, then the attachment process between the infant and the mother may be disrupted (Davies, 2004).

Once a FXS diagnosis is confirmed, parents, especially mothers, continue to wrestle with the stress of raising a child with an intellectual disability. As described, children with FXS often have difficult and challenging behaviors and those behavioral and sensory processing challenges may influence mothers’ emotional well-being and interactions with their children. It is very difficult to deal with the behavior and sensory outbursts of a child with FXS on a regular basis on top of coping with the reality that the child has a disability.

**Delayed diagnosis of FXS.**

Pediatricians are most often the first professionals that parents go to when they begin to have concerns about the development of their child (Bailey, 2004). Pediatricians in a family practice may or may not have any experience or knowledge of genetic disorders, in particular the developmental influences of FXS. Studies report that parents initially have concerns about their child before they reach one year of age (Bailey, 2004; Taaffe Young, Davis, Schoen, & Parker, 1998), although when parents mention their concerns to their pediatrician, they are often dismissed and characterized as overly anxious parents. Due to the busy schedules of doctors’ offices, the pediatricians may spend only a short amount of time with the concerned parents and observe the child instead of using standardized developmental screening instruments to address concerns.
One study (Mirrett et al., 2004) argues for a more systematic screening process through the pediatricians’ office to accurately identify very young children with FXS. Mirrett and colleagues (2004) first recommend that pediatricians perform a thorough screening of phenotypic signs of FXS during the well-baby examinations, although with the numerous dysmorphic features associated with a number of developmental disorders, this action could be inaccurate and could lead to a misdiagnosis or undue worry for the parents. Developmental screenings through the pediatricians’ office could be a logical next step. The authors explored questions related to this theme. They used three relatively well-used screening tools to find out if children already identified with FXS would score in the developmentally delay or at-risk range. Even though their sample was a small and convenient one, they found that all of the tools used identified a majority of the sample for having developmental delays at nine, twelve, and eighteen months of age.

Implications from this study reveal the needs for developmental screenings by the first professional families go to when concerns arise which would be their pediatrician. Earlier identification of developmental delays may lead to an earlier genetic diagnosis and ultimate treatment of FXS.

A national study of parental perceptions of physicians’ found that a majority of parents desire more information from their physician regarding growth, development, and child rearing practices (Taaffe Young et al., 1998). The authors also found that physicians specializing in pediatrics are often unwilling to address nonmedical concerns of parents. The pediatrician may be unaware of this genetic disorder and dismiss the parents’ strong feelings that something is wrong with their child. The typical “wait and see” position taken by many pediatricians could lead to a loss of valuable intervention.
This phenomenon puts families in an untenable situation because they may not be able to access interventions without a medical or developmental diagnosis. As mentioned above, FXS is usually not diagnosed at birth, as is the case with infants with Down syndrome. The parents are sent home with what they believe to be a “healthy” infant, only to realize later that they have deep concerns for their children’s development. The diagnosis is generally confirmed after the age of 3 years and sometimes even later (Bailey, 2004). The lapse of time in between birth and the age of diagnosis is a time of extreme stress and anxiety for both parents, especially mothers, due to uncertainty and lack of information.

**Impact of mothers’ knowledge of their carrier status.**

 Mothers who discover that they are carriers of the *FMR1* gene mutation that has caused their children to have FXS often have feelings of guilt, anger, and sorrow (Epstein et al., 2002b; Lewis, Abbeduto, Murphy, Richmond, Giles, Bruno, Schroeder et al., 2006). Perpetual feelings of guilt and sorrow could affect the emotional well-being of these mothers. Few studies have explored the subject of mothers of children with FXS and their particular phenotype, due to their carrier status. Some studies have explored the neurocognitive, emotional, and behavioral profile of women who are carriers of the *FMR1* gene mutation (Bennetto, Taylor, Pennington, Porter, & Hagerman, 2001). The results showed that women with the premutation had increased levels of anxiety and depression as well as learning disabilities related to mathematical ability (Lachiewicz, Spiridigllozzi, Dawson, Cuccaro, & McConkie-Rosell). “Premutation carriers, male and female, do not usually exhibit overt cognitive or behavioral problems. However, mounting evidence suggests that premutation carriers may be at risk for specific disorders not present among the full-mutation carriers” (Sherman, 2002).
All of the previous factors that may account for the emotional well-being of mothers of children with FXS could negatively affect the relationship between a mother and her child with FXS. First, the delayed diagnosis could lead to unrealistic expectations of the child because parents do not have a definitive diagnosis and therefore unknown prognosis. Often times, parents of children with an undiagnosed disability feel frustrated by the challenging behaviors and lack of skills their children display. Extended family or community members may characterize the children as “lazy” or “defiant” when in reality the expectations placed on the child may simply be inappropriate for their developmental age. In addition, the children’s inability to control their behaviors may be organic in nature. The help that parents seek from educational and behavioral specialists may prove ineffective or inappropriate due to the lack of information about the origin of their children’s challenging behaviors.

**Grief cycle of parenting a child with FXS.**

Parents with a child with a disability experience similar stages of grief compared to individuals that have experienced the death of a loved one or are facing death themselves. The work of Kubler-Ross (1969) outlines the five stages of grief which individuals may go through when facing the death (literally or symbolically) of a child or loved one. The stages include denial and isolation, anger, bargaining, depression, and acceptance (Kubler-Ross, 1969). Parents who have a child born with a disability, or have a child that is diagnosed with a disability later, may experience the symbolic loss of that “perfect” child and therefore go through the process of mourning and stages of grief mentioned above.
During pregnancy, the parents of the unborn child are full of hopes and dreams for their baby. The parents develop strong emotional bonds to their unborn child, which continue throughout infancy to create an attachment relationship between the parents and the baby. The sudden disruption of the attachment due to the diagnosis of a disability at birth or shortly after is a loss for the family. "The closer the relationship of attachment, the more intense the grief of the relationship is disrupted" (Teel, 1991). The loss of the "perfect” baby that the parents had dreamed of becomes the center of their grief.

Through this devastating loss, the parent of a child with a disability continues to interact with the child and make decisions in the best interest of the child while he/she grieves. The constant reminder of their loss may interfere with their grief process (Teel, 1991). Parents do not necessarily go through the stages of grief sequentially, and may not even reach the last stage of grief (acceptance) due to the constant reminder of their child’s disability, and the loss of their “perfect baby.” The length of time in each stage is variable and may affect the emotional well-being of the parents. Oshlansky (1962) termed the ongoing loss or the daily reminders of the symbolic death of a child as "chronic sorrow.” Chronic sorrow encompasses the concept that the parents of a child with a disability experience recurrent episodes of sadness and depression, as well as anger and acceptance (Oshlansky, 1962). Professionals may view the reoccurrence of a parental grief as abnormal if their only construct of the grief process is the staged model. "A critical attribute chronic sorrow is the experience of episodic sadness that lasts the lifetime of the child or the parent, regardless of the physical proximity of the disabled child to the parents (Teel, 1991, p. 1314). Mothers of children with FXS, similar to mothers of children with other disabilities, struggle with their grief; therefore, it is
important to discuss the relationship between the balance of the mothers’ emotional well-being and their children’s behavioral and sensory characteristics (Teel, 1991; Weber, 2000b).

Lack of support for mothers with children with FXS.

Finally, a factor that affects the emotional well-being of mothers of children with FXS is the lack of support and knowledge regarding FXS. Even though FXS is the leading cause of inherited mental retardation, the public has very little knowledge of this syndrome and the impact it has on families. Unlike families with children with Down syndrome or Autism, which is widely covered by the media, families with children with FXS may have little family and community support. Lewis and colleagues (2006) studied this particular issue and found that mothers of sons with FXS, with and without a comorbid diagnosis of Autism, experienced higher levels of familial conflict and maternal pessimism about their sons’ future in comparison to mothers of sons with Down syndrome and/or Autism. In addition, mothers of children with Down syndrome and Autism feel more support by external networks than mothers of children with FXS.

The difficult behaviors of children with FXS could impact the mothers’ emotional well-being and in turn impact their responses to their child, thus increasing the severity and frequency of problem behaviors (Wheeler, 2005). Children with sensory processing disorders will often over react or under react to sensory stimuli within their environment. These reactions manifest themselves as inappropriate or maladaptive behaviors that impact the child and the family on a daily basis. The lack of support coupled with the feelings of guilt and sorrow about the mothers’ carrier status could lead to inconsistent responses to the child’s behavior and sensory characteristics. These responses may in
turn influence the attachment relationship between the mother and her child with FXS and lead to more maladaptive behaviors. All of the conditions that contribute to the mothers’ emotional well-being and the mother-child relationship could make intervention for the family a difficult reality.

**Impact of the mother-child relationship: expressed emotion.**

The construct of expressed emotion has grown out of the field of psychopathology research first focusing on family functioning of adult schizophrenic patients (G. Brown et al., 1972; G. Brown & Rutter, 1966; McCarty & Weisz, 2002a; Vaughn & Leff, 1976; Vostanis & Nicholls, 1992). EE is a measure of the emotional family climate connected to the verbal and nonverbal expressions of family members toward an individual. The initial basis for EE research was the finding that adult schizophrenic patients from families characterized by high levels of emotional involvement, hostility, or criticism – later collectively labeled ‘EE’ – were significantly more likely to relapse than their counterparts from families low on the characteristics (McCarty & Weisz, 2002a). Early EE studies used the Camberwell Family Interview (CFI), and linked high expressed emotion to the likelihood of a psychiatric relapse and the return to a psychiatric hospital (G. Brown et al., 1972; G. Brown, Monck, Carstairs, & Wing, 1962). Vostanis and Leff (1995) studied the relationship between parental expressed emotion and their child’s behavior in a non-clinical population through the use of the CFI, the Child Behavior Checklist (CBCL), and the Family Environment Scale (FES). Their results revealed, “Even within this ‘normal’ range, there was still a positive association between maternal criticism and child behaviour” (p. 255). Thus begins the argument that critical

---

1 As previously mentioned “expressed emotion” will represent the construct, while the abbreviation “EE” will represent the measurement.
statements made by mothers about their children could affect the child’s behavior in maladaptive ways or that maternal criticism may be viewed as a response to the child’s behavior problems. Other studies that link maternal expressed emotion and childhood behavior problems include populations of low birth weight children (St. Jonn-Seed & Weiss, 2002; Vostanis & Leff, 1995), clinically referred children and youth (Bolton et al., 2003; McCarty & Weisz, 2002b), preschool children with attention deficit-hyperactivity disorder (Daley et al., 2003), mothers with depression (Nelson, Hammen, Brennan, & Ullman, 2003), and African-American families (Kwon et al., 2006). There is now substantial evidence that supports a positive association between maternal expressed emotion and poor childhood behavioral outcomes (G. W. Brown, Harris, & Eales, 1996).

One of the early studies on expressed emotion by Brown, Birley & Wing (1972) began to define the components of the construct, which included critical comments, hostility, dissatisfaction, warmth, emotional over-involvement, and the overall index of expressed emotion. The sample of relatives not only included parents, but siblings as well as spouses of discharged patients diagnosed with schizophrenia. The authors found that the most significant component of expressed emotion related to symptomatic relapse was the number of critical comments made by a relative. In addition, the components of emotional over-involvement and hostility added depth to the interpretation of expressed emotion. Further studies provide evidence that EE is a significant predictor of schizophrenic relapse in addition to other mental illnesses relating to mood and attention disorders (Butzlaff & Hooley, 1998; Calam & Peters, 2006; Daley et al., 2003).

Jacobsen and colleagues (2000) provide further evidence for a link between mothers’ high expressed emotions and their children’s insecure attachment behaviors.
EE measures the continuum of parental warmth to parental criticisms and hostility, which relates to the degree of sensitivity parents show their child when attachment is measured. Attitudes and feelings identified through a measure of expressed emotion may impact the parent-child attachment bond and the treatment of the child by the parent (Jacobsen, Hibbs, & Ziegenhain, 2000). The results showed that “As expected, mothers who had High EE were more prone than other mothers to have children with disorganized attachment in early childhood” (p. 904). Future researchers should cautiously interpret these results due to the study’s small sample size and homogenous population.

EE is primarily a snapshot of the emotional family climate, although one longitudinal study explored the stability of expressed emotion over time and found that the level of EE was influenced by crises (Santos et al., 2001). This study is important because many families with children with FXS are often in crisis, regardless of age, especially during times of transition. Interventionists can provide support during these times to help decrease expressed emotions and problematic behaviors that may have manifested.

**Measuring expressed emotion: Five minute speech sample.**

One primary way of measuring EE is through an instrument called the Five Minute Speech Sample (FMSS) (Magana-Amato, 1993). The FMSS is a non-scripted monologue designed to measure the expressed emotion, hence the emotional relationship between a parent, or a close relative, and their child. Adapted from a longer interview process, the Camberwell Family Interview (CFI) (G. Brown & Rutter, 1966; Vaughn & Leff, 1976), the FMSS is coded for content and tone of the parents’ speech sample in relation to criticisms, emotional over-involvement, relationship status, and parental
warmth. Specifically, the speech sample is coded for Criticisms (CRIT), which may involve critical statements about the child, negative initial statements and/or statements about a negative relationship between the parent and child. The speech sample is also coded for Emotional Over-involvement (EOI) which is determined by excessive praise, emotional display, such as crying, statements about self-sacrificing over protective behavior and/or excessive detail about the child’s birth or diagnosis. The sub-scales, CRIT and EOI in combination make up the overall EE rating, which can be high or low.

EE is a qualitative and quantitative measure of the emotional climate in a family’s home. The EE dimensions, emotional over-involvement, and criticisms are potential risk factors for children in relation to behavior problems. The purpose of this study is to explore the relationship between the expressed emotion of mothers of children with FXS and the behavioral and sensory characteristics of their children.
Chapter Two: Method

Rationale for the Study

The field of fragile X syndrome (FXS) is in need of additional research regarding the effects of the mother-child relationship on therapeutic interventions and subsequent development of their children with FXS. The majority of studies on FXS highlight specific advancements in biomedical and genetic fields (Bailey, Hatton et al., 2001a; W. T. Brown, 2002; K. M. Cornish et al., 2009; Dyer-Friedman et al., 2002; Hagerman, 2006a). That research is extremely important for understanding the complex genetic and biological repercussions of FXS; however, there is a gap in scientific research regarding the relational factors between mothers and their children with FXS. The paucity lies within the information regarding how mothers of children with FXS (who are also carriers of the fragile X premutation gene) describe and view their children. Exploring the way maternal expressed emotion affects children’s behavioral functioning is worth exploring because relevant intervention strategies may be developed in response to such research. Parents and professionals need to understand how high expressed emotion affects the emotional family climate and then learn strategies on how to balance expressed emotion to optimize therapeutic interventions. This study could influence how mothers’ expressed emotion can improve or diminish the immediate and long-term effects of behavioral/sensory interventions for their children (Hatton et al., 2002). In addition, this study is worth conducting because the information collected will positively
add to the field of fragile X research. Currently, there is limited research exploring expressed emotion as measured by the Five Minute Speech Sample (FMSS) within the population of fragile X syndrome (FXS) and affected families. This research could provide additional insight and information to relevant discussions such as how the FXS diagnosis influences the mother-child relationship in terms of attachment and early intervention (J. P. Shonkoff & Phillips, 2000). Without pathologizing mothers of children with FXS, additionally this study could provide insight into the relationship between mothers and their children with FXS, as well as informing intervention strategies to promote positive mother-child relationships.

**Design**

The design of this study was based on the research questions and proposed relationships between the dependent and independent variables. The research questions are stated again below.

1. **What is the relationship between the expressed emotion of mothers of children with FXS and the perception of their children’s behavioral challenges?**
   a. Expression of emotion (EE) will be measured by the Five Minute Speech Sample (FMSS) completed by the mother (Magana-Amato, 1993).
   b. Perception of children’s behavior will be measured by the Behavior Assessment System for Children (BASC) completed by the mother (Kamphaus & Reynolds, 1998).

2. **What is the relationship between the expressed emotion of mothers of children with FXS and their perception of their children’s sensory processing?**
   a. The perception of the children’s sensory processing will be measured by the Sensory Profile (Dunn, 1999) completed by the mother.

The dependent variables for this study included the measurement of mothers’ expressed emotion and their children’s behavioral and sensory characteristics.
Independent variables (i.e. demographics) presented an accurate description of sample characteristics. Relationships between the dependent variables were explored while holding the independent variables constant. To that end, a correlation design was the most appropriate design for this study.

**Sample Size and Participants**

For correlational studies, the recommended minimum sample size is 30 participants (Cohen, 1968). In selecting an adequate sample size for this study, the author used a Power Analysis calculator website from Columbia University ([http://www.biomath.info/power/corr.htm](http://www.biomath.info/power/corr.htm)) and found that in order to have a correlation coefficient of .50, with an Alpha level of .05, and Power of .80; thirty participants were required for reliable results. The sample for this study was primarily a convenience sample (study recruitment procedures addressed below); therefore, generalizations to the population of mothers of children with FXS should be made with caution.

**The mothers.**

Due to the low-incidence of FXS, the inclusion criteria was broadly defined as any mother with a child with the full or partial mutation of the FMR1 gene, regardless of age, gender, or culture/language. FXS is an inherited genetic developmental disorder, and therefore does not discriminate based on race, religion, or socioeconomic factors (Hagerman & Hagerman, 2008). Every effort was made to recruit participants that reflect diverse cultural, economic, and familial systems, although the diversity of participants for this study was limited. This may be due to the small population of mothers who access the recruitment sites (see below) and volunteer for research protocols.
Thirty-four mothers completed the demographic questionnaire, although the entire sample for the study involved 37 mothers. Missing data was present for each of the measures and questionnaires, primarily due to lack of return. Therefore, the demographic information presented below reflects an \( n \) of 34, instead of the total \( n \) of 37.

Within the demographic questionnaire, the mothers provided information about themselves, their children, and families. A majority of the mothers were between the ages of 31-50 years, married, and had a family household income over $60,000. Many of the mothers reported that they were college graduates and chose to be stay-at-home parents. Culturally and ethnically, there was little diversity in the sample, with a majority of the mother self-selecting their race as “Caucasian.” Even though the sample was not culturally or ethnically diverse, it was geographically diverse, as the sample included mothers from many states across the United States and one from South America as shown in Table 2.

Table 2

<table>
<thead>
<tr>
<th>Demographic Characteristics of Mothers of Children with FXS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Characteristic</td>
</tr>
<tr>
<td>Maternal Age (n=34):</td>
</tr>
<tr>
<td>31-40 years</td>
</tr>
<tr>
<td>41-50 years</td>
</tr>
<tr>
<td>51-60 years</td>
</tr>
<tr>
<td>Over 60 years</td>
</tr>
<tr>
<td>Family Income (n=34):</td>
</tr>
<tr>
<td>Less than $10,000</td>
</tr>
<tr>
<td>$10-20,000</td>
</tr>
<tr>
<td>$20-40,000</td>
</tr>
<tr>
<td>$40-60,000</td>
</tr>
<tr>
<td>$60-100,000</td>
</tr>
<tr>
<td>Over $100,000</td>
</tr>
<tr>
<td>Characteristic</td>
</tr>
<tr>
<td>----------------------</td>
</tr>
<tr>
<td><strong>Marital Status (n=34):</strong></td>
</tr>
<tr>
<td>Single, never married</td>
</tr>
<tr>
<td>Separated</td>
</tr>
<tr>
<td>Divorced</td>
</tr>
<tr>
<td>Married</td>
</tr>
<tr>
<td>Widowed</td>
</tr>
<tr>
<td><strong>Education (n=34):</strong></td>
</tr>
<tr>
<td>High School or GED</td>
</tr>
<tr>
<td>Some College</td>
</tr>
<tr>
<td>College Graduate</td>
</tr>
<tr>
<td>Post-College Degree</td>
</tr>
<tr>
<td><strong>Employment (n=34):</strong></td>
</tr>
<tr>
<td>Full-time</td>
</tr>
<tr>
<td>Part-time</td>
</tr>
<tr>
<td>Stay-at-home parent</td>
</tr>
<tr>
<td>Seeking employment</td>
</tr>
<tr>
<td><strong>Ethnicity (n=34):</strong></td>
</tr>
<tr>
<td>Latino/Hispanic</td>
</tr>
<tr>
<td>African American</td>
</tr>
<tr>
<td>Caucasian</td>
</tr>
</tbody>
</table>

**The children.**

The children with FXS as described by their mothers had a mean age of 13 years with a range of 2-40 years. Due to the low incidence of FXS, the inclusion criterion was broad. It was necessary to exclude age as a significant factor that could confound correlational results because of the wide range of children’s ages in this sample. To that end, four groups based on Erikson’s stages of human development were created and a test of analysis of variance (ANOVA) completed to identify the existence of group differences as defined by age (Erikson, 1950). Children in the youngest group, labeled *Early Childhood*, consisted of children ages birth to five years (n = 7). Children in the *Middle Childhood* group were ages 6 to 12 years (n = 14); children in the *Adolescence* group were 13-18 years (n = 9), and the *Young Adulthood* group contained 19-40 year
olds (n =7). The largest was the middle childhood group, followed by the adolescents, young adults, and early childhood. The ANOVA concluded that the only significant demographic differences between the age groups were in regards to maternal age ($F =17.02; df =3; p<.05$) and age of diagnosis ($F =8.97; df =3; p<.05$). In both of these variables, the means young adult group stood out because the mothers tended to be older and their age of diagnosis was also higher. This is also the group with the widest range of age (19-40 years). There were no significant differences for the remaining demographic variables. The youngest child in the sample was two years of age and the eldest child was 40 years. Figure 2 illustrates the breakdown of age groups within this sample.

**Figure 2.** Child Age Groupings
Additional child information collected from the mothers included, gender, children’s ethnicity, FXS diagnosis, age at diagnosis, and additional diagnoses. Seventy-six percent of the children were male and 24% were female. Like the mothers, the ethnicity of the children remained relatively undiversified, with a majority of the children identified as Caucasian.

As for the diagnosis at FXS, 94% of the mothers reported that their child was diagnosed with the full mutation, while two mothers reported a mosaic status or partial mutation. The mean age at diagnosis was 4.8 (SD = 6.39) corroborating earlier research that indicated that the average age of FXS diagnosis is after 3-4 years of age (Bailey, 2004; Hagerman, 2006a). The age of diagnosis ranged from six months to 32 years of age, with a clear association between the younger children in the sample and a younger age of diagnosis.

Children with FXS often have multiple diagnosis and researchers reported a high rate, up to 30% of comorbidity with Autism (Bailey Jr, Hatton, Skinner, & Mesibov, 2001; Hagerman, 2006a; Hagerman et al., 2008) in addition to other behaviorally-based disorders. Therefore, when asked if their children had other diagnoses in addition to FXS, 40% of the mothers reported that their children had two or more diagnoses. Other diagnoses included sensory processing disorder, Autism spectrum disorder (ASD), attention deficit hyperactivity disorder (ADHD), and learning disabilities. Table 3 illustrates the profile of the children with FXS of the participating mothers in this sample.
Table 3

Profile of Children with FXS

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Mean</th>
<th>Standard Deviation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age (n=37)</td>
<td>13.2</td>
<td>9.2</td>
</tr>
<tr>
<td>Age of FXS Diagnosis (n=34)</td>
<td>4.8</td>
<td>6.4</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Frequency</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gender (n=37)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>28</td>
<td>76%</td>
</tr>
<tr>
<td>Female</td>
<td>9</td>
<td>24%</td>
</tr>
<tr>
<td>Ethnicity (n=34)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Latino/Hispanic</td>
<td>1</td>
<td>3%</td>
</tr>
<tr>
<td>African American</td>
<td>1</td>
<td>3%</td>
</tr>
<tr>
<td>Caucasian</td>
<td>32</td>
<td>94%</td>
</tr>
<tr>
<td>FXS Diagnosis (n=37)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Full Mutation</td>
<td>35</td>
<td>95%</td>
</tr>
<tr>
<td>Partial Mutation or Mosaic</td>
<td>2</td>
<td>5%</td>
</tr>
<tr>
<td>Additional Diagnoses (n=33)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Autism Spectrum Disorder</td>
<td>4</td>
<td>12%</td>
</tr>
<tr>
<td>Sensory Processing Disorder</td>
<td>2</td>
<td>6%</td>
</tr>
<tr>
<td>ADHD</td>
<td>13</td>
<td>40%</td>
</tr>
<tr>
<td>2 or more diagnoses</td>
<td>8</td>
<td>24%</td>
</tr>
<tr>
<td>No other diagnoses</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

The family.

Within the families of the participants, 56% of the mothers reported that they had one other child, 28% reported that they had two or more children, and 20% had no other children. Of the families with other children, 41% said they had another child with FXS in their family. When asked what other transitions their family was experiencing at the time of data collection, many families reported that they were in the middle of Individualized Education Plan (IEP) reviews, additional diagnoses, and/or other family changes. Sixty-eight percent of the mothers also said that they were involved in or
receiving family support through local or national FXS groups. Table 4 illustrates the family characteristics and transitions that could have potential effects on their children with FXS.

Table 4

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Frequency</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Other children in the family</td>
<td></td>
<td></td>
</tr>
<tr>
<td>No additional children</td>
<td>7</td>
<td>21%</td>
</tr>
<tr>
<td>1 additional child</td>
<td>19</td>
<td>56%</td>
</tr>
<tr>
<td>2 additional children</td>
<td>6</td>
<td>18%</td>
</tr>
<tr>
<td>3 additional children</td>
<td>2</td>
<td>6%</td>
</tr>
<tr>
<td>Other children with FXS</td>
<td>14</td>
<td>41%</td>
</tr>
<tr>
<td>Transitions</td>
<td></td>
<td></td>
</tr>
<tr>
<td>IEP Review</td>
<td>8</td>
<td>24%</td>
</tr>
<tr>
<td>Medical or educational diagnosis</td>
<td>3</td>
<td>9%</td>
</tr>
<tr>
<td>Birth of a sibling</td>
<td>1</td>
<td>3%</td>
</tr>
<tr>
<td>Parental change (i.e. employment/re-location)</td>
<td>3</td>
<td>9%</td>
</tr>
<tr>
<td>2 or more transitions</td>
<td>5</td>
<td>15%</td>
</tr>
<tr>
<td>No transitions</td>
<td>13</td>
<td>39%</td>
</tr>
<tr>
<td>Involvement of FXS Support Group</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>23</td>
<td>68%</td>
</tr>
<tr>
<td>No</td>
<td>11</td>
<td>32%</td>
</tr>
</tbody>
</table>

Note. N = 35

Recruitment.

The participants were recruited for voluntary participation through flyers distributed through private therapists that provide services to families with children with FXS, hospitals that serve families with children with FXS, and a national website that provides information on all aspects of FXS. Specifically, the first recruitment site was a private therapy agency in Denver, CO, Developmental FX, a not-for-profit organization.
that specializes in clinical support to families who have children with fragile X syndrome and/or other neurodevelopmental disorders. The second recruitment site was The Children’s Hospital, Child Development Unit (CDU). The CDU evaluates a variety of children with developmental and behavioral difficulties, particularly children with FXS. The National Fragile X Foundation website (www.fragilex.org) was the final recruitment site. This website is a clearinghouse of information about FXS for families, therapists, practitioners, and researchers. Many families with children with FXS use this website for support and information relating to FXS. The author distributed recruitment flyers to each recruitment site’s personnel for authorization, and did not request permission for participants’ private information.

The sample was purposeful and convenient because the author targeted locations where families with children with FXS frequent. Part of the sample may have also come from a “snowball effect” in which one participant informed other mothers of the value and ease of this study (via internet list serves). This snowball effect was an important recruitment strategy because many families touched by FXS connect with other families who also have children or family members with FXS. These families were also eager to participate in studies that further the awareness, knowledge, and effects of FXS. Consequently, the recruitment of families through word of mouth from other families across the nation was an effective approach to increase participation in this study.

Data Collection Procedures

The mothers who agreed to participate in this study were asked to complete a short audio taped interview, two self-administered standardized instruments, and one
demographic questionnaire for a one-time data collection procedure. The author collected the data by visiting the mothers in their homes, if they lived within the Denver metropolitan area. Mothers, who lived outside of the Denver metropolitan area, completed the interview process through long-distance phone calls, and instruments/questionnaires were completed and collected via postal service. Due to the low-incidence of FXS and a limited sample in the Denver metro area, nation-wide and long-distance data collection was necessary. Through direct communication with other researchers who have used the FMSS, it was determined, that long-distance data collection through audio taped phone interviews would be valid and reliable for this measure (personal communication Mary Klinnert, 2007).

The short interview mentioned above was The Five Minute Speech Sample (FMSS; Magana-Amato, 1993) and the dependent variable of this study (a detailed description of this instrument is below). The FMSS was specifically chosen for its unique characteristics and overall construct of expressed emotion \(^2\) in which a parent or relative expressed their feelings towards their child. Drs. Wamboldt and Klinnert at the National Jewish Hospital in Denver, Colorado conducted the required training for this author in order to track the standardized administration and coding of the FMSS over the course of 8 weeks in 2005. The other instruments, described below, are published self-administered instruments with standardized scoring procedures. This study's methodology required direct contact with the participants rather than obtaining required

\(^2\) As previously mentioned “expressed emotion” will represent the construct, and the abbreviation, “EE” will represent the final rating of the Five Minute Speech Sample.
information from indirect sources. The following numbered list summarized the recruitment and data collection procedures.

1. Potential participants viewed the recruitment flyer on the National Fragile X Foundation’s web site or other participating families referred participants to the recruitment flyer.

2. Potential participants called or emailed the author about their interest in participation.

3. The author communicated with potential participants and informed them of confidential consent procedures and the purpose of the study.

4. The potential participants either declined or chose to participate. Those who chose to participate provided the author with their mailing address, telephone number for future data collection.

5. The author mailed participants the packet of questionnaires, consent form, and project information sheet together.

6. After the author confirmed that the participants received the packet, reviewed, and signed the consent form, the FMSS was scheduled for a later date, and completed through a phone interview or in-person home visit.

7. At the scheduled date for data collection, the FMSS was audio-taped with a telephone-recording device connected to a digital audio recorder, or the author audio-taped the FMSS during a home visit.

8. The participants returned the questionnaires in a self-addressed stamped envelope provided by the author via postal service.

**Consent procedures.**

Since this study engaged humans as participants, procedures to protect them from undo harm were implemented. First, informed consent was obtained before any data was collected. The potential participants contacted the author if interested in participating in the study and the author set up a convenient time for the family to review the consent procedures and the purposes of the study. At the initial meeting or phone call, the author discussed the consent form and procedures of the study with each family to ensure their
understanding of participation (including an audio-taped interview and the completion of questionnaires) and withdrawal if so desired. All information regarding the participants was kept confidential. The author was the only other person in the room at the time of data collection so that the participants would feel comfortable expressing their emotions about their child. Parents were also encouraged to make sure their child (whom they were talking about) was in a safe place during the interview because it is not appropriate to talk about the child’s behavior in his/her presence, and may cause stress to the child (Weber, 2000b). The participants’ responses were coded by number and those codes were kept separately from other data to protect the participants’ identifying information. The data was kept in a locked filing cabinet at all times and the only other person to view the data was the author’s advisor, a professional transcription contractor, and a professional colleague for determination of inter-rater reliability. In reporting the results of the study, no identifying or discernable information are revealed. The report is comprised of collapsed data and therefore the specific identities of individuals are not revealed. The data reflect averages of the group as well as general wording from the transcribed Five Minute Speech Sample.

Risks and benefits.

Previous research revealed that women who are carriers for the FMR1 gene (mothers of children with FXS) are more anxious and may exhibit more emotional volatility than mothers of children who are developing in a typical manner (C. Johnston et al., 2003). Mothers of children with FXS are usually the primary care takers of their children with FXS, so there may be an unexpected emotional reaction from these
participants in particular. The author was aware of these characteristics, and took care not to create undue stress for the mothers. This population of mothers has successfully participated in survey research in the past, and there was no reason to anticipate that they would have difficulty with this study.

The potential risks to the mothers participating in this study were minimal. Although, risks may be associated with psychological distress due to the emotional nature of mothers speaking about their child and the effects of FXS on their families. Developmental disabilities can be an emotional topic for many families, particularly regarding early developmental issues. Therefore, if a parent experienced any emotional distress due to the completion of the measures mentioned above, they were encouraged to contact the author, and she would refer them to a clinical psychologist for a mental health consultation, if need. Throughout the study, none of the subjects indicated emotional distress. There was minimal time commitment for participation in this study because it consisted of a one-time data collection process. The time commitment consisted of five minutes for the FMSS and 45 minutes to one hour for questionnaire completion. No adverse effects were anticipated, or revealed.

The field of fragile X research is a growing area of study, especially in relation to intervention for families. Historically, much of the research was medically based, focusing on genetics or bio-medical advancements. Therefore, a benefit to conducting this research was that it could lead to important intervention strategies for families. In the opinion of the researcher and the IRB, this outweighed the potential risks to the participants. This study proposed to draw conclusions about the relationship between
how mothers talk about their child (expressed emotion) and the behavioral and sensory characteristics of their child with FXS. The existence of a relationship between these two factors can lead to possible intervention strategies for mothers and their children with FXS, particularly infants and toddlers with FXS and the mother/child dyad. For instance, these findings may provide added information and awareness to the early identification and intervention of children with FXS. Given the later diagnosis of children with FXS, families do not receive their first diagnosis of FXS until their child is about 3-4 years of age, losing valuable intervention time for the child and family (Bailey et al., 2000; Bailey et al., 2003). This diagnosis delay is notably different for the population of families with children with Down syndrome, resulting in less stress and more support services early on for families with children with Down syndrome as opposed to families with children with FXS (Poehlmann et al., 2005).

**Instruments/Measures**

The data was obtained from audio-taped interviews with the mothers of children with FXS, and self-administered questionnaires. The following instruments were used:

3. The Sensory Profile (Dunn, 1999)

Demographic questionnaire developed by the author.

**Five minute speech sample (FMSS).**

Five Minute Speech Sample (FMSS) (Magana-Amato, 1993, 2002) is a standardized instrument used to rate the expressed emotion (EE) of a relative towards an
individual within their family. Expressed emotion is the primary construct measured by the FMSS. For the purposes of this dissertation, the term, expressed emotion, is used when referring to the construct, and the abbreviation, EE, will be used when referring the overall rating of the FMSS. “The Five Minute Speech Sample (FMSS) EE coding system distinguishes the nature of the respondent-relative’s environment by identifying emotions, feelings and attitudes expressed by a respondent about a relative during a 5-minute monologue” (Magana-Amato, 1993, 2002). The interviews were transcribed and analyzed for content and tone. The primary coding for the FMSS are NEUTRAL, POSITIVE, and NEGATIVE for the following categories (a) initial statement, (b) relationship, (c) criticism, and (d) emotional over-involvement. The subscale scores include Emotional Overinvolvement (EOI) and Criticism (CRIT), which are coded as LOW, HIGH, or BORDERLINE; and the final EE score is dichotomous, coded as either LOW or HIGH. The categories and scoring procedures were largely based on a longer familial interview process, called the Camberwell Family Interview (CFI; Brown & Rutter, 1966). The CFI was developed as a standardized instrument for measuring the construct of expressed emotion, in particular, the emotional climate verbally expressed between a family member and his/her relative with mental or developmental disorders.

The guidelines provide extensive coding instructions for each of the categories with multiple caveats and examples. For example, there are guidelines for administration concerning the following:

- **Physical setting**: The guideline state that only the examiner and the respondent be in the room during the interview process, so that the respondent can talk freely without distraction or inhibition.
• **Adequate equipment:** Proper equipment is essential to data collection. The manual recommends that the examiner have a good tape recorder, a high-quality microphone, and a stopwatch allowing the respondent to speak for exactly 5 minutes. “If the respondent is unable to continue for the full 5 minutes, even after the appropriate prompts has been given, then the tape machine must continue to record until the time has elapsed” (Magana-Amato, 2002; p. 5).

• **Verbatim instructions:** In order to maintain reliability of instruction, the administrator reads following instructions read aloud to the participant:

> “I’d like to hear your thoughts and feelings about (relative’s name), in your words and without my interrupting with any questions or comments. When I ask you to begin I’d like you to speak for 5 minutes, telling me what kind of person (relative’s name) is and how the two of you get along together. After you begin to speak, I prefer not to answer any questions until after the 5 minutes are over. Do you have any question before we begin?” (Magana-Amato, 2002).

• **Answering questions before, during, and after the procedure:** The guidelines give specific responses to questions that the respondents may have to maintain consistency of the instrument.

• **Dealing with issues or concerns from the participant during the procedure:** The issues or concerns that arise from the procedure may include respondents finishing their talk before the five minutes have elapsed, pausing and looking for reassurance, or showing emotional distress, such as crying.

• **Actions the examiner should not take:** These are very specific actions that will disrupt the reliability of the instrument:

1. “Do not say anything while the respondent is delivering the speech, not even ‘Mm-hmm.’

2. Do not use leading prompts, such as ‘Could you tell me a little bit more about you and (relative’s name) get along?’ or ‘Could you tell me more about what type of person (relative’s name) is?’

3. Avoid looking at the respondent while he/she is giving the speech sample. Seem busy with papers, etc. Do not disturb the respondent in any way. Some respondents may find a lack of eye contact distressing in this event, minimal eye contact may alleviate verbal blocking by the speaker” (Magana-Amato, 2002; p. 8).
General and specific scoring procedures are included within the guidelines for more detail. The guidelines provide extensive direction for coding independently and for interrater reliability, although no reliability or validity statistics are presented. Several studies have been implemented to document the reliability and validity of the FMSS with both mothers and fathers. Beginning with two studies by Magana and colleagues (1986), in which they documented the consistency of the FMSS with the longer, and already established Camberwell Family Interview (CFI). Results of the first study revealed agreement between three raters of the FMSS and CFI classifications (Kappa statistic-.70, .70, .80), as well as a significant correlation between “proportion of variance shared by the two systems” ($r = .38, df=49, p < .005$) (Magana, et al., 1986; p.210). The second study was not only a replication of the first, but also encompassed another language component, with a Mexican-American, Spanish-speaking sample. The results were very similar to the English-speaking sample and revealed that, “A person who is classified as high EE on the FMSS has a very high probability of receiving the same rating on the CFI” (p. 210). These two studies provided the foundation for future research on the FMSS, which was originally intended to be used as a screening instrument for the CFI, but has developed into a valid and reliable instrument in its own right.

Additional studies have been completed on the validity of the FMSS, with a variety of populations, including children with asthma (Wamboldt, O’Connor, Wamboldt & Klinnert, 2000), infants from low-income families (Kaugars, Moody, Dennis, Gavin & Klinnert, 2007), children with depressive disorders (Asarnow et al., 1993), patients with schizophrenia in Japan (Shimodera, Mino, Inoue, Izumoto, Kishi and Tanaka, 2004), and
children with behavior problems (Calam & Peters, 2006). These studies provide evidence for the accurate reflection of the content and construct development of the FMSS. Study after study has shown that the FMSS can accurately measure the expressed emotion of diverse caregivers and their children or relatives with mental, physical, or developmental challenges (Baker et al., 2000; Daley et al., 2003; Jacobsen et al., 2000; Kopelowicz, Zarate, Gonzalez, Lopez, & Ortega, 2002; Lenior, Dingemans, Schene, Hart, & Linszen, 2002; McCarty & Weisz, 2002b). Therefore, the FMSS appears to be an acceptable measure for the FXS population.

**FMSS subcategory ratings.**

Within the FMSS, four categories are scored discretely or by frequency count. Subcategories can be found within the broader categories. The following list is a description of each of the categories from the FMSS guidelines (Magana-Amato, 2001).

1. **“Initial statement:** The initial statement is based on the first thought or idea expressed by the respondent about his/her relative.

2. **Relationship:** The relationship is based on statements that describe the relationship between the respondent and the relative. These statements are taken into account when making the overall “quality of relationship” rating.

3. **Criticism:** A criticism is a comment indicating that the respondent dislikes, resents, disapproves of, or is angered or annoyed by the relative’s behavior or characteristics.

4. **Dissatisfaction:** Statements of dissatisfaction indicate that the respondent is bothered, irritated, or upset by the relative’s behavior or characteristics.

5. **Emotional Over-involvement (EOI):** Emotional over-involvement is indicated by statements that demonstrate that the respondent is excessively involved with the relative. There are five subcategories used when coding EOI” (p. 10).
The subcategories of Emotional Over-involvement (EOI) require further explanation due to the subtlety of scoring. Within the guidelines, there are several examples for weak or strong evidence of each of the following EOI subcategories as well as cautionary messages about scoring.

“Self-sacrificing/Overprotective Behavior (SSOP) (Overall rating)

Scored as present when:

- The respondents reports that he/she has sacrificed him/herself in an extreme and/or unusual manner for the relative, and/or
- Respondent’s behaviors indicate extreme and/or unusual overprotection of, or over-involvement with the relative, and/or
- The respondent implies that he/she does not enjoy the self-sacrificing/overprotective behavior he/she describe.
- Evidence of extreme interdependency with behavioral consequences, with behavioral descriptions in text” (p. 31).

“Emotional Display (Overall rating)

- Emotional display is scored as present when the respondent cries, becomes “choked up,” tears, exhibits emotion that disrupts the speech flow, or is unable to speak during the interview due to what seems to be emotional sentiment regarding the relative” (p. 34).

“Excessive Detail about the Past (Overall rating)

- Excessive detail is scored when the respondent gives an inordinate amount of extraneous or irrelevant information about the relative’s distant past. These statements must be made without associating the thoughts or ideas to the present—the statements cannot be relevant to current life situation” (p. 35).

“Positive Remarks (Frequency count)

- A positive remark is one in which a person’s behavior or personality is praised or complimented. Five or more positive remarks constitute excessive praise” (p. 36).
“Statements of Attitude (Frequency count)

- A statement of attitude is scored present when the respondent expressed very strong feelings of love for the relative or willingness to do anything for the relative in the future” (p. 42).

The following section outlines the criteria for the final EE ratings as stated within the FMSS guidelines.

Criteria for assigning the final expressed emotion rating.

A. “High Expressed Emotion is assigned when one of the following is present:
   1. **Critical**—The presence of any of the following is necessary for a high critical rating.
      a. A negative initial statement.
      b. A negative relationship rating.
      c. One or more criticisms
   2. **Emotional Over-involvement (EOI)**—The presence of any of the following statements and/or behaviors is necessary for a high EOI rating.
      a. Self-sacrificing/over-involvement behavior
      b. Emotional display during the interview
      c. Any two of the following:
         i. Excessive detail about the past
         ii. One or more statements of attitude
         iii. Excessive praise (5 or more positive remarks)
   3. **Critical and EOI**—this rating is assigned when both 1 or 2 above are present.

B. **Low Expressed Emotion**—This rating is assigned when none of the ratings 1-3 above applies

C. **Borderline Expressed Emotion**—A borderline rating is given to speech samples that contain evidence for, but do not qualify as high—EE. There are two classifications of borderline rating—Critical and EOI. However, with populations where a lower threshold, high—EE might be more appropriate, borderline speech samples may be considered high—EE. This could be particularly important when coding for groups that are reluctant to express strong attitudes about their relatives, for example, parents of young children (italics added by author).

There are five individual components, which present alone, would be rated at borderline EE, and are further categorized as either borderline critical or EOI.
1. **Borderline Critical**—The presence of one or more statements of dissatisfaction is necessary for a borderline high—EE—critical rating.

2. **Borderline EOI**—The presences of one of the following:
   a. Borderline Self-Sacrificing/Over-protective behavior
   OR
   b. One or more statements of attitude
   OR
   c. Excess detail about the past
   OR
   d. Five or more positive remarks” (pp. 44-45).

**Behavior assessment system for children (BASC-2).**

The BASC-2 (Reynolds & Kamphaus, 2004) is a system for assessing the behavior and self-perceptions of children and young adults, aged 2-25 years. This self-administered questionnaire includes two rating scales, one for parents and one for teachers, but for the purposes of this study, only the rating scale for parents will be used because the purpose is to explore relationships between mothers’ expressed emotion and the perception of their children’s behavior, rather than to triangulate the scales. In this comprehensive rating scale, parents rate their child’s observable behavior using the Parent Rating Scale (PRS) and those ratings provide scores in four areas: (a) attention problems, (b) hyperactivity, (c) internalizing problems, and (d) adaptive skills. The response format for the PRS is a four-point scale based on the frequency of the responses: *Never, Sometimes, Often,* and *Almost Always.* The PRS asks parents to rate their child’s behavior using the previous response format on statements from a variety of behavioral constructs such as:

- Has trouble following regular routines. (Activities of Daily Living)
- Gets upset when plan are changed. (Adaptability)
- Disrupts the play of other children. (Aggression)
• Worries about making mistakes. (Anxiety)
• Is easily distracted. (Attention Problems)
• Shows feelings that do not fit the situations. (Atypicality)
• Uses others’ things without permission. (Conduct Problems)
• Is easily frustrated. (Depression)
• Responds appropriately when asked a question. (Functional Communication)
• Has poor self-control. (Hyperactivity)
• Gives good suggestions for solving problems. (Leadership)
• Has trouble keeping up in class. (Learning Problems)
• Offers to help other children (adolescents). (Social Skills)
• Complains of being sick when nothing is wrong. (Somatization.)
• Has good study habits. (Study Skills)
• Clings to parents in strange situations. (Withdrawal)

There are three age levels of the BASC-2 that cover a wide range of ages;

Preschool (2-5 years); Child (6-11 years); and Adolescent (12-21 years). The BASC-2 is a comprehensive behavior rating system that may be used for multiple reasons, including clinical diagnosis, educational classification, program evaluation, forensic evaluation, and research. “The original BASC has been used in more than 125 such research studies, many of which were large-scale, longitudinal analyses of both developmental psychopathology and treatment efficacy” (Reynolds & Kamphaus, 2004, p. 10).
The reliability and validity for the Parent Rating Scales (PRS) are very high as reported in the manual and technical report (Reynolds & Kamphaus, 2004). The sample size for studies on PRS is over 4800 across the United States from August 2002 to May 2004. A representative sample encompassed children and young adults from 2-21 years, and college-aged students, 18-25. The sample also includes children and young adults who received special services due to identified disabilities.

Internal consistency or the coefficient alpha of the composite scores range from .85 to .95 for the general population. Coefficient alphas are equally high for the clinical norm sample. The PRS test-retest reliability was measured by asking parents to rate the same child twice over several weeks. The researchers found scores ranging from the low .80s to low .90s, with the exception of one subscale, Internalizing Problems, at .78. The PRS interrater reliability measures the type of agreement from different caregivers for the same child at the same time. “In general, interrater correlations are lower than those obtained in the coefficient alpha and test-retest reliability studies. Median interrater reliabilities are .74, .69, and .77 for preschool, child, and adolescent levels, respectively” (Reynolds & Kamphaus, 2004, p. 167).

The BASC-2 manual reports four types of validity; 1) intercorrelations and factor analysis; 2) PRS correlations with other behavior instruments; 3) PRS group scores with children with specific clinical diagnoses; and 4) scale content developed through test construction. Intercorrelations between the composite and subscales and factor analyses were generally high indicating that scales are significantly related in content. The PRS of the BASC has been correlated to three other highly respectable behavior instruments; the
Child Behavior Checklist (Achenbach & Rescorla, 2000); the Conner’s Parent Rating Scale-Revised (Conners, 1997); and the Behavior Rating Inventory of Executive Functioning (BRIEF; Goia, Isquith, Guy, & Kenworthy, 2000). The BRIEF was originally targeted for use in this current study, although after initial analysis of the pilot study (see below), and a high correlation between the BASC and the BRIEF, the duplication was deemed unnecessary. “Overall, correlations between the PRS and BRIEF scales are moderate to high. The Global Executive Composite (which includes all BRIEF subscales) correlates highly with the PRS Externalizing Problems composite and the BSI (Behavior Systems Index), ranging from .58 to .67 for the child sample and .80 to .86 for the adolescent sample” (Reynolds & Kamphaus, 2004, p. 184).

Through test construction and content validation, the BASC-2 has been found to be useful and highly correlated with various groups of children who typically have profiles consistent with behavior, emotional, physical, and/or learning problems. Item content also came from parents, psychologists and the Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision (DSM-IV-TR; American Psychiatric Association, 2000). The norm samples included sufficient cases for the following groups:

- Attention Deficit/Hyperactivity Disorder
- Bipolar Disorder
- Depression Disorders
- Emotional/Behavioral Disturbance
- Hearing Impairment
- Learning Disability
• Mental Retardation or Developmental Delay
• Motor Impairment
• Pervasive Developmental Disorders
• Speech or Language Disorder

In general, the BASC-2 is a sound instrument that is reliable and valid, and typically used in many different populations for a variety of purposes. It was chosen for this study because of its accuracy in detecting behavioral challenges rated by parents, particularly mothers of children with FXS. As noted in the literature review, children with FXS commonly have behavioral challenges.

Table 5 below represents the classification system of the BASC and is used in descriptive report writing for individuals or reporting research results.

Table 5

BASC Classifications

<table>
<thead>
<tr>
<th>Classification</th>
<th>Adaptive Scales</th>
<th>Clinical Scales</th>
<th>T-Score Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Very High</td>
<td>Clinically Significant</td>
<td>70 and above</td>
<td></td>
</tr>
<tr>
<td>High</td>
<td>At-Risk</td>
<td>60-69</td>
<td></td>
</tr>
<tr>
<td>Average</td>
<td>Average</td>
<td>41-59</td>
<td></td>
</tr>
<tr>
<td>At-Risk</td>
<td>Low</td>
<td>31-40</td>
<td></td>
</tr>
<tr>
<td>Clinically Significant</td>
<td>Very Low</td>
<td>30 and below</td>
<td></td>
</tr>
</tbody>
</table>

*Note. (Reynolds & Kamphaus, 2004)*
Sensory profile.

The Sensory Profile (Dunn, 1999) is a questionnaire completed by parents and caregivers which measures children’s responses to sensory stimulation in their natural environments. According to the technical report by Dunn (2008), the Sensory Profile is a “judgment-based questionnaire designed to contribute to a comprehensive assessment of a child’s sensory performance when combined with other evaluations, observations, and reports” (p. 1).

Three Sensory Profiles measure the sensory process of children from infancy to adulthood. The first, titled, the Sensory Profile, addresses children aged 3-10 years; next, Infant/Toddler Sensory Profile, addresses children birth to 36 months, and lastly the Adolescent/Adult Sensory Profile, is for individuals 11-90 years. All variations of this instrument provide information corresponding to all sensory systems and the individuals’ reactions to sensory input.

The Sensory Profile contains a long form (125 items) and a Short Sensory Profile (38 items) in a 5-point Likert scale format (almost never, seldom, occasionally, frequently, almost always). For the purposes of this study, mothers of children with FXS were asked to complete the short form of the Sensory Profile, depending on the child’s age. Dunn reported that the Short Sensory Profile is often used and most appropriate for research protocols, which target sensory modulation only (2008).

The following list outlines the three main sections of the Sensory Profile (Dunn, 1999) and the multiple categories within each one.

1. Sensory Processing
   a. Auditory Processing
b. Visual Processing  
c. Vestibular Processing  
d. Touch Processing  
e. Multisensory Process  
f. Oral Sensory Processing  

2. Modulation  
   a. Sensory Processing Related to Endurance/Tone  
   b. Modulation Related to Body Position and Movement  
   c. Modulation of Movement Affecting Activity Level  
   d. Modulation of Sensory Input Affecting Emotional Responses  
   e. Modulation of Visual Input Affecting Emotional Responses and Activity Level  

3. Behavioral and Emotional Responses  
   a. Emotional/Social Responses  
   b. Behavioral Outcomes of Sensory Processing  
   c. Threshold for Response  

Reliability and validity research for the Sensory Profile was completed with over 1,200 children with and without disabilities from 1993 to 1999 across four large regions in the United States. A separate standardization process was completed for the Infant/Toddler Sensory Profile from 2000-2001 and included more than 1,100 infants and toddlers, and the Adolescent/Adult Sensory Profile which included 615 people from 17-79 years.  

Throughout all three versions of the test, the reliability and validity appear to be accurate and are explained in full in three separate technical reports. Table 6 highlights the statistics of all three versions of the Sensory Profile.
Table 6

*Sensory Profile Reliability and Validity*

<table>
<thead>
<tr>
<th>Reliability &amp; Validity</th>
<th>Sensory Profile</th>
<th>Infant/Toddler Sensory Profile</th>
<th>Adolescent/Adult Sensory Profile</th>
</tr>
</thead>
<tbody>
<tr>
<td>Internal Consistency (Coefficient Alpha)</td>
<td>.47-.91</td>
<td>.42-.86 (7-36 months).17-.83 (0-6months)</td>
<td>.64-.78</td>
</tr>
<tr>
<td>Test-Rest Reliability</td>
<td>None reported</td>
<td>.86 (sensory processing) .74 (quadrants)</td>
<td>None reported</td>
</tr>
<tr>
<td>Content Validity</td>
<td>Literature and expert panel review 80% agreement between therapists</td>
<td>Literature and expert panel review</td>
<td>Literature and expert Panel Review</td>
</tr>
<tr>
<td>Convergent &amp; Discriminant Validity</td>
<td>Correlated to the <em>School Function Assessment</em></td>
<td>Correlated to the <em>Infant/Toddler Symptom Checklist</em></td>
<td>Correlated to the <em>NYLS Adult Temperament Questionnaire</em></td>
</tr>
</tbody>
</table>

Dunn (2008) reported reliability statistics for internal consistency (Cronbach’s coefficient alpha) for all three versions of the *Sensory Profile*, which measures the homogeneity of item content. Evidence of test-retest reliability also speaks to the stability of the test, whereas, content validity relates to the sampling adequacy and the expert judgment based on the theoretical relationships of variables. Finally, clinical group studies with children with disabilities, including Autism, attention deficit-hyperactivity disorder, and sensory integrative dysfunction provide evidence that this instrument is useful as part of a comprehensive assessment process.
**Demographic questionnaire.**

The short demographic questionnaire (developed by the author) sought to obtain relevant information pertaining to the participants, from mothers, their children with FXS and their families. The description of participants at the beginning of this chapter was collected through this demographic questionnaire. The questions were demographic in nature, and yielded information below. Please see Appendix A for a copy of the demographic questionnaire.

1. Maternal age
2. Level of education
3. Employment status
4. Family income
5. Maternal and child ethnicity
6. Marital status
7. Age and gender of the child with FXS
8. Age of child when FXS diagnosis was made
9. Other diagnoses of the child with FXS
10. Other children in the home with and without FXS
11. Family transition processes
12. Involvement with FXS support groups

All of these instruments were carefully chosen and reflect the characteristics of children diagnosed with FXS in the literature, and their family relationships. The constructs (maternal well-being, behavior challenges, and sensory processing) in each of the instruments have been studied in the fragile X field and connected to the
characteristics associated with FXS (Bailey et al., 2000; Epstein et al., 2002b; P. J. Hagerman, 2002; Hooper, Hatton, Baranek, Roberts, & Bailey, 2000; Miller et al., 1999). However, these constructs have yet to be explored in relation to expressed emotion measured by the Five Minute Speech Sample. It is the belief of this author that each of these instruments provide insight into the relationship between expressed emotion and the behaviors of children with FXS and their mothers.
Chapter Three: Results

Two primary questions guided the research for this dissertation. (1) What is the relationship between the expressed emotion of mothers of children with fragile X syndrome (FXS) and their perception of their children’s behavioral challenges? (2) What is the relationship between the EE score on the Five Minute Speech Sample of mothers of children with FXS and their perception of their children’s sensory processing? While both of these research questions provide information about the relationship between EE and perceptions of behavior, they also provide insight into the nature of the construct of expressed emotion with mothers of children with FXS. Researchers have studied the construct of expressed emotion in many populations, but there is a lack of expressed emotion research within the FXS population. As noted in the literature review, children with FXS have significant challenges with behavior and sensory processing from early childhood through adulthood (Epstein et al., 2002a; Hagerman, 2006a; Hatton et al., 2002; Sullivan et al., 2007). In addition, high maternal expressed emotion has been associated with relapses in mild to severe mental illness disorders, including young children with challenging behaviors (Boger et al., 2008; Peris & Baker, 2000), emotional and anxiety disorders (G. W. Brown et al., 1996; Calam & Peters, 2006), and schizophrenia (Lenior et al., 2002; McCarty & Weisz, 2002b). To address the relationship between these variables, correlational analysis was completed between the measurement of EE, the Five Minute Speech Sample (FMSS) (Magana-Amato, 2001)
and the perception of children’s’ behavior through the Behavior Assessment System for Children (BASC) (Reynolds & Kamphaus, 2004) and sensory processing through the Sensory Profile (Dunn, 1999).

The organization of this results chapter first addresses the descriptive statistics of the measures, followed by inferential statistics. The descriptive and correlational statistics begin to tell the story of the expressed emotion of mothers of children with FXS. An alpha level of .05 was used for all statistical tests.

**Five Minute Speech Sample (FMSS) Descriptive Statistics**

The primary variable in this study is the expressed emotion of mothers of children with FXS as measured by the Five Minute Speech Sample (FMSS) and yields a dichotomous EE score of High or Low (Magana-Amato, 2001). The score also yields a sub-rating score of criticism (CRIT) and/or emotional overinvolvement (EOI), which can be a rating of High, Low, or Borderline. Thirty-five mothers completed the FMSS in person (n = 14) during home visits or FXS clinic consultations or by phone (n = 21). There were no significant differences in EE scores between the mothers who completed the FMSS in person or by phone ($F = 3.831, p > .05$). Therefore, location of data collection was not found to be a confounding variable.

There are five scoring categories of the FMSS that include Initial Statement, Relationship, Criticisms, Dissatisfaction, and Emotional Overinvolvement. The first two categories of the FMSS, initial statement and relationship, were scored discretely as **positive, neutral, or negative** (Magana-Amato, 2001). The initial statement is an essential component and provides initial thoughts about the relationship between the
respondent and his/her relative. For this study, the relationship is between the mother and her child with FXS. Seventy-seven percent of the mothers scored “neutral” initial statements, typified by a factual description of their child’s age and diagnosis of FXS. For example, the mothers would say, “My son is 13 years old and has fragile X syndrome.” The remaining mothers provided “positive” initial statements, expressing a general positive remark about their child, such as “Betsy is a wonderful person. She is the sweetest person.”³ None of the mothers expressed a “negative” initial statement.

The relationship within the FMSS is an overall rating within the measure, which is also coded as positive, neutral, or negative. Table 7 shows that a majority of relationship codes in the sample were “positive,” followed by 40% “neutral,” and only 6% negative. As stated above, a positive, neutral, or negative relationship was determined by statements that speak to the “quality of the relationship,” such as the quality of communication between the dyad and shared activities.

Several mothers talked about their “special relationship” with their children with FXS. Often mothers reported they had the closest relationship with their child, regardless of other family members (including fathers), even though they described themselves as the disciplinarian, while the fathers “had all of the fun.” The relationship was described as very close with mutual admiration and enjoyment. Emerging themes from the relationship included heartening statements that clearly established quality time together that led to positive relationships. The following are examples of statements that demonstrate the dyads’ positive relationships:

³ All names used in quotes from the FMSS have been changed to protect the identity of the participants.
• “I enjoy playing with him and reading books with him.”
• “We sing together.”
• “We have a special bond.”
• “We have a unique communication with each other.”
• “We are very in sync with each other.”
• “We are attached at the hip.”

Table 7

<table>
<thead>
<tr>
<th>FMSS Variable</th>
<th>Positive</th>
<th>Neutral</th>
<th>Negative</th>
</tr>
</thead>
<tbody>
<tr>
<td>Initial Statement</td>
<td>23% (n=8)</td>
<td>77% (n=27)</td>
<td>0%</td>
</tr>
<tr>
<td>Relationship</td>
<td>55% (n=19)</td>
<td>40% (n=14)</td>
<td>6% (n=2)</td>
</tr>
</tbody>
</table>

*Note. N = 35*

Table 8 illustrates FMSS categories coded as present or absent throughout the entire speech sample, although the EOI subcategory of Self-Sacrificing Overprotective Behavior (SSOP) can be coded as a borderline, with several “hints” or evidentiary statements indicating the behavior may be present. SSOP represents statements by respondents in which they make sacrifices for the child, regardless of the impact on themselves. Some of the statements even though they may reflect a positive relationship, indicate a lack of boundaries between the mother and child, and a loss of individuality of the child and mother. Attachment theory supports this notion of the development of an intense bond between mother and child; although as the child ages and then begins to distinguish him/herself from the mother both physically and emotionally. For a child
with special health care or development needs, it is often hard for the mother to let the child explore and learn from his/her environment due to the development challenges. So the mother is cautious about letting her child develop the independent skills even though the mother remains the “secure base” for the child (Bowlby, 1982). The following statements are examples of hints of SSOP statements that were coded High or Borderline.

- “He does everything with me, and goes everywhere with me.”
- “I had to stay home a lot because I couldn’t handle him.”
- “I spend a lot of time worrying about him.”
- “Everything went into helping him.”

Many mothers said that their lives were chaotic and frustrating, but there was little evidence that they were truly critical or dissatisfied with the behavior of their children. They understood the impact of FXS and built their daily lives around the strengths and challenges of their children. One mother stated, “I understand her because I have fragile X myself.” Statements of dissatisfaction were primarily centered on the child’s ability to “push their mother’s buttons” with challenging behaviors related to sensory overstimulation. Interestingly, mothers reported that their children’s aggressive behavior was targeted at them and not other family members, such as this statement, “Sometimes I get hurt from his aggressive behavior, but that is ok.” Therefore, the perception of the mothers is that their children with FXS are closer to them, which allows them to feel comfortable enough in their relationship to be targets of their aggression.
<table>
<thead>
<tr>
<th>FMSS Variable</th>
<th>Present</th>
<th>Absent</th>
<th>Borderline</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dissatisfaction</td>
<td>31% (n=11)</td>
<td>65% (n=24)</td>
<td></td>
</tr>
<tr>
<td>Emotional Display</td>
<td>0%</td>
<td>100% (n=35)</td>
<td></td>
</tr>
<tr>
<td>Excessive Detail</td>
<td>3% (n=1)</td>
<td>97% (n=34)</td>
<td></td>
</tr>
<tr>
<td>Self-Sacrificing Over</td>
<td>3% (n=1)</td>
<td>78% (n=29)</td>
<td>14% (n=5)</td>
</tr>
<tr>
<td>Protective Behavior</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Note. N = 35

Lastly, Table 9 illustrates FMSS subcategories that are continuous by the use of a frequency count taken over the entire speech sample. There were very few criticisms within this sample, but examples are, “I get angry with him sometimes; he knows how to push buttons and when he is frustrated he will do things that are so irritating.” Another example of a critical statement was, “Her biggest problem is just not being so lazy and getting up and doing things when we ask her too.” Criticisms are determined by content and tone, indicating some level of frustration or unfavorable view of the child.

A mother’s love for her child with FXS cannot be disputed, and Statements of Attitude are an expression of that intense love. There were several Statements of Attitude articulated by the mothers which were coded as either present or absent (Statements of Attitude is a subcategory of Emotional Over-involvement). The reason these statements are coded as a subcategory of EOI is that within a brief period (5 minutes), the mother verbalized statements that would indicate intense love for the child and the willingness to
do anything to help the child. Such statements included, “I love him very much,” “She is my angel,” and “I love him more than anyone in the world.”

The mothers in this sample had very positive things to say about their children. They were truly proud of their accomplishments, such as adjusting to overstimulating experiences, taking care of themselves, and communicating with peers. There was a cumulative count of 25 Positive Remarks made by mothers. The coding of “Positive Remarks” also falls under the EOI subcategory and encompasses a curvilinear result because of the frequency count. More specifically, zero to four Positive Remarks do not affect the EOI rating, although when participants express five or more Positive Remarks within five minutes, a high EOI rating results. Within this sample, the range of Positive Remarks was from 0-11, with an average of two. Five Positive Remarks is the tipping point when the coding changes from low to high. Positive Remarks include statements such, “He is a very sweet child,” “she is very sensitive,” “he has the best laugh ever,” and “he is a great sense of humor.”

Table 9

*Descriptive Statistics for Continuous Codes of FMSS*

<table>
<thead>
<tr>
<th>FMSS Variable</th>
<th>Frequency</th>
<th>Range</th>
<th>Mean</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td>Criticism</td>
<td>2</td>
<td>0-1</td>
<td>.06</td>
<td>.236</td>
</tr>
<tr>
<td>Statements of Attitude</td>
<td>7</td>
<td>0-2</td>
<td>.31</td>
<td>.676</td>
</tr>
<tr>
<td>Positive Remarks</td>
<td>25</td>
<td>0-11</td>
<td>2.11</td>
<td>2.386</td>
</tr>
</tbody>
</table>

*Note.* $N = 35$
Final FMSS expressed emotion rating.

The outline in Appendix A represents the criteria for assigning the final EE ratings taking into account the subcategory ratings. The final EE ratings are dichotomous, either High or Low, although the Low EE rating may encompass a Borderline Critical or Borderline EOI rating. Magana-Amato (2001), the FMSS developer, stated in the guidelines that with “lower threshold” populations, relatives might be hesitant to reveal intense feelings about their children; therefore, a Borderline rating can be reassigned as High EE. Other studies have used this rule when conducting FMSS research with mothers of young children, stating “Borderline EE should be counted as High EE in samples where parents may be reluctant to express strong attitudes of criticism or overinvolvement about a relative” (Jacobson et al., 2000, p. 901).

Unfortunately, this rule is sparsely cited within the two articles and the FMSS guidelines (Jacobsen et al., 2000; Stubbe, Zahner, Goldstein, & Leckman, 1993). Taking the lead from prior research and the FMSS guidelines, the author has also applied this rule to the FXS population because mothers of children with FXS may be more guarded in their expressions of criticisms or negative statements toward their children. This bias toward non-critical statements may be a reflection of cultural norms when talking about children with intellectual disabilities in which criticisms may seem unfair or unloving.

Additionally, mothers of children with FXS are keenly aware that their children have inherited the FMR1 gene mutation from themselves and feel guilt about their child’s condition, which may also cause the reluctance of expressed emotion. To that end, a lower threshold for EE ratings was applied. Although this rule has been applied,
descriptive and inferential statistics are reported with and without the application of the rule to show how the rule influenced the results.

**FMSS subgroup ratings.**

The descriptive statistics for the EE subgroup ratings are broken down into eight groups, taking into account all of the possible rating combinations. The subgroups’ ratings are assigned before the final EE ratings and involve the subgroups of Low, Critical, and Emotional Overinvolvement (EOI), because the subgroup rating will determine the final EE rating. Figure 3 below illustrates that 37% of the mothers in the sample were rated Low, followed by 27% rated as Borderline EOI. All Borderline EOI and Critical subgroups together constitute 52% (n=18) of the sample. There were no mothers who were rated with the combination of Critical and Borderline EOI (CRIT & b/EOI).

![EE Subgroup](image)

*Figure 3. Descriptive Statistics for EE Subgroups*
The first set of analyses encompasses final EE ratings with a dichotomous High/Low rating reveal that 86% (n=30) of the sample were rated Low EE, while only 14% (n=5) were rated High EE. Figure 4 below illustrates the final FMSS rates absent the borderline rule.

**Figure 4.** Final FMSS EE Ratings absent the Borderline Rule

In Figure 4 above, the final Low ratings included the Borderline ratings and when broken down further, 37% (n=13) of the mothers were rated Low EE, 11% (n=4) were rated High EE, and 51% (n=18) were rated Borderline EE (EOI and Critical together). Figure 5 illustrates all ratings, including High, Low, and Borderline.
Figure 5. Final FMSS EE Ratings with Borderline Rating

Alternatively, when applying the Borderline rule regarding “low-threshold” populations for a dichotomous High/Low EE rating, 57% of the sample (n=20) were rated High EE, and 43% (n=15) were rated Low EE (see Figure 6).

Figure 6. Final FMSS EE Rating with Borderline Rule Applied
Clearly, a majority of mothers fall within the Borderline EE rating, particularly from the Emotional Overinvolvement subgroup as reference in Table 1. Even within this small sample, a trend of Emotional Overinvolvement is emerging for mothers of children with FXS. This trend will be discussed further in the discussion chapter.

**Behavior Assessment System for Children (BASC) Descriptive Statistics**

The Behavior Assessment System for Children (BASC) is a well-known comprehensive assessment used to evaluate the behavior of children and young adults (Reynolds & Kamphaus, 2004). This effective tool measures the behavior of children in school and home environments. The BASC encompasses individual scales and composite scores that measure externalizing and internalizing behavioral characteristics, along with adaptive behavior and functional communication. \( T \)-scores classify the scores on the adaptive and clinical scales, ranging from clinically significant on the lower end to clinically significant on the higher end.

Thirty-two mothers completed this self-administered behavior assessment, and results of the individual and component scales are reported below. There was missing data from four of the participants because of the lack of return of questionnaires after the completion of the Five Minute Speech Sample.

Descriptive statistics for all completed BASCs resulted in variability of classifications within the individual and component scales. The BASC is broken down into Externalizing and Internalizing component scales. The mean for each of the scales varies from the average classification to very high and very low (see Table 5 in Chapter 2 for reference). Of particular interest, the Hyperactivity scale (within the Externalizing
component), defined in the BASC guidelines as “The tendency to be overly active, rush through work activities, and act without thinking” (p. 60) resulted in a mean of 62.53, which is in the “High” classification for the Adaptive scales and “Clinically Significant” for the Clinical scales. This was consistent with prior research on attention and hyperactivity disorders and children with FXS, which indicated that many children with FXS, especially boys are diagnosed with ADHD (K. Cornish, Sudhalter, & Turk, 2004). Additional Externalizing scales such as Aggression (M = 51.09) and Conduct Problems (M = 48.64) had means in the “average” range. The mean of the Externalizing composite scores also fell within the “average” range (M = 54.62).

The Internalizing scales (i.e. Anxiety, Depression, and Somatization), were within the “average” range including the Internalizing composite score of 52.62, although there is a good deal variability within the sample (SD=12.73). Conversely, the scales of Atypicality (M=66.53), and Attention Problems (M=65.00) were in the “high” classification, and Withdrawal (M=70.62) was in the “very high” classification. The composite score of the Behavioral Symptoms Index (BSI), “reflects the overall level of problem behavior” (p. 67), resulted in a mean of 65.25 and a “high” classification. The BSI includes the Hyperactivity, Aggression, Depression, Attention Problems, Atypicality, and Withdrawal scales.

The Adaptive Skills composite includes the scales of Adaptability, Activities of Daily Living, Functional Communication, Social Skills, and Leadership. The means for these scales resulted in a low trend from “Clinically Significant” for Functional Communication (M=25.81) to the relatively highest mean in the group of 38.50 for Social
Skills which is still in the “low” classification. The Adaptive Skills composite mean was also in the “low” classification with a mean of 31.00. Table 10 below displays the descriptive statistics of the BASC.

Table 10

**BASC Descriptive Statistics**

<table>
<thead>
<tr>
<th>BASC Variables</th>
<th>N</th>
<th>Minimum</th>
<th>Maximum</th>
<th>Mean</th>
<th>SD</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hyperactivity</td>
<td>32</td>
<td>44</td>
<td>80</td>
<td>62.53</td>
<td>10.70</td>
</tr>
<tr>
<td>Aggression</td>
<td>32</td>
<td>38</td>
<td>79</td>
<td>51.09</td>
<td>9.69</td>
</tr>
<tr>
<td>Conduct Problems</td>
<td>28</td>
<td>40</td>
<td>73</td>
<td>48.64</td>
<td>8.07</td>
</tr>
<tr>
<td>Externalizing</td>
<td>32</td>
<td>41</td>
<td>80</td>
<td>54.62</td>
<td>8.62</td>
</tr>
<tr>
<td>Anxiety</td>
<td>32</td>
<td>32</td>
<td>80</td>
<td>48.84</td>
<td>10.91</td>
</tr>
<tr>
<td>Depression</td>
<td>32</td>
<td>39</td>
<td>99</td>
<td>54.56</td>
<td>12.49</td>
</tr>
<tr>
<td>Somatization</td>
<td>32</td>
<td>39</td>
<td>76</td>
<td>52.78</td>
<td>11.14</td>
</tr>
<tr>
<td>Internalizing</td>
<td>32</td>
<td>38</td>
<td>89</td>
<td>52.62</td>
<td>12.73</td>
</tr>
<tr>
<td>Atypicality</td>
<td>32</td>
<td>45</td>
<td>105</td>
<td>66.53</td>
<td>12.39</td>
</tr>
<tr>
<td>Withdrawal</td>
<td>32</td>
<td>43</td>
<td>91</td>
<td>70.62</td>
<td>10.79</td>
</tr>
<tr>
<td>Attention Problems</td>
<td>32</td>
<td>41</td>
<td>82</td>
<td>65.00</td>
<td>8.68</td>
</tr>
<tr>
<td>BSI</td>
<td>32</td>
<td>42</td>
<td>93</td>
<td>65.25</td>
<td>8.94</td>
</tr>
<tr>
<td>Adaptability</td>
<td>32</td>
<td>21</td>
<td>58</td>
<td>37.47</td>
<td>9.03</td>
</tr>
<tr>
<td>Social Skills</td>
<td>32</td>
<td>20</td>
<td>63</td>
<td>38.50</td>
<td>10.83</td>
</tr>
<tr>
<td>Leadership</td>
<td>28</td>
<td>20</td>
<td>40</td>
<td>30.00</td>
<td>4.43</td>
</tr>
<tr>
<td>Daily Living</td>
<td>29</td>
<td>15</td>
<td>50</td>
<td>31.24</td>
<td>9.27</td>
</tr>
<tr>
<td>Functional</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Communication</td>
<td>32</td>
<td>11</td>
<td>47</td>
<td>25.81</td>
<td>8.78</td>
</tr>
<tr>
<td>Adaptive Skills</td>
<td>32</td>
<td>16</td>
<td>54</td>
<td>31.00</td>
<td>9.85</td>
</tr>
</tbody>
</table>

The BASC scores consisted of high/at-risk Hyperactivity; low/at-risk Adaptive Skills, and “average” Internalizing and Externalizing scales, but the standard deviations
showed variability within the sample. Therefore, the “typical” child with FXS within this sample would be characterized as hyperactive, with low functional communication, daily living, and social skills, but not particularly aggressive or defiant. This “typical” child would also be portrayed as a child that is somewhat withdrawn, with atypical behaviors and attention challenges. This profile is consistent with prior research that characterized children with FXS with a variety of behaviors consistent with internalizing and externalizing behaviors mentioned above (K. Cornish et al., 2008; Hagerman, 2006a; Hatton et al., 2002; Kau et al., 2004).

**Sensory Profile Descriptive Statistics**

*The Sensory Profile* (SP) is a caregiver questionnaire designed to elicit feedback from caregivers about their children’s reactions to everyday sensory stimulation and processing. The caregivers, or the mothers within this study, responded to questions about how frequently their children exhibited behaviors in response to a continuum of sensory processing. The SP consists of three sections 1) Sensory Processing; 2) Modulation and Behavior; and 3) Emotional Responses. Each of the sections contains subsections breaking down the behavioral reactions to sensory stimulation even further. Cutoff scores determine the classification for each of the sections, which include 1) Typical Performance, 2) Probable Difference, and 3) Definite Difference. The mean of all of the subcategories collected from data within this study are classified as “Probable Difference” with a range of all three classifications (Dunn, 1999). Due to the rapid development of children ages birth to three, Dunn (1999) developed the *Infant/Toddler Sensory Profile* (ITSP), and encouraged users to complete the profile consistent with the
ages of children they were evaluating for the purposes of reliability and validity. Therefore, two different SP’s were used because of the wide age of range within this study; *The Sensory Profile* and the *Infant/Toddler Sensory Profile*.

Table 11 below shows the descriptive statistics for 31 children and adolescents above the age of four years whose mothers complete *The Sensory Profile*. Individual items were placed into one of nine factors listed below and yields classifications of Typical Performance, Probable Difference, or Definite Difference. The mean scores below reflect that a majority of mothers answered questions that would place their children within the “Probable Difference” category due to their perception of their children’s sensory processing. The variation in N reflects incomplete questions by participants.

Table 11

*Descriptive Statistics of the Sensory Profile Factor Summary*

<table>
<thead>
<tr>
<th>SP Variables</th>
<th>N</th>
<th>Minimum</th>
<th>Maximum</th>
<th>Mean</th>
<th>SD</th>
<th>Classification</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sensory Seeking</td>
<td>28</td>
<td>47</td>
<td>84</td>
<td>65.75</td>
<td>11.800</td>
<td>Probable Difference</td>
</tr>
<tr>
<td>Emotionally Reactive</td>
<td>30</td>
<td>26</td>
<td>62</td>
<td>46.97</td>
<td>10.311</td>
<td>Probable Difference</td>
</tr>
<tr>
<td>Low Endurance</td>
<td>31</td>
<td>21</td>
<td>45</td>
<td>34.65</td>
<td>6.586</td>
<td>Probable Difference</td>
</tr>
<tr>
<td>Oral Sensation</td>
<td>29</td>
<td>11</td>
<td>45</td>
<td>33.34</td>
<td>10.926</td>
<td>Probable Difference</td>
</tr>
<tr>
<td>Inattention</td>
<td>30</td>
<td>11</td>
<td>31</td>
<td>22.43</td>
<td>4.352</td>
<td>Probable Difference</td>
</tr>
<tr>
<td>SP Variables</td>
<td>N</td>
<td>Minimum</td>
<td>Maximum</td>
<td>Mean</td>
<td>SD</td>
<td>Classification</td>
</tr>
<tr>
<td>-----------------</td>
<td>----</td>
<td>---------</td>
<td>---------</td>
<td>--------</td>
<td>-------</td>
<td>----------------------</td>
</tr>
<tr>
<td>Poor Regulation</td>
<td>31</td>
<td>15</td>
<td>40</td>
<td>31.48</td>
<td>5.328</td>
<td>Probable Difference</td>
</tr>
<tr>
<td>Sensitivity</td>
<td>30</td>
<td>5</td>
<td>20</td>
<td>15.20</td>
<td>4.468</td>
<td>Probable Difference</td>
</tr>
<tr>
<td>Sedentary</td>
<td>30</td>
<td>4</td>
<td>19</td>
<td>10.57</td>
<td>3.775</td>
<td>Probable Difference</td>
</tr>
<tr>
<td>Perceptual</td>
<td>31</td>
<td>3</td>
<td>14</td>
<td>7.61</td>
<td>3.127</td>
<td>Probable Difference</td>
</tr>
</tbody>
</table>

Figure 7 below illustrates the range of SP factor summary classifications within the whole sample and emphasizes that a majority of factor summary scores were in the probable difference classification.

![SP Factor Summary Classifications](image)

*Figure 7. Range of Sensory Profile Factor Summary Classifications*
**Infant-toddler Sensory Profile.**

Only two mothers in the sample had children young enough to use the *Infant/Toddler Sensory Profile* (ITSP). Both young children varied in their classification from Typical Performance to Definite Difference. In the category of Low Regulation, one child was classified in the Definite Difference category, while the other was classified in Probable Difference. Both children were classified in the Typical Performance category for Sensory Sensitivity, and one child for Sensation Avoidance. Table 12 illustrates the scores for these two young children.

Table 12

*Descriptive Statistics for the Infant/Toddler Sensory Profile Factor Summary*

<table>
<thead>
<tr>
<th>ITSP Variables</th>
<th>N</th>
<th>Minimum</th>
<th>Maximum</th>
<th>Mean</th>
<th>SD</th>
<th>Classification</th>
</tr>
</thead>
<tbody>
<tr>
<td>Low Regulation</td>
<td>2</td>
<td>37</td>
<td>55</td>
<td>46.00</td>
<td>12.728</td>
<td>Definite &amp; Probable Difference</td>
</tr>
<tr>
<td>Sensation Seeking</td>
<td>2</td>
<td>29</td>
<td>51</td>
<td>40.00</td>
<td>15.556</td>
<td>Probable &amp; Definite Difference</td>
</tr>
<tr>
<td>Sensory Sensitivity</td>
<td>2</td>
<td>45</td>
<td>50</td>
<td>47.50</td>
<td>3.536</td>
<td>Typical Performance</td>
</tr>
<tr>
<td>Sensation Avoidance</td>
<td>2</td>
<td>41</td>
<td>55</td>
<td>48.00</td>
<td>9.899</td>
<td>Probable Difference &amp; Typical Performance</td>
</tr>
</tbody>
</table>
**Sensory Profile section summary.**

The Sensory Profile Section Summary encompasses the three overall sections mentioned above 1) Sensory Processing, 2) Modulation and Behavior, and 3) Emotional Responses. Within each of the sections are subsections that evaluate the children’s responses to specific sensory stimulation, such as Auditory, Visual, Vestibular, Touch, Multisensory, and Oral. The Modulation and Behavior section evaluates the behavior children exhibit in response to sensory stimulation in terms of organization and appropriateness to the stimuli. The subsections are Modulation of Tone, Movement, Activity, Emotional, and Visual. Finally, the section of Emotional Responses provides evidence of behavioral outcomes in relation to sensory stimulation and processing. The subsections are Emotion-Social, Behavioral Outcomes, and Threshold (Dunn, 1999). Taken together, all three of these sections provide a profile of the variability of sensory responses to everyday sensory stimulation that children encounter at home, school, and in their community. The following three figures (Figure 8, 9, and 10) provide a classification breakdown for each of those sections.
Figure 8. The Sensory Profile—Sensory Processing Summary

Figure 9. The Sensory Profile—Modulation & Behavior Summary
As the figures above illustrate, there is variability in sensory processing for children with FXS, as reported by their mothers in this sample. Notably, a majority (49%) were classified in the Definite Difference category for Touch, which is consistent with research on children with FXS, and their aversion to tactile stimulation (Scharfenaker, O'Connor, Stackhouse, Braden, & Gray, 2002b). Also within the Definite Difference classification were Modulation of Tone (52%) and Behavioral Outcomes (68%). Children with FXS often have trouble modulating their sensory experiences and behavioral responses, especially in relation to tone in which they may be required to be seated and stay alert for periods of time at school, in therapy, or at family/community functions (Dunn, 1999; Symons et al., 2003; Weber, 2000b).
Correlations

FMSS & BASC.

The first research question addressed the relationship between the expressed emotion of mothers of children with FXS and the perception of their children’s behavioral characteristics, measured by the Five Minute Speech Sample (FMSS) (Magana-Amato, 2001) and the Behavior Assessment System for Children (BASC) (Reynolds & Kamphaus, 2004). The overall ratings of the FMSS (High/Low/Borderline) resulted in no significant correlations with the BASC summary scores, indicating no omnibus relationship between the expressed emotion of mothers with children with FXS and the perceptual reporting of their children’s behavior. Further analysis revealed significant correlations between two of the subcategories of the FMSS and one subcategory of the BASC. There was a significant, positive relationship between the FMSS subcategory, Self-Sacrificing Over Protective Behavior and the BASC’s subcategory, Atypicality (r=.517, p<.05). This relationship suggested that mothers of children with FXS were more likely to be overprotective and sacrifice their own needs and wants when they viewed their children’s behavior as atypical (i.e. displaying odd or immature behaviors). There was a significant, negative relationship between the FMSS subcategory, Positive Remarks with again the BASC’s subcategory Atypicality (r= -.370, p<.05). This negative relationship indicated that mothers made fewer positive remarks about their children the more atypical they perceived their behaviors. To that end, both of these correlations suggested that when mothers perceived their children’s behavior as atypical, they expressed more overprotection and self-sacrifices and few positive
remarks. Figure 11 illustrates the linear relationship between the FMSS Positive Remarks and the BASC’s Atypicality, although there is one outlier of a significant number of positive remarks that may add to the correlation statistic.

![Graph showing the negative correlation between Positive Remarks and Atypicality]

**Figure 11.** Negative Correlation between Positive Remarks and Atypicality

A negative relationship was also found between the number of Positive Remarks expressed by mothers and the Behavioral Symptoms Index of the BASC (r. -.464, p < .05) indicating that the more positive remarks mothers expressed, the less challenging behaviors they perceived their children to exhibit, including internalizing and externalizing behaviors. The Figure 12 demonstrates the linear relationship between the variables, including the outlier mentioned previously.
Figure 12. Negative Correlation between Positive Remarks and Behavioral Symptoms Index

**FMSS & Sensory Profile.**

The second research question involved the variables of expressed emotion measured by the Five Minute Speech Sample (FMSS) and sensory processing measured by the Sensory Profile (Dunn, 1999; Magana-Amato, 2002). Results indicate no significant relationship between the overall EE ratings and summary sections of the Sensory Profile, although there were several significant relationships between subcategories of the FMSS and subsections of the Sensory Profile. The Table 13 illustrates the significant correlations between FMSS subcategories and continuous factor raw scores.
Table 13

*Correlations between FMSS and Sensory Profile Factor Raw Scores*

<table>
<thead>
<tr>
<th>FMSS/SP Variables</th>
<th>Initial Statement</th>
<th>Positive Remarks</th>
<th>State of Attitude</th>
<th>SSOP</th>
<th>Dissatisfaction</th>
</tr>
</thead>
<tbody>
<tr>
<td>Low Endurance</td>
<td>.41*</td>
<td>-.15</td>
<td>-.51**</td>
<td>-.18</td>
<td>-.12</td>
</tr>
<tr>
<td>Sedentary</td>
<td>-.10</td>
<td>.45*</td>
<td>-.38*</td>
<td>-.21</td>
<td>-.03</td>
</tr>
<tr>
<td>Vestibular</td>
<td>-.26</td>
<td>-.22</td>
<td>-.42*</td>
<td>-.04</td>
<td>.18</td>
</tr>
<tr>
<td>Auditory</td>
<td>-.01</td>
<td>.535**</td>
<td>.07</td>
<td>-.46**</td>
<td>.02</td>
</tr>
</tbody>
</table>

Note. * Correlation is significant at the .05 (2-tailed), ** Correlation is significant at the .005 (2-tailed)

Another way to look at this data is to examine the correlations between the FMSS subcategories and the discrete classifications of the SP Factor Summary (i.e. typical performance, probable difference, and definite difference). Table 14 shows the correlations between these factors.

Table 14

*Correlations between FMSS and Sensory Profile Factor Summary Classifications*

<table>
<thead>
<tr>
<th>FMSS/SP Variables</th>
<th>Initial Statement</th>
<th>Positive Remarks</th>
<th>State of Attitude</th>
<th>SSOP</th>
<th>Dissatisfaction</th>
</tr>
</thead>
<tbody>
<tr>
<td>Low Endurance/Tone</td>
<td>.46*</td>
<td>.09</td>
<td>.31</td>
<td>.24</td>
<td>-.13</td>
</tr>
<tr>
<td>Inattention/Distractibility</td>
<td>.10</td>
<td>-.14</td>
<td>.19</td>
<td>.45</td>
<td>*-.03</td>
</tr>
<tr>
<td>Sensory Sensitivity</td>
<td>.13</td>
<td>-.17</td>
<td>.41*</td>
<td>.08</td>
<td>-.39*</td>
</tr>
<tr>
<td>Sedentary</td>
<td>.01</td>
<td>-.41*</td>
<td>.36</td>
<td>.24</td>
<td>-.09</td>
</tr>
</tbody>
</table>

Note. * Correlation is significant at the .05 (2-tailed), ** Correlation is significant at the .005 (2-tailed)
**FMSS and demographics.**

Lastly, analyses provided significant relationships between the EE variables and demographic variables. Notably, the subcategories in the FMSS correlated with the categorical demographic variables. There was a positive relationship between the Initial Statement (coded as positive-1, neutral-2, and negative-3) and the mothers’ age ($r = .370$) indicating that younger mothers expressed more positive initial statements than older mothers. Marital status also was negatively correlated with a number of FMSS subcategories such as Criticisms, Self-Sacrificing Over Protective Behavior (SSOP), and Emotional Overinvolvement (EOI). This relationship suggests that mothers who were married (85%) were more likely to have fewer Criticisms, Self-Sacrificing Over Protective Behavior, and Emotional Overinvolvement, although, it may be due to the lack of variability in marital status. Ethnicity was another factor negatively correlated to FMSS subcategories, again possibly due to the lack of diversity in the sample.

One relationship that was approaching significance was the number of additional diagnoses to FXS and the rating of Borderline EOI ($r = .316$, $p = .084$). This possible relationship may indicate mothers are more likely to become more emotionally over-involved when their children receive additional diagnoses. Table 15 shows the significant relationships between the FMSS subcategories and demographic variables.
Table 15

*Correlations between FMSS Subcategories and Demographics*

<table>
<thead>
<tr>
<th>Variables</th>
<th>Initial Statements</th>
<th>Criticism</th>
<th>SSOP</th>
<th>Positive Remarks</th>
<th>EOI</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mothers’ Age</td>
<td>.37*</td>
<td>.04</td>
<td>.16</td>
<td>-.29</td>
<td>-.22</td>
</tr>
<tr>
<td>Marital Status</td>
<td>.00</td>
<td>-.48**</td>
<td>-.62**</td>
<td>-.01</td>
<td>-.38*</td>
</tr>
<tr>
<td>Ethnicity</td>
<td>-.15</td>
<td>.07</td>
<td>.11</td>
<td>-.49**</td>
<td>-.44*</td>
</tr>
</tbody>
</table>

*Note.* * Correlation is significant at the .05 (2-tailed), ** Correlation is significant at the .005 (2-tailed)

**Conclusion.**

In this chapter, descriptive and inferential statistics were reported. There were interesting findings for each of the measures, particularly within the FMSS in which descriptive statistics provided a profile of how mothers of children with FXS talk about their children. Overall, EE findings revealed that mothers of children with FXS had positive relationships with their children and many were emotionally over-involved. This finding is not surprising; however, it has not been documented.

Descriptive statistics from the BASC and Sensory Profile also provided a picture of how mothers with children with FXS perceive their children’s behavior and sensory processing. On average, mothers reported that their children with FXS exhibited hyperactive behaviors, low functional communication, and limited daily living and social skills. Mothers also reported sensory processing problems that affect behavior, such as modulation and tactile sensitivity. This profile is consistent with the literature describing the behavioral phenotype of children with FXS (Dykens et al., 1996; Hagerman, 2006a; Hagerman et al., 2008; Hatton et al., 2002; Lachiewicz & Mirrett, 2000).
Additionally, results of the research questions were reported with Pearson $r$ correlations between instrument and demographic variables. The results provided a clear picture of the sample and analyses of the relationship of variables. There were no significant omnibus relationships between the variables, although several interesting correlations between sub-scores and sub-sections. Of particular interest was the expressed emotion profile of mothers of children with FXS. To that end, the following chapter will present an interpretation of the results within the context of the FXS population.
Chapter Four: Discussion

This dissertation has explored the relationship between the expressed emotion of mothers of children with FXS and the behavioral and sensory characteristics of their children, as reported by the mothers, through a unique method called the Five Minute Speech Sample. To this end, descriptive and inferential statistics were completed. Results partially confirmed the argument made in the literature review that there are multiple factors affecting mother-child relationships, in particular, the way mothers talk about their children (i.e. expressed emotion). The research questions for this dissertation begin to explore the potential relationships between expressed emotion of mothers and perception of the behavioral outcomes for their children and the method opens the door to the utilization of the FMSS.

Expressed Emotion and the Five Minute Speech Sample

The majority of children with FXS have inherited the \textit{FMR1} gene from their mother and have endured an intellectual disability. The mothers are well aware of their carrier status after their child’s diagnosis, and it is this author’s belief that this knowledge limits the mothers’ expressed emotion in a formal setting with a standardized instrument, such as FMSS, and, in fact may limit their expressed emotion in general. With that in mind, the application of the “borderline rule” as described in Chapter Three, is necessary for respondents of a “lower threshold” population because they may be more “reluctant to express strong attitudes about their relatives, for example, parent of young children”
(Magana-Amato, 2002, p. 45). To clarify, if a respondent receives a borderline rating for Emotional Over-Involvement (EOI) or Critical (CRIT), then that rating would be scored as High EE as opposed to Low EE. The FMSS guidelines state “samples that receive a borderline rating would be considered a low EE-subgroup. However, with populations where a lower threshold is high—EE might be more appropriate, borderline speech samples may be considered high—EE” (p. 44). A “lower threshold population” is a population that may be more reluctant to fully express their emotions relating to the intense relationship with their child. The argument for the application of this rule to mothers of children with FXS is because of the author’s belief that these mothers have a lower threshold due to the inheritability factor of the FMR1 gene mutation that causes their children’s subsequent intellectual disability. This is consistent with other “low threshold” populations, such as mothers of very young children in that mothers of children with FXS share similar parenting responsibilities.

Expressed emotion is not a construct that has been explored in the population of families with children with FXS, nor has the method of the Five Minute Speech Sample (FMSS). The literature on expressed emotion has included families with adult children with schizophrenia, and other behavioral and medical disorders, such as eating disorders and asthma. In addition, research conducted with young children recognized the difficulty parents have expressing their emotions in full due to the evolving attachment relationship. Researchers have concluded that maternal expressed emotion measured by the FMSS was associated with the quality of attachment between mother and child and thus the behavior of the children (Jacobsen et al., 2000). There is a paucity of research on
expressed emotion and children with significant intellectual disorders, such as FXS. In many ways, there are similarities between young children and children with intellectual disabilities because both sets of children need constant care and attention as well as nurturing caregivers who provide opportunities for learning and development. There are also maternal factors that can disrupt the attachment relationship, regardless of the child’s age or ability level, such the mother’s well-being. This dissertation attempted to explore these relationships by addressing the expressed emotion of mothers of children with FXS and their perception of their children’s behavior.

**Initial statements and relationships.**

The “initial statement” of a mother reveals the foundation of her relationship with her child. This first statement sets the tone for the rest of the speech sample and contributes to the coding of the overall relationship. In this sample, a majority of mothers began the FMSS with neutral statements: in general, reporting child’s name, age, and diagnosis of FXS. There were fewer positive initial statements articulated by mothers of younger children, and no negative initial statements in the sample. The initial statements were similar to those of other mothers found in the literature. For example, mothers of young children typically began their speech samples with positive or neutral statements, while parents of adult children with schizophrenia typically began with negative statements (Butzlaff & Hooley, 1998; Daley et al., 2003; Jacobsen et al., 2000; Kwon et al., 2006).

The initial statement, while coded separately adds to the overall relationship rating. Therefore, a negative initial statement provides evidence of a negative
relationship. This study yielded only neutral and positive initial statements, which added to the overwhelming positive relationships of mothers in this sample. The positive relationships, reported by a majority of mothers were evidenced by statements about engaging in enjoyable activities with their children regardless of age. Describing mutually enjoyable activities is one of the criteria for a “positive relationship” in the FMSS guidelines. The speech samples were sprinkled with statements like “we do everything together” demonstrating that the pair spent a lot of time together that was satisfying for both mother and child. Activities that the pairs enjoyed included shopping, exercising, reading, playing games, and preparing meals.

**Dissatisfactions and criticisms.**

The presence of dissatisfactions and the frequency of critical statements add to the overall coding of the relationship and final EE ratings. Even though the relationships between mothers and their children with FXS were positive for the most part, some mothers expressed “dissatisfaction” with their child and their child’s behavior. Other mothers while expressing dissatisfaction with the situation (i.e. diagnosis of FXS), did not attribute the dissatisfaction to their children. This form of dissatisfaction does not add to the coding of the relationship or final EE coding because the respondent does not directly “blame” the child for the situation. Therefore, these types of statements, such as “sometimes it is hard or frustrating” were not coded as “dissatisfactions.”

Similar to the lack of dissatisfaction expressed by mothers, there were also limited “criticisms.” In the sample, only two criticisms were coded. This may be because FXS is an inherited genetic disorder and mothers are more forgiving for the behavior of their
children because of their limited intellectual ability. Another explanation could be that despite their challenges, the mothers are generally positive because they are accustomed to the children’s behaviors. Parents of children with schizophrenia and other mental health disorders tend to be more critical of their children’s behavior.

One argument supporting the discrepancy of EE ratings, especially dissatisfactions and criticisms, between children with intellectual disabilities (i.e. FXS) and children with mental health disabilities (i.e. schizophrenia), is the dichotomy of “visible” versus “invisible” disabilities. Children with FXS have a “visible” disability, even though they are born with few dysmorphic characteristics and are often not diagnosed until three or four years of age. The visibility of FXS emerges more as they age and the developmental gap widens. Criticisms may not be attributed to children with FXS because their behavior is perceived to be no fault of their own, due to their diagnoses. Additionally, disabilities that are more noticeable may elicit patience and sympathy. Children with mental health disorders, on the other hand, have an “invisible” disability and are often criticized for their behavior because they appear “normal” at times, even though their challenges are not physically obvious. Children with learning disabilities, attention/behavioral disorders, or sensory processing disorders encompass invisible disabilities and the public may have higher and/or unrealistic expectations for their functioning, in relation to their actual capabilities (Mitchell, 2002). Parents and family members, while knowledgeable of their children’s diagnosis, still have higher expectations and sometimes “blame” the children for their intermittent and erratic behavior (Todd & Shearn, 1997). Society, in general is less sympathetic and
compassionate to individuals with invisible disabilities because of a common misconception that their behavior is a result of no self-control, therefore contributing to criticism, dysfunction, and discrimination (Mitchell, 2002). Historically, children and adults with visible disabilities or disfigurements were discriminated against and denied civil liberties which resulted in low expectations and adjustment to adult life (Goldberg, 1974). In recent years, society has become more compassionate and accepting of individuals with visible disabilities through systematic inclusion and civil rights legislation (Schalock et al., 2002; Thorn, Pittman, Myers, & Slaughter, 2009).

The age at diagnosis for children with “visible” and “invisible” disabilities is also a factor. Children with FXS are often not diagnosed until 3-4 years of age, which is late according to early intervention standards (Bailey Jr, Roberts et al., 2001; Carmichael, Pembrey, Turner, & Barnicoat, 1999). However, children with mental health disorders, are often diagnosed at a later age yielding less adjustment time for the parents (NIMH, 2008). Children with FXS are typically diagnosed when they are children and stay “child-like” due to their intellectual disability. Alternatively, children with mental health disorders are diagnosed at a later age, often into adolescence or adulthood. Parents of these children have witnessed their children develop typically in many ways, such as physically and cognitively and may express more dissatisfaction and criticisms when they behave poorly. (G. W. Brown, J. L. T. Birley, & J. K. Wing, 1972; McCarty & Weisz, 2002b). Parents of children with invisible disabilities may know intellectually that their child has a disorder that results in challenging or aberrant behaviors, but emotionally it is harder to reconcile. Therefore, they may less sensitive to their children’s behavioral
outbursts. Alternatively, parents of children with visible disabilities, particularly inherited genetic disorders, may be more sensitive and accommodating to their children’s difficult behaviors. This dichotomy provides more rationale for the argument that mothers of children with FXS may be reluctant to express strong criticisms and/or dissatisfactions.

**Emotional over-involvement (EOI).**

Emotional over-involvement is most salient to the attitudes and characteristics of mothers of children with FXS. Research on mothers who are carriers of the *FMR1* gene mutation, suggest that they have a tendency to experience more anxiety, depression, low self-esteem, social isolation, and higher levels of stress than mothers of children with other disabilities (Grigsby et al., 2008; C. J. Johnston et al., 2003). Previous descriptions of female carriers are consistent with mothers in this sample who were coded Borderline and High EOI due to increased statements of concern, worry, and anxiety about their children. Excessive worry is one of the indications that lead to the Self-Sacrificing Over Protective (SSOP) rating. Statements such as “I worry about my son constantly” or “It’s hard to go on vacation or out of town for a short time, because I worry about how she is doing” provide evidence of an SSOP rating. Every parent worries about his/her child to some degree, but it is the “excessiveness” that is the relevant feature that can be detrimental to the emotional climate in the home environment. Mothers of children with FXS may have good reason to worry about their children due to frequent medical and behavioral problems, although it is difficult to discern whether this attribute is a result of the carrier phenotype or common concerns that come with having children with
disabilities. McConkie-Rosell and colleagues (2005) reported that mothers with the FMR1 premutation may be predisposed to psychological issues such as anxiety and depression and may be exasperated by parental guilt and blame.

The subcategories in the EOI category entail specific maternal behaviors that are potentially damaging to the emotional family climate. The subcategories are emotional display, excessive detail, Self-Sacrificing Over Protective Behavior (SSOP), statements of attitude, and positive remarks. Most notably, more mothers in this sample tended to be rated EOI, rather than Critical due to the high expression of positive remarks. Positive Remarks are an expression by the mother that the child does something “very well” or is “very good” at something. According to the FMSS guidelines, a high frequency of positive remarks (>5) within a short speech sample results in a borderline EOI rating. That coupled with other components of EOI, such as statements of attitude, leads to a score of High EE. Many mothers in this sample often exclaimed that their child was very good at engaging in social acts with others and that their child was popular either at school or in the community. Statements like “everyone knows him” or “he has a friend everywhere he goes” often followed or preceded the positive remarks. These statements are supported by the FXS literature that report children with FXS are inherently social beings although sensory over-stimulation often times gets in the way (Bailey Jr et al., 1998; K. Cornish et al., 2008; Epstein et al., 2002a). These Positive Remarks may be seen an example of how mothers of children with FXS continually highlight the best attributes of their child.
As reported in the results chapter, emotional display and excessive detail were for the most part, absent. Statements of Attitude, Self-Sacrificing Over Protective Behavior (SSOP), and Positive Remarks were more prevalent, indicating mothers’ Emotional Overinvolvement in the lives of their children. Statements of Attitude are a mother’s expression of intense love for her child, or “willingness to do anything for the relative in the future” (Magana-Amato, 2002, p. 42). The unconditional devotion and willingness to do anything for their children could be detrimental to the children and the parent-child relationship. The rating of SSOP was small, but also important to recognize, because more mothers were coded as borderline SSOP rather than full SSOP. This rating indicated that mothers would sacrifice much of themselves to help their child. In many of the speech samples there were “hints” of SSOP but not enough for a borderline or full rating, but statements that the mother-child dyad were inextricably linked but that the mother did not feel she was sacrificing. Such statements included “we are joined at the hip,” or “he/she is closer to me than anyone else in the family.”

**Implications of Expressed Emotion**

The results of the final ratings of EE suggest an emerging trend of Emotional Overinvolvement for mothers of children with FXS. This trend has implications not only for the family but also for practitioners working with the families. The excessive worry, anxiety, and Emotional Overinvolvement can lead to undesirable behaviors from the child and misperceptions of the child’s ability level by the practitioner. The research on expressed emotion revealed that mothers who expressed critical or emotionally over-involved views of their children are more likely to have harsher parenting practices and to
be more over-protective (Calam & Peters, 2006; Vostanis & Nicholls, 1992). In addition, research indicates that when children live in a home environment with a parent who has high expressed emotion (either Critical or Emotional Overinvolvement), their clinical treatment is more likely to be compromised, as evidenced by increased rates of schizophrenic relapse and subsequent hospitalization (G. Brown et al., 1972; Jacobsen et al., 2000; Vaughn & Leff, 1976). Following this expressed emotion research, mothers of children with FXS who are emotionally over-involved (and High EE) could be at risk of mediating the full effects of the children’s therapeutic intervention, although this claim cannot be verified without further study. The implications for these findings on intervention are unknown and speculative at this time, however based on previous research possible links could have powerful consequences. For example, children with FXS spend a limited number of hours in therapy, relative to the time they spend with their family. The skills learned in therapy may be negated because of parents’ critical or over-involved behaviors and the children may regress back to homeostasis because of the unintended stress by the parents. Brain research has provided evidence that when individuals experience stress, they use the more primitive parts of the brain and do not retain newly learned skills (Conger, Conger, Elder, Lorenz, & Simons, 1994; Howes et al., 2008; J. Shonkoff et al., 2007; J. P. Shonkoff & Phillips, 2000). Parents are often not involved in the therapeutic sessions and may not have a full understanding of the environmental conditions needed to elicit optimal behavior of their children and therefore, unintentionally place stress on the child. By involving parents in the therapeutic sessions and/or discussing the therapeutic interventions with parents, they
may understand how to reinforce intervention strategies at home in an appropriate and balanced way (i.e. not over critical or involved). This is very important for mothers in this study because they tended to be rated High EE with Emotional Overinvolvement and including them in the therapy session may only exasperate their overinvolvement, therefore, it is important for practitioners to emphasis the need for mothers to be mothers first and not therapists.

Practitioners working with families with children with FXS need to be aware of the potential “emotional over-involvement” of mothers and its effects, particularly possible mediating effects on therapeutic interventions. When practitioners are aware of the influence of expressed emotion, particularly EOI, then they can adjust their expectations for therapeutic interventions by working closely with the parents about following through with intervention strategies and helping the parents set realistic expectations. Practitioners can also help mothers understand and balance their expressed emotion so that the therapeutic interventions can result in the best possible outcomes.

Expressed emotion and perceptions of behavior.

The primary research questions of this dissertation were centered on the relationship between the expressed emotion of mothers of children with FXS and the perceptions of their children’s behavior. The results of this study found no significant correlations between the overall scales of the FMSS and the BASC and Sensory Profile, and few significant associations between with the subscales. There may be several explanations for these findings, such as the mothers’ reluctance to express the emotional and behavioral views of their children in full. This was suggested previously when
discussing the “low threshold” populations for the FMSS resulting in borderline EE ratings. The same argument could be applied to the BASC and Sensory Profile measures. Mothers may be more reluctant to express their emotions through an interview, as well as through pen and paper because it is difficult for parents to be objective about their children and express their perceptions in a “public” way. This study was asking mothers to do a very difficult task of talking about their children’s most difficult characteristics in a unique way. To that end, these measures were not objective, but the mothers’ perceptions of their children’s behavior and sensory processing. Additionally, the instrument is by design, intended to measure perception.

The correlations within the subscales reveal some interesting findings. There was a significant correlation between demographic variables and the FMSS. The results suggest an association between both ethnicity and marital status and EE. In this sample, mothers that self-identified themselves as Caucasian and married were less critical and emotionally over-involved than mothers who were single and ethnically diverse. This may be a reflection of societal stressors on single parents as well as individuals of color. It is worth noting again, that this sample was not diverse in terms of ethnicity, marital status, or income so the distribution of data is skewed. The question of cultural diversity and expressed emotion is worth exploring in future studies to determine whether mothers of color are more likely to have high expressed emotion. A replication study of FMSS with Mexican-American, Spanish-speaking relatives found no differences between the samples of Mexican-Americans and Caucasians (Magana et al., 1986).
Further analysis revealed associations between two of the subcategories of the FMSS and one subcategory of the BASC. First, there was a significant, positive relationship between the FMSS subcategory, Self-Sacrificing Over Protective Behavior (SSOP) and the BASC’s subcategory, Atypicality ($r=.517$, $p<.05$, $n=30$). As mentioned before, SSOP refers to statements a mother makes during the FMSS that would indicate that she sacrificed herself for the benefit of her child in an extreme way. The Atypicality scale on the BASC measures “a child’s tendency to behave in ways that are considered odd or strange” (p. 61). Such behaviors may include chewing on clothes or objects, talking or singing to oneself, or exhibiting socially inappropriate behaviors. These behaviors are typical for children with FXS, and even more so for children who are dually diagnosed for FXS and Autism (Hagerman, 2006d). It stands to reason that mothers of children with FXS would be more over protective when their child exhibits atypical behaviors. It is a natural maternal response to want to protect your child from the harsh realities of the public eye.

Second, a significant, negative relationship between the FMSS subcategory, Positive Remarks, and the BASC’s subcategory of Atypicality was identified. A negative relationship reveals that the more positive remarks mothers made about their children, the less atypical behaviors were perceived and reported. Alternatively, the more atypical behaviors perceived, the fewer positive statements expressed by mothers. This relationship makes sense because when mothers (or other family members) view their children exhibiting atypical behaviors that are embarrassing or require explanation to others, they may not view them in a positive light. Atypical behaviors do not necessarily
engender warm, positive responses. Parents typically try to reduce the number of atypical behaviors because they make children stand out and often elicit criticisms or ridicule from others.

**Expressed emotion and sensory processing.**

The second research question involved the constructs of expressed emotion and sensory processing, measured by the Five Minute Speech Sample (FMSS) (Magana-Amato, 2001) and the Sensory Profile (SP) (Dunn, 1999). Results indicated no significant relationship between the overall FMSS EE ratings and summary sections of the SP, although there are many significant relationships between FMSS subcategories and SP subsections. It is not surprising that mothers in this sample would identify their children with sensory processing problems because the literature has documented an array of sensory processing disorders present in children with FXS (R. J. Hagerman, 2002; Hagerman et al., 2008; Scharfenaker et al., 2002a). There were positive and negative relationships between EOI subcategories and SP subsections suggesting that mothers in this sample were more often EOI and perceived their children to have variable sensory processing difficulties, adding to the complexity of the mother-child relationship. One of the positive relationships was between Positive Remarks and the Sedentary subsection of the SP, indicating that as Positive Remarks increase so does the mothers’ perception of their children’s sedentary or inactive responses. The relationship makes sense because children that are more sedentary and do not display out-of-control behaviors are more likely to receive positive feedback from caregivers because they are not causing disruption or troubling behaviors. Alternatively, the negative relationships
between EOI ratings and other SP subsections present a confusing pattern. For example, there were negative relationships between Statements of Attitude (i.e. statements of extreme devotion) and Vestibular, Modulation and Low Endurance indicating that more Statements of Attitude correspond with decreased identification of problems with vestibular, modulation, and low endurance. In other words, the more mothers talked about how much they loved their child and would do anything for their children, the less sensory problems they identified. This relationship may mean that mothers could project unrealistic improvements of their children’s behaviors. Alternatively, it could be an accurate reflection of the children’s behavior, although without corroborating evidence, this remains an unknown.

The subcategory of Dissatisfaction on the FMSS was negatively correlated with the sensitivity classification on the Sensory Profile, meaning that as Dissatisfaction decreased, the classification of sensitivity increased and mothers did not express criticisms or dissatisfaction of this sensory challenge of their children. This significant relationship indicates that mothers who are more aware and understanding of their children’s sensitivities, are less dissatisfied and with their children or situation. This finding has positive implications for families with children with FXS because professionals or providers can inform parents of expected sensory challenges and parents become more aware and less likely to blame the children for erratic behaviors, but they may also exhibit high expressed emotion and inhibit optimal benefit.

Overall, children with FXS are both over and under stimulated by a variety of environmental sensory stimuli; therefore, it is difficult to discern the possibilities related
to these associations. Parenting a child with FXS is extremely difficult because he/she often has unpredictable behaviors that may stem from sensory processing problems. Consequently, this author supports the position that mothers of children with FXS are more likely to be EOI because children with FXS are extremely difficult to parent due to their unpredictable and challenging behaviors. This question should be explored further to address the interaction between factors with a larger sample to account for confounding variables. It is not the intention to begin to make a causal argument with this data, but acknowledge that the data provides interesting information within the context of the FXS population.

Statements about sensory processing that did not necessarily fit the coding of the FMSS supported these correlations. This may be a result of the varying ages of children and the fluctuating pattern of sensory processing as children age. Many of the mothers reported that they knew something serious was wrong with their child very early on due to their reactions to experiences that resulted in difficult behavior. Mothers reported overstimulating behavior during transitions, mealtimes, and in community settings. They went on to talk about the strategies they employed to decrease sensory processing difficulties that resulted in undesirable behaviors. Interestingly, several mothers reported using Hippotherapy, a growing therapeutic technique involving the use of horses with children with developmental delays, as well as music to calm their children during overstimulating experiences. Mealtimes were also reported very challenging times for the mothers due to either the children’s avoidance or commitment to particular foods, as well as over eating behavior. These statements, while not coded in the FMSS
demonstrate that the mothers were committed to finding strategies that work well for their children.

**Limitations**

It is important to acknowledge the limitations of this study. The primary limitations of this study involved the limited sample and the selected measures. This was a small sample with a subset of mothers who were inclined to participate in FXS research. Even though the sample came from across the United States, the make-up of the demographic characteristics portrayed a homogeneous population of white, middle-class, stay-at-home mothers. Cultural and socioeconomic differences may affect the results and the utility of the measure and should be examined in future studies. The age range of children was also a limitation and prevented conclusions related to specific ages such as early childhood, school age, or adolescence. In future studies, it would be beneficial to study distinct age ranges independently using a larger sample to see if the expressed emotion of mothers differs according to the age of their children.

The sample size also limited the analysis of the data. Correlational analysis was utilized effectively however a larger sample would have expanded the analysis options including regression analysis that could have added a predictive component to the study. Moreover, the data could have been analyzed in quartiles to address the variability of the age range and the different subcategories of the FMSS.

Additionally, random measurement errors may exist especially in the self-reporting measures that the mothers completed (i.e. BASC and Sensory Profile), although the instruments’ validity checks try to account to such errors. There may have also been
measurement errors in the coding of the FMSS and next steps should include inter-rater reliability analysis.

The FMSS provided rich information about the population of mothers of children with FXS, which measures a construct that has not been explored before with this population, however the FMSS instrument, can be considered a limitation. Traditionally, the FMSS was developed and used in the fields of psychology and psychoanalysis, as opposed to educational or therapeutic fields such as special education, speech pathology, or occupational therapy. The translation into these fields may or may not be appropriate. Its primary use was to examine the family climate of patients with mental illness. Therefore, the instrument may be viewed as a tool that pathologizes parents. It may appear to judge parents even though their statements may be an appropriate reaction or situationally specific. For instance, when parents talked about their worries for their children, this was an appropriate worry because their children have significant medical and developmental challenges. The labels used in the FMSS (i.e. Critical, Emotional Overinvolvement, Self-Sacrificing Over Protective behavior) hold negative connotations and may be alarming to parents and practitioners. These labels may also lead to inaccurate generalizations of mothers of children with FXS. The effectiveness and usefulness of such as tool has not been explored to a sufficient degree. This study produced a profile of mothers of children with FXS, however at this time the meaning behind this profile remains unclear. There is no documentation to support or refute the consistency of issues within the labeled categories across populations. SSOP may or may not have the same meaning for families of children with special needs and those of adults.
with mental illness. The grieving process has been well documented, however we do not know about Emotional Overinvolvement for these families, nor do we know if that construct is actually adaptive and necessary given their situation. For these reasons, the FMSS may not be a tool that is ready to be used for intervention planning for children with FXS or other disabilities.

Limited correlations were found between the behavioral and sensory processing instruments and the FMSS. This finding may indicate that the BASC and the Sensory Profile were not the optimal measures for this study. Both tools are used for intervention planning and therapy, whereas the FMSS is not and therefore they may have not been a good match for analysis. Even though the BASC and Sensory Profile were chosen because children with FXS have significant issues with behavior and sensory integration, many of the mothers commented that certain items did not apply to their child. In future studies, the measures of behavior and sensory processing need to be more sensitive to the FXS population as well as the age range to address the applicability for the population.

**Future directions.**

The next step for this line of research includes recruiting a larger sample to look solely at the expressed emotion of mothers with children with FXS to understand this construct within this population, specifically the phenotype of mothers with the *FMR1* premutation. A comparison of expressed emotion between mothers of children with FXS and mothers of children with other developmental disabilities, such as Down syndrome, Autism, or Williams syndrome would be crucial to sort out the differences between the FXS phenotype and environmental effects within the relationship between
EE and FXS, or other children with systemic delays. Further research could also explore how much of the variance of EE ratings are explained by the mothers’ own challenges of being a carrier of the \textit{FMR1} premutation.

Additionally, exploring the expressed emotion of fathers of children with FXS and siblings would have great promise to view the whole families expressed emotion and possible implications such as interaction patterns. Calam et al. (2006) raised this question by stating, “An important assumption is that the way in which parents talk about their child reflects important aspects of the way that they interact with their child in everyday life. Studies should establish the extent to which the various dimensions of EE relate to actual parenting behaviour” (p. 114). It would also be important to look at the differences in parenting behavior and the dimensions of EE.

In the future, the developers of the FMSS or other researchers may want to review the coding labels and change them to reflect language that is less pathologizing and reactionary. A critique of the whole measure would be an area of future research to address the aforementioned limitations. Another limitation of the FMSS is the limited range of scoring, which is either dichotomous (High, Low) or a range of three points (High, Low, Borderline; Positive, Negative, Neutral) for the overall rating and many of the subcategories. Future psychometric studies could address an increased range within the FMSS to address variability within the scores.

This study began to explore the expressed emotion of mothers of children with FXS in relation to their behavior and sensory processing. The initial findings provide striking information on the high number of mothers with FXS who met the criteria for
High EE, particularly Emotional Over-involvement. These findings may lead to intervention strategies that can help mothers balance their expressed emotion and improve the impact for their children’s therapeutic interventions, in light of previous research on the well-being of mothers of children with FXS. Even though findings thus far have revealed few relations between the FMSS and the instruments measuring the mothers’ perception of their child’s behavior, with a larger sample size, these associations may become more apparent through statistical correlations.

Research has provided evidence of the difficulties of parenting a child with a disability such as FXS due to the lack of awareness, carrier phenotype, and the grieving process. Therefore, it is important to explore the parent-child relationship and begin to understand how the diagnosis of FXS can affect the relationship. Practitioners, providers, and all types of professionals working within the FXS field need work with parents to provide meaningful support so that their child can receive optimal benefits from treatment. It is primarily the practitioners’ job to be objective about a child’s abilities and challenges, as opposed to the parents’ responsibility. It is difficult for any parent to view their child through a critical lens and provide the important intervention needed for success. Although, parents are often told that, they need not be a “therapist,” they must follow through with therapeutic interventions at home, if they want the desired outcomes. These mixed messages provide a platform for mothers who are already at risk for anxiety and social isolation, to be even more cautious, worried, and anxious about how they talk about their children with FXS. Consequently, careful interpretation of the expressed emotion of mothers of children with FXS is required. These findings lead to more
questions, such as; how does this perception affect their child’s behavior and what level of intervention is needed to change undesirable behavioral characteristics? How do the mothers’ perceptions color their expressed emotion of their children? Practitioners need to be aware of how parents perceive their children and talk about them, as they are with their children for a majority of time. Practitioners, on the hand, are only with the child for as few as 2-4 intervention hours a week and have to implement therapy based on their observations and parents’ reports. Both of these sources of information may not coincide and may lead to ineffective or inappropriate intervention. The expressed emotion of mothers with children with FXS is an intriguing topic that warrants further research and discussion among physicians, therapists, researchers, and families and this dissertation provides a beginning to that discussion.
References


Lachiewicz, A. M., Spiridigliozzi, G., Dawson, D., Cuccaro, M., & McConkie-Rosell, a. *A study of cognitive, emotional and physical characteristics in women with the fragile X premutation.*


Appendix A

Demographic Survey

Study Title: The relationship between the Expressed Emotion of mothers of children with fragile X syndrome and the behavioral and sensory characteristics of their children

Directions: Please fill in or circle the answer that best describes you or your family.

Mother’s Information

1. Mother’s Name: ____________________________________________________

2. Mother’s age:
   - 20-30
   - 31-40
   - 41-50
   - 51-60
   - Over 60

3. What is your gross annual income?
   - Less than $10,000
   - $10,000-$20,000
   - $20,000-$40,000
   - $40,000-$60,000
   - $60,000-$100,000
   - Over $100,000

4. What is your marital status?
   - Single, never married
   - Separated
   - Divorced
   - Married
   - Living together as if married
5. What is your highest level of education?
   - Grades 0-8
   - Grades 9-11
   - High School or GED
   - Some College
   - College Graduate
   - Post-College Degree

6. Are you working right now?
   - Yes, full time
   - Yes, part time
   - Stay-at-home parent
   - Not working, but looking for a job

7. What is your ethnicity?
   - Mexican, Mexican-American
   - Other Latino or Hispanic
   - African American
   - American Indian
   - Asian
   - Pacific Islander
   - Caucasian
   - Other

Child’s Information

8. Child’s Name: _____________________________________________________

9. Child’s age:
   - __________________

10. Child’s gender:
    - Male
    - Female
11. What is the ethnicity of your child?
   - Mexican, Mexican-American
   - Other Latino or Hispanic
   - African American
   - American Indian
   - Asian
   - Pacific Islander
   - Caucasian
   - Other

12. Is your child diagnosed with the full mutation of fragile X syndrome (FXS)?
   - Yes
   - No

13. What was your child’s age at the time of the FXS diagnosis?
   - ______________

14. Does your child have any of the following diagnoses in addition to fragile X syndrome?
   - Sensory Integration Disorder
   - Autism Spectrum Disorder
   - Attention Deficit Disorder
   - Vision or Hearing disability

15. How many other children live at the home with you? ________________

16. Are any other children diagnosed with FXS?
   - Yes
   - No

17. Is your family going through any of the following transitions?
   - Individual Education Plan (IEP) initial placement
   - Individual Education Plan (IEP) review
   - Medical or educational diagnoses of your child(ren)
• Birth of another child
• Family relocation
• Parental change/loss of employment

18. Is your family connected with any regional or national FXS support groups?
• Yes
• No