Autism Spectrum Disorder in Multiplex Families: A Qualitative Study of Diagnostic Experiences and Parental Perceptions

Master’s Thesis

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by
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ABSTRACT

Autism Spectrum Disorder in Multiplex Families: A Qualitative Study of Diagnostic Experiences and Parental Perceptions

A Thesis presented to the Department of Biological Sciences, Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

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Autism spectrum disorders (ASDs) are among the most commonly diagnosed neurodevelopmental disorders with an incidence of approximately 1 in 110 individuals, and are characterized by impairments in communication, reciprocal social interaction, and stereotyped patterns of behavior. For parents, receiving an ASD diagnosis in a child can be a distressing experience that can significantly affect coping and adaptation. Recent studies have demonstrated that the recurrence risk for siblings may be as high as 25%, suggesting that multiplex families may be much more common than originally believed. To date there is limited research focused on multiplex families, and in particular there has been no exploration of the emotional impact of receiving an ASD diagnosis in more than one child. The aim of this study was to investigate the diagnostic and lifestyle experiences of multiplex ASD families through qualitative interviews with seven parents.

In concordance with previous literature, parents in this study reported feelings of confusion leading up to and during receipt of an ASD diagnosis and encounter cyclic re-emergence of worry and grief following a diagnosis. The parents we interviewed told us
that although they were familiar with ASDs, they still did not feel prepared when faced with the possibility of a second ASD diagnosis. Additionally, they did not feel that medical professionals took their concerns about their second or third child seriously, despite the fact that one or more children in the family had already been diagnosed. Finally, parents of multiplex families reported that having more than one child with an ASD creates additive emotional and lifestyle challenges and believe that they experience increased feelings of isolation and stress due to a "laser-beam" focus on autism. Results from this study help to reinforce areas in which clinical professionals can provide additional support to parents of multiplex families surrounding an ASD diagnosis.
# TABLE OF CONTENTS

**INTRODUCTION** ......................................................................................................................... 1  
  Overview of Autism Spectrum Disorders ................................................................. 1  
  The Impact of Autism Spectrum Disorders on Parents ........................................ 2  
  ASD Etiology, Risk Perception and Family Planning ........................................... 3  
  Study Purpose ....................................................................................................................... 4  

**METHODS** ................................................................................................................................. 6  
  Sampling Methods and Study Population ............................................................. 6  
  Study Design ..................................................................................................................... 8  
  Data Management and Analysis ............................................................................ 8  

**RESULTS** ...................................................................................................................................... 9  
  Overview of Major Themes ........................................................................................... 9  
  Description of Themes and Supporting Quotes .................................................... 10  

**DISCUSSION** ............................................................................................................................... 14  
  Discussion of Major Themes ...................................................................................... 14  
  Limitations and Future Research ............................................................................ 18  

**CONCLUSIONS** ........................................................................................................................... 20  

**REFERENCES** .............................................................................................................................. 22  

**APPENDICES** ............................................................................................................................. 25  
  Appendix A: Recruitment Notice .............................................................................. 25  
  Appendix B: Eligibility Screening Tool ................................................................. 26  
  Appendix C: Informed Consent Form ..................................................................... 27  
  Appendix D: Interview Guide ................................................................................... 31
LIST OF TABLES

Table 1. Participant Demographics ................................................................. 7
Table 2. Demographics of Participants' Children ............................................. 7
INTRODUCTION

*Autism Spectrum Disorders – Overview*

Autism spectrum disorders (ASDs) include Autistic Disorder, Asperger Disorder and Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS). These disorders are characterized by impairments in communication and reciprocal social interaction, as well as stereotyped patterns of behavior and interests. According to the current DSM classification, these three disorders are part of a broader category referred to as Pervasive Developmental Disorders, which also include Childhood Disintegrative Disorder (CDD) and Rett syndrome (*Diagnostic and statistical manual of mental disorders (Revised 4th ed.),* 2000). However, it is anticipated that in the near future DSM classification will change, and individuals with an ASD will receive a diagnosis of Autism Spectrum Disorder rather than Autistic Disorder, Asperger Disorder or PDD-NOS (Wing, Gould, & Gillberg, 2011).

Once considered rare, ASDs are now among the most commonly diagnosed neurodevelopmental disorders with a recent incidence of approximately 1 in 110 people. There has been much speculation surrounding the origins of this dramatic increase in prevalence, with the greatest consensus attributing the rise in ASDs to broadened diagnostic criteria, increased awareness, and improved identification (Dosreis & Weiner, 2006; King & Bearman, 2009; Matson & Kozlowski, 2011). ASDs are diagnosed by clinical professionals such as psychologists or physicians trained to assess a child's
communication, behavior, and developmental levels in accordance with the DSM-IV criteria (Lord C & Corsello, 2005).

The precise etiological factors that predispose a child to ASDs are not fully understood, however, current research has substantiated considerable evidence for genetic, syndromic and environmental contributions (Folstein & Rosen-Sheidley, 2001; Freitag, Staal, Klauck, Duketis, & Waltes, 2010; Landrigan, 2010; Schaefer & Mendelsohn, 2008). Approximately 10-15% of individuals with an ASD have an identified Mendelian condition or genetic syndrome with distinct clinical features such as fragile X syndrome, tuberous sclerosis and chromosome abnormalities (Folstein & Rosen-Sheidley, 2001). Nevertheless, the most significant etiology for ASDs to date remains idiopathic, or non-syndromic, with the cause(s) for the largest percentage of cases remaining largely unknown. Current recommendations for genetic testing and diagnosis use a tiered approach with chromosomal microarray and fragile X testing included in the first tier (Folstein & Rosen-Sheidley, 2001; Schaefer & Mendelsohn, 2008; Shen et al., 2010).

**The Impact of Autism Spectrum Disorders on Parents**

For families of children diagnosed with an ASD, getting a diagnosis can be a distressing experience that can significantly affect coping, adaptation, and future family planning (Keenan, Dillenburger, Doherty, Byrne, & Gallagher, 2010; Siklos & Kerns, 2007). Despite the substantial impact of this diagnostic experience, recent literature has shown that many parents of children with an ASD remain dissatisfied with the diagnostic process (Brogan & Knussen, 2003; Howlin & Moore, 1997; Keenan, et al., 2010; Mansell & Morris, 2004). These survey-based studies have highlighted the critical elements of the
diagnostic experience including timing of diagnosis, quality of information, manner of the health professional during disclosure, as well as acceptance of parental suspicions prior to diagnosis. Further research suggests that parents regularly encounter stressful delays before finally receiving an appropriate diagnosis, and often experience feelings of confusion, self-blame or guilt, and frustration (Howlin & Moore, 1997; Keenan, et al., 2010; Midence & O'Neill, 1999).

When receiving the diagnosis, parents of children with an ASD have been found to experience a variety of emotions including shock, confusion, worry, grief, fear, isolation, anger, sadness and feeling overwhelmed (Cameron, 1992; Siegel, 1997). A primary theme described by these parents is a profound sense of sadness and loss similar to experiencing the death of a loved one. However, in contrast to the stages of grief and acceptance described by Kubler-Ross (1969), research examining the emotional experiences of mothers caring for a child with a developmental disability show that there is no clearly defined final stage of acceptance in these parents. Rather, emotions emerge and re-emerge with the developmental milestones such as birthdays, adolescence and schooling, which serve as reminders of the loss (Cameron, 1992).

**ASD Etiology, Risk Perception and Family Planning**

A small, but significant body of research has focused on parents’ beliefs about the causes of ASD in their children. Although parents have identified a wide range of causes, including childhood illness, vaccination, pregnancy complication, diet, and environmental exposures, the most consistently cited cause is genetic factors (Elder, 2001; Gray, 1995; Mercer, Creighton, Holden, & Lewis, 2006; Selkirk, Veach, Lian, Schimmenti, & LeRoy, 2009). Inextricably tied to this perceived genetic contribution in
ASDs are parental perceptions of recurrence risk. Mercer et al. surveyed 37 parents regarding perceived recurrence risk for ASD, and while one third of the participants believed the recurrence risk to be 25%, the majority of participants believed the recurrence risk to be as high, or higher than 75% (2006). Until recently, actual recurrence risks to siblings were estimated to be 5-8%, with a higher risk of recurrence for a child with two affected siblings (Folstein & Rosen-Sheidley, 2001; Mercer, et al., 2006). In 2011, Ozonoff demonstrated that the sibling recurrence risk may be as high as 25% for a child with only one affected sibling, citing the two strongest predictors of an ASD diagnosis as gender of the infant sibling and number of affected older siblings. Prior to these findings, little emphasis was placed on multiplex ASD families as sibling recurrence risks were believed to be relatively low. However, with this new research clearly demonstrating significantly higher recurrence risks, health care providers must be attuned to the unique experiences and challenges facing multiplex ASD families.

*Study Purpose*

While there is a wealth of emerging research and information on autism spectrum disorders, there is a noticeable absence of qualitative studies that capture the experiences of parents who have more than one child with an ASD. Previous research has demonstrated that multiplex families may be much more common than originally believed, therefore, the purpose of this study was to:

- Describe parents’ experiences receiving a diagnosis of an autism spectrum disorder in more than one child.
- Explore the significance of this experience as it relates to family planning and beliefs about the disorder.
• Describe the potentially unique lifestyle challenges in multiplex ASD families. Highlighting the experiences of parents who have more than one child on the autism spectrum will help providers involved in the care of these children gain insight into the emotional, social and diagnostic issues experienced by multiplex ASD families.
METHODS

The study design was approved by the Brandeis University Institutional Review Board.

**Sampling Methods and Study Population**

We recruited participants for this study through local and national autism support groups including Community Autism Resources of Massachusetts, Northeaster Arc – Autism Support Center, *Autism Speaks*, Family Autism Center at South Norfolk County Arc, the Autism Resource Center of Central MA, and the Boston Autism Consortium. A recruitment notice (Appendix A) was provided to the points of contact in each organization, and was subsequently posted to their organizations website and/or included in their weekly email update to organization members. Interested individuals were encouraged to contact us directly via email, and we assessed eligibility using a brief questionnaire (Appendix B). Subject inclusion criteria were as follows: a parent or caregiver of child with an ASD, 18 years of age or older, fluent in English, with 2 or more children of any age that have been diagnosed with an ASD by a medical professional and have been diagnosed at different points in time.

Eleven respondents expressed interest, and seven were eligible for study participation. A copy of the informed consent form (Appendix D) was e-mailed to each participant and collected prior to completing a telephone interview. Of the seven participants we recruited for this study, five had two children with an ASD, and two had three children with an ASD. In total, there were fifteen children with an ASD and ten of these fifteen children were male. Diagnoses included autistic disorder, Asperger disorder,
and PDD-NOS. Demographics of participants as well as participants’ children are included in Table 1 and Table 2, respectively.

Table 1. Participant Demographics

<table>
<thead>
<tr>
<th>ID</th>
<th>Age</th>
<th>Gender</th>
<th>State of Residence</th>
<th>Occupation</th>
<th>Ethnicity</th>
<th>Education</th>
<th>Marital Status</th>
</tr>
</thead>
<tbody>
<tr>
<td>P1</td>
<td>53</td>
<td>F</td>
<td>MA</td>
<td>Librarian</td>
<td>Caucasian</td>
<td>Some graduate work</td>
<td>Married</td>
</tr>
<tr>
<td>P2</td>
<td>44</td>
<td>F</td>
<td>MA</td>
<td>Homemaker</td>
<td>Caucasian</td>
<td>Bachelor's degree</td>
<td>Married</td>
</tr>
<tr>
<td>P3</td>
<td>29</td>
<td>F</td>
<td>AL</td>
<td>Consultant</td>
<td>Caucasian</td>
<td>Bachelor's degree</td>
<td>Married</td>
</tr>
<tr>
<td>P4</td>
<td>49</td>
<td>F</td>
<td>MA</td>
<td>Site Manager</td>
<td>Caucasian</td>
<td>Bachelor's degree</td>
<td>Married</td>
</tr>
<tr>
<td>P5</td>
<td>N/A</td>
<td>F</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>P6</td>
<td>41</td>
<td>F</td>
<td>MA</td>
<td>Business Coordinator</td>
<td>Caucasian</td>
<td>Bachelor's degree</td>
<td>Married</td>
</tr>
<tr>
<td>P7</td>
<td>46</td>
<td>F</td>
<td>MA</td>
<td>Homemaker</td>
<td>Caucasian</td>
<td>Masters</td>
<td>Married</td>
</tr>
</tbody>
</table>

*P1-P7 = Participants 1-7; P5 opted not to respond to demographic questions.

Table 2. Demographics of Participants' Children

<table>
<thead>
<tr>
<th>ID</th>
<th>Total number of Children</th>
<th>Number of Children with an ASD</th>
<th>Diagnosis in Child</th>
<th>Gender</th>
<th>Current Age</th>
<th>Age at Diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>P1</td>
<td>4</td>
<td>2</td>
<td>Asperger Disorder</td>
<td>M</td>
<td>26</td>
<td>13yr</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Asperger Disorder</td>
<td>M</td>
<td>14</td>
<td>10yr</td>
</tr>
<tr>
<td>P2</td>
<td>3</td>
<td>3</td>
<td>Autism</td>
<td>F</td>
<td>13</td>
<td>3yr</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Autism</td>
<td>F</td>
<td>11</td>
<td>22mo</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>PDD-NOS</td>
<td>F</td>
<td>4</td>
<td>3yr</td>
</tr>
<tr>
<td>P3</td>
<td>3</td>
<td>2</td>
<td>Autism (mild-moderate)</td>
<td>M</td>
<td>5</td>
<td>3yr</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Autism (mild-moderate)</td>
<td>M</td>
<td>5</td>
<td>5yr</td>
</tr>
<tr>
<td>P4</td>
<td>2</td>
<td>2</td>
<td>Asperger Disorder</td>
<td>M</td>
<td>14</td>
<td>14yr</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>PDD-NOS</td>
<td>M</td>
<td>13</td>
<td>3yr</td>
</tr>
<tr>
<td>P5</td>
<td>2</td>
<td>2</td>
<td>Autism</td>
<td>M</td>
<td>10</td>
<td>18mo</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Asperger Disorder</td>
<td>F</td>
<td>8</td>
<td>7yr</td>
</tr>
<tr>
<td>P6</td>
<td>3</td>
<td>3</td>
<td>PDD-NOS</td>
<td>M</td>
<td>12</td>
<td>7yr</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Autism</td>
<td>M</td>
<td>9</td>
<td>~2yr</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Autism</td>
<td>M</td>
<td>7</td>
<td>~1yr</td>
</tr>
<tr>
<td>P7</td>
<td>2</td>
<td>2</td>
<td>Asperger Disorder</td>
<td>16</td>
<td>F</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Asperger Disorder</td>
<td>12</td>
<td>F</td>
<td></td>
</tr>
</tbody>
</table>
**Study Design**

To explore the experiences of parents who have more than one child with an ASD, we created a semi-structured interview guide (Appendix D) with open-ended interview questions. Questions were designed to explore participants’ first and second diagnostic experiences and the time leading up to each diagnosis, risk perception and family planning considerations, as well as beliefs about the etiology of ASD. Question design was based on previous literature regarding ASD and other childhood diagnostic experiences. The same semi-structured interview guide was used in every interview and questions were adapted based on the thoughts and experiences expressed by each participant.

**Data Management and Analysis**

All interviews were digitally recorded and transcribed by a professional transcription service, and data was imported into ATLAS.ti (version 6.2). Data analysis was rooted in grounded theory and followed qualitative method procedures outlined in Miles and Huberman (Glaser, 1992; Miles MB, 1984). Accordingly, interview transcripts were read through line-by-line, and sections of text were assigned codes based on a priori interview guide discussion points and emergent themes. Individual codes were then further grouped into "Super Code Families" reflecting broader themes found in the majority of interview transcripts. If a code was only mentioned on a single occasion, it was excluded from the data. In order to protect participant confidentiality, all databases containing interview transcripts in addition to any other identifying information were password protected.
RESULTS

Study participants were asked directly about their experiences surrounding the first and second ASD diagnosis, risk perceptions, and lifestyle challenges. Three major themes emerged from the data based on their appearance in four or more participant interviews. These themes include:

1. Parents experience feelings of confusion leading up to and during receipt of an ASD diagnosis and experience feelings of worry and grief for their child's future following receipt of this diagnosis.

2. Parents who have experienced a diagnosis of an ASD in one child are not more prepared for the diagnosis of an ASD in a second child.

3. Having a second child with an ASD creates additive emotional and lifestyle challenges in multiplex families. Parents of multiplex ASD families believe that they are more overwhelmed and more isolated than parents of simplex families.
Theme #1: Parents experience feelings of confusion leading up to and during receipt of an ASD diagnosis and experience feelings of worry and grief for their child's future following receipt of this diagnosis.

There were several questions in the interview that asked participants about their feelings leading up to the receipt of an ASD diagnosis. As previously documented in the literature, the majority of participants in this study described feeling lost prior to receiving an official diagnosis.

And it was just feeling lost, because you didn't know what was wrong with your child, you didn't know how to help him (P1).

In addition to feeling lost and alone, participants also discussed feeling confused leading up to and following receipt of the ASD diagnosis:

[I felt] very confused, because I had no idea what she was talking about. I will never forget that day (P5).

Consistent with literature on the emotional responses of parents receiving a diagnosis of a disability in a child, participants in this study described feeling grief for the loss of a "typical child," and expressed worry and concern about the uncertainties in their child's future.

When we officially had a diagnosis probably also a little grief… when you have something that is going to be with your child, a diagnosis that's going to be part of who he is for the rest of his life-- And you have to understand, Brian* was my first, and he's just so brilliant, I mean he still is. He was my child that was going to Harvard… and you [don’t] know… how this was going to affect his future, whether he's going to go to college. So you're relieved you finally have something, but then at the same time it's like a grieving process (P1). *name changed

You have these hopes and dreams, and you picture your children in a certain way and things they’ll do, and then you get that piece of paper, and all of that changes (P2).

Research examining the emotional experiences of mothers caring for a child with a developmental disability show that there is no clearly defined final stage of acceptance, and that there may be cyclic emergence and re-emergence of grief. Participants discussed...
having good days and bad days and the difficulty coping with the reminders of their child's limitations on a bad day.

You're always in step one. You're always just trying to figure out what to do. And then maybe this will work this week, but it doesn't work next week... you feel very bipolar. Like when you make progress you feel like, 'Ok good, we're getting past this. The skill has been achieved.' And you forget that part of autism is that some days a fully potty trained child will walk up to the toilet and pee his pants in front of the toilet, because he just forgets to pull his pants down (P3).

**Theme #2: Parents who have experienced a diagnosis of an ASD in one child are not more prepared for the diagnosis of an ASD in a second child.**

Questions during the interviews also specifically focused on the emotions surrounding the *second* diagnostic experience. Many participants described a time leading up to the second diagnosis when they were fixated on the behavioral *differences* between their first child with an ASD and their second child, for whom they had suspicions of an ASD. While participants often acknowledged seeing similarities in hindsight, participants described their focus on any evidence that could reassure them that their second child did not have an ASD:

But with us it was tough, because Alex* was reading and writing at an early age and talking, so we didn’t see the comparison at that point in time (P4).

So, for us the problem with that, when we thought of autism we thought of Noah*. So because Jack's* behaviors do not match Noah's behaviors truly, we thought, 'Oh this is something different. This is him mimicking behavior, this is a twin thing. This is learned behavior. This is environment. This is something different (P3).

Participants had difficulty accepting their suspicions of an ASD in their second child and their familiarity with autism spectrum disorders did not seem to make them more able to recognize the diagnosis in a second or third child, nor did it make them better prepared emotionally to receive the subsequent diagnosis. In addition, many participants felt that once they did share their concerns about their subsequent child/children with healthcare providers, the concerns were not taken seriously.
I knew, but they kept telling me, "No." And then you second-guess yourself again, because you feel like, 'Okay, am I just seeing this?' (P1).

So I took him back. Denied again. They said he didn't need speech services. But it just would not sit with me. And then there were too many-- It wasn't just the communication. There were too many [signs]… (P3).

Not only is clinician acceptance of parental suspicion prior to diagnosis a key factor contributing to parental satisfaction during the diagnostic process, but also, the provision of resources and delineation of a plan is critical to parents when receiving a first or second diagnosis.

That would have been really helpful if I left with-- I had to find the resource center myself. You would think that would have come with your packet. You should leave with a diagnosis and a packet of information and, 'This is what he has. This is who you contact. This is when you follow up' (P4).

Additionally, participants spoke to the innate variability of autism spectrum disorders and the difficulty such variation presents for resource identification:

But I think that is so important, that parents are equipped…Nobody tells you what to do, because honestly nobody knows what to do…It's not like it's muscular dystrophy or cancer or something tangible that you say, "This is what it is. This is what we do." And I'm not saying it's worse than those things, because it's not, but it's different, because… you're in a constant state of research (P3).

**Theme #3: Having a second child with an ASD creates additive emotional and lifestyle challenges in multiplex families. Parents of multiplex ASD families believe that they are more overwhelmed and more isolated than parents of simplex families.**

Participants were subsequently asked about their family planning decisions following both diagnoses and most participants agreed that having additional children with an ASD would be emotionally overwhelming and potentially unmanageable.

Oh no, I decided not to have any more children after I had Steven* diagnosed with autism… because from what everybody was telling me it was more than just having a regular kid, it was like having Steven being sextuplets, like being six (P5).

The idea that caring for one child with an ASD can be equated to caring for more than one typical child was a reoccurring theme:
So it’s like instead of three kids, they’re like 12. And if you have one child with autism, that is difficult, but it’s just not as difficult as if you have three kids with autism (P6).

On more than one occasion in each narrative, participants stressed the distinction between their children regardless of their similar diagnoses. A recurrent theme that emerged from these narratives was that participants wanted others to understand that every ASD diagnosis is distinct and unique:

{People] don't understand that one child is not going to be another child (P1).

When you have kids with multiples, they’re not all the same and their autism diagnosis - their autism is not all the same. They’re all different… (P6).

As described by participants, the variable phenotype of autism created additive parenting challenges:

I've just crossed my fingers and hope that only one of them will be broken at a time so I can fix them and then get to the other one later when she has a problem (P7).

Furthermore, participants expressed feeling a degree of isolation unique to their multiplex situation, and unanimously agreed that they feel that having more than one child with an ASD is more difficult and overwhelming than having a single child with an ASD.

I know that I’ve found it kind of isolating, and I think that rather than feel more connected to other parents I felt less and less over time (P7).

And the whole ride home I'm just thinking, 'Okay, we have three kids. Most of them are autistic, like two out of three kids.' I just felt like my whole life laser beamed in on autism, like, this is very much what our world is going to be about. I feel a little more isolated… and I do feel like it affects the family differently, because like I said you're not just accommodating this one person. I think once you hit two it sort of takes the family over, like that's what you're about all of a sudden, whether you want to be or not (P3).
DISCUSSION

In many ways, this study demonstrates that parents of multiplex ASD families have experiences similar to parents of simplex ASD families and other parents receiving a special needs diagnosis in a child. Previous literature documenting parental experiences in the diagnosis of autism spectrum disorders has consistently demonstrated that receiving an ASD diagnosis can be a distressing event where parents may experience significant feelings of confusion and frustration (Howlin & Moore, 1997; Keenan, et al., 2010; Midence & O'Neill, 1999). Congruent with the earlier literature, six out of the seven participants in this study stated feeling confused leading up to and during receipt of an ASD diagnosis in their child/children. Confusion during this process exacerbated participants' isolation throughout the experience, which led to difficulties sharing the diagnosis with family and friends. As found in other studies, participants reported feelings of distress and despair in their inability to understand their child's behaviors and diagnosis, which can lead to strained communication with family and friends (Midence & O'Neill, 1999).

In addition to these feelings of confusion leading up to an ASD diagnosis in a child, parents have reported a variety of feelings when receiving a diagnosis of an autism spectrum disorder including shock, worry, grief, fear, isolation, anger and sadness (Cameron, 1992; Siegel, 1997). Consistent with this literature on the emotional responses of parents of children with disabilities, participants in this study described feeling grief
for the loss of a "typical child," in addition to feelings of worry and concern for their child's uncertain future. Research examining the emotional experiences of mothers caring for a child with a developmental disability show that there is no clearly defined final stage of acceptance in these parents, and that these initial emotions of loss and grief emerge and re-emerge with developmental milestones (Cameron, 1992). Many of the participants in this study described this phenomenon of cyclic grief, especially with regard to parenting more than one child with an ASD. Participants discussed the feeling of having good days and bad days and the surge of negative emotion with the reminders of their child's limitations on a bad day. Participants also described the added grief and confusion when parenting two children with an ASD whose needs are similar, but distinct, and the struggle of balancing the inevitable bad days. The findings of cyclic grief in this study add to the previous research by confirming the significance of this phenomenon in parents of children with an autism spectrum disorder, similar to parents of children with other disabilities or genetic diseases. Furthermore, this study demonstrates that although parents of two children with an ASD are grieving the same diagnosis in both children, the grief is not only distinct, but also additive.

While the emotions surrounding an ASD diagnostic experience may be similar among simplex and multiplex families, and even analogous from the first to the second diagnosis in a multiplex family, this study also demonstrates that parents of multiplex families have unique experiences distinct from the simplex ASD community. Narratives from this study reveal that while parents of multiplex ASD families may be familiar with autism spectrum disorders, they still do not feel prepared when faced with the possibility of a second ASD diagnosis. For example, three study participants (P1, P3, and P4)
reported an initial fixation on the behavioral differences between their first child with an ASD and their second child, for whom they had suspicions of an ASD. While they acknowledged similarities in hindsight, they described a time when they were focused on what differentiated their children from one another. This intense focus on differences may represent the initial stage of adaptation where acceptance of a diagnosis is still challenging. More specifically, participants may have been focused on the behavioral differences as a way to remain hopeful of the alternative until they were truly ready to accept their suspicions of an ASD in their second child. Therefore, while parents of multiplex families have a familiarity with autism spectrum disorders, acceptance of an ASD diagnosis in a second child is not only challenging, but also emotionally complicated by parents' difficulty acknowledging their suspicions of an ASD and their initial focus on substantiating evidence for the alternative.

Parents' preoccupation with these differences most likely dissipates with the progression through the process of adaptation to a stage of acceptance, and is subsequently replaced with a focus on obtaining the ASD diagnosis in their second child. Previous literature has demonstrated that a contributing anxiety in families is that many parents are worried and suspicious about their child's behavior before professionals acknowledge the problem (Howlin & Moore, 1997). In this study, the same participants (P1, P3 and P4) that initially fixated on their children's differences felt that once they were able to accept the possibility of an ASD in their second child, their views were not given appropriate consideration and weight by clinical professionals. While recent studies have demonstrated positive trends in parental satisfaction with increasing acknowledgment of parental suspicion by professionals, these studies are limited in that
they capture only single diagnostic experiences (Brogan & Knussen, 2003). It could be that among our study participants, a lack of acceptance by clinicians was due to a lingering ambivalence expressed by the participants. In other words, these parents might have felt as though they were ready to accept their own suspicions of an ASD, but outwardly expressed uncertainties regarding the ASD diagnosis. Additionally, as previously mentioned, sibling recurrence risks were believed to be low (5-8%) and thus health care providers might not have felt that there was sufficient reason to be highly attuned to subsequent diagnoses in siblings. Previous research clearly shows that clinician acceptance of parental suspicion prior to diagnosis is a key factor contributing to parental satisfaction during the diagnostic process, and it is therefore important that clinicians remain aware of and sensitive to this for multiplex ASD families (Howlin & Moore, 1997).

Following receipt of an ASD diagnosis, the research literature has documented the importance of providing parents with information, resources and a plan (Brogan & Knussen, 2003). Our participants almost unanimously emphasized the importance of providing information, resources and a plan to parents receiving a first and second diagnosis of an ASD in a child. These findings clearly support the previous research and solidify the recommendations that clinicians should provide a clear plan when delivering a diagnosis to families in addition to providing families with information about ASDs and accessible resources.

Not yet explored or documented in the literature are the experiences and emotions surrounding parenting more than one child with an ASD. Of note in autism spectrum disorders is the potential variability of phenotype – participants not only stressed the
distinction among their children with an ASD, but also explained that these differences create additive parenting challenges and lifestyle stressors. Participants also spoke to the degree of isolation experienced by their families, and unanimously agreed that they perceived having more than one child with an ASD as more difficult and overwhelming. While they recognized that parenting any child with a disability is filled with unique challenges, participants discussed feeling a distinct difference when the majority of children in a family need extra care and attention. For example, one participant described her experience as having her life "laser-beamed" in on autism. It is thus important for health care providers to understand that having one child with an ASD does not prepare parents for having an additional child with an ASD and feelings of increased isolation and stress in multiplex families may require improved attention by providers and increased provision of resources and support.

**Limitations and Future Research**

This was a pilot study using qualitative methods to explore parents' experiences receiving a diagnosis of an autism spectrum disorder in more than one child and to identify the potentially unique lifestyle challenges in multiplex families. The primary limitation in this study was the small, homogeneous sample, and the subsequent limited ability to generalize the findings in this study to all families of children with more than one child diagnosed with an ASD. Additionally, participants in the study were recruited through online support groups and agencies that provide services to children with ASDs, suggesting that these parents were active in the ASD community, and therefore would be more likely to volunteer than the general population. As such, the results of this study
may not represent the views and experiences of all parents who have more than one child with an autism spectrum disorder.

Another limitation in this study is that all interviews were conducted over the phone for convenience to participants. Conducting interviews over the phone allowed greater access for participation, but may have influenced the way the interviewer built rapport with the participant and vice versa. However, it is important to note that phone interviews also allow participants anonymity and the opportunity to openly express emotions. A final limitation of this study is that the information was obtained through self-report. Participants in this study may have been biased in their self-disclosure procedures regarding their experiences. In order to minimize fear of identity or judgment, we assured each participant of confidentiality procedures.

While limitations to this study exist, the themes identified in this data can be used to guide further research into the experiences of multiplex ASD families. Further research should include additional qualitative studies with a large number of participants that compare and contrast the first and second diagnostic experiences in these families. Another recommendation for future research would include a deeper look into family planning and risk perception in multiplex families. Also, with the more recent studies showing increased sibling recurrence risk as well as increased knowledge of genetic influences, it might be interesting to survey genetic counselors regarding their role in and experience with the ASD diagnostic process and risk assessment procedures.
CONCLUSIONS

In this study we explored the emotions surrounding the diagnostic experiences of parents who have more than one child with an ASD. Through semi-structured interviews, participants described their experiences receiving and coping with a diagnosis of an ASD in more than one child, explored the significance of these experiences as they related to family planning decisions and discussed the unique emotional and lifestyle challenges that accompany raising more than one child with an autism spectrum disorder.

Data from this study is concordant with previous literature suggesting that parents experience significant levels of confusion leading up to and during receipt of a single ASD diagnosis. However, this study further demonstrates that while parents may be familiar with autism spectrum disorders and have suspicions in subsequent children, they are still emotionally unprepared to acknowledge an additional diagnosis. With previous literature demonstrating significant sibling recurrence risks and the current data revealing the emotional challenges surrounding the receipt of a second ASD, it is important that health care providers not only be mindful of the potential of an ASD in more than one child within a family, but also remain sensitive to the unique emotions experienced by these multiplex families.

As reported in previous literature following receipt of an ASD diagnosis, participants in this study identified feelings of grief for the loss of a "typical child." Although parents of two children with an ASD are grieving the same diagnosis in both children, it is apparent from this study that the cyclic grief experienced surrounding each
child is not only distinct, but also additive. Furthermore, parents of multiplex families discussed feeling increased levels of isolation when the majority of children in a family need extra care and attention. More specifically, multiplex families may have a lifestyle that is defined by a "laser-beamed" focus on autism. These findings make a significant contribution to the literature as they provide insight into the unique experiences of parents who have more than one child with an ASD. Results from this study help to reinforce areas in which health care professionals can provide additional support to parents of multiplex families during the diagnostic process including increased sensitivity to a second diagnosis, access to resources and information following receipt of both diagnoses, and continued counseling and support to help prevent further isolation from the community.
REFERENCES


Are you a parent or caregiver with more than one child with an autism spectrum disorder (ASD)?

If you answered YES, you have the opportunity to participate in a research study looking at parents’ experiences receiving an ASD diagnosis in more than one child.

What will study participation involve?
A one hour audiotaped face-to-face or telephone interview. All participants will receive a $25 gift card in appreciation of your time.

When will the interviews take place?
Interviews will take place during the month of February and will ideally be completed no later than February 29th, 2012.

It is our hope that the stories shared by parents and caregivers will be useful in educating genetic counselors and other health professionals about the experiences in families with more one child on the autism spectrum.

If you are interested in participating in this study, please contact me by email at: ichilton@brandeis.edu

I look forward to hearing from you!

Sincerely,
Ilana Chilton
Genetic Counseling Graduate Student
Brandeis University, Waltham MA
APPENDIX B
Eligibility Screening Tool

Basic Demographics
Name: ___________________________________
Location (City, State): ___________________________________
Contact Information (phone, email): ___________________________________
How did you hear about this study? _________________________________________

Questions for potential participants:

1. Are you over the age of 18? YES NO

2. How many children do you have? How old are they?
________________________________________________________________________
________________________________________________________________________
________________________________________________
________________________________________________

3. What are their specific diagnoses and how were they diagnosed?
   • e.g. child neurologist, child psychiatrist, developmental behavioral pediatrician, early intervention specialist
________________________________________________________________________
________________________________________________________________________
________________________________________________________________________

4. Would you be willing to meet for an in-person interview lasting for about 1hr? If not, would you be willing to participate in a telephone interview lasting for about 1hr?
________________________________________________________________________
________________________________________________________________________

☐ Participant meets eligibility criteria
APPENDIX C
Informed Consent Form

BRANDEIS UNIVERSITY
DEPARTMENT OF BIOLOGY
GENETIC COUNSELING GRADUATE PROGRAM

Informed Consent to Participate in Research

Autism spectrum disorder (ASD) in multiplex families: a qualitative study of diagnostic experiences and parental perceptions

Principal Investigator: Beth Rosen-Sheidley
Student Researcher: Ilana Chilton

INTRODUCTION
Ilana Chilton is a graduate student in the Genetic Counseling Program at Brandeis University. She is conducting a research study to learn more about the diagnostic experiences and perceptions of parents who have more than one child with an Autism Spectrum Disorder. Beth Rosen-Sheidley is a Professor of the Practice of Genetic Counseling and Co-Director of Research and Professional Development for the Brandeis University Genetic Counseling Program.

You are being invited to participate in this study because you are a parent/caregiver of more than one child on the autism spectrum.

Your participation in this study is completely voluntary. You should not feel any pressure to participate. You can decide to stop taking part in this research study at any time for any reason.

Please read all of the following information carefully. Ask any questions that you have about this research study. Do not sign this consent form unless you understand the information in it and have had your questions answered to your satisfaction.

If you decide to take part in this research study, you will be asked to sign this form. You will be given a copy of the signed form. You should keep your copy for your records. It has information, including important names and telephone numbers, to which you may wish to refer in the future.
PURPOSE OF STUDY
The purpose of this study is to (1) describe parents’ experiences of receiving a diagnosis of an autism spectrum disorder in more than one child, (2) explore the significance of this experience as it relates to family planning and beliefs about the disorder, and (3) describe the potentially unique lifestyle challenges in families with multiple children on the autism spectrum.

It is our hope that the information shared by participants in this study will be useful in educating genetic counselors and other health professionals about the unique emotional, social, and diagnostic experiences in parents who have more than one child on the autism spectrum, and, ultimately, help to inform the diagnostic management and care for these families.

PROCEDURES TO BE FOLLOWED
You will be asked to participate in an audiotaped face-to-face or telephone interview lasting approximately one hour. During this interview you will be asked questions regarding your experience surrounding the diagnosis of your second child with an autism spectrum disorder, as well as your beliefs and perceptions about this disorder.

RISKS
Participation in this study presents no more than minimal risk. However, it is possible that by taking part in this interview, you may experience thoughts or feelings that are upsetting to you. Should that occur, Beth Rosen-Sheidley is available to talk with you.

BENEFITS
There will be no direct benefit to you for your participation in this study. We hope that, in the future, information obtained from this study will help us gain a better understanding of the unique diagnostic and lifestyle experiences of parents with multiple children on the autism spectrum.

ALTERNATIVES
An alternative is to not participate in this research study.

PRIVACY AND CONFIDENTIALITY
All records containing identifying information, such as names, email addresses, telephone numbers, and home or work addresses will be kept strictly confidential during the study. All study related documents and materials (including consent forms, interview transcripts, and audiotapes) will be kept in a secure location accessible only to the student researcher. Any databases containing identifying information will be password protected using a password only known to the student researcher. Transcripts, interview notes, and audiotapes will be labeled with a coded ID number, which will be assigned to you upon
enrollment in the study. If you are quoted or referred to in any written or oral reports of the study, you will be given an alternative name. You will never be referred to by your real name or any other identifying information in any written or oral reports based on the interview.

PAYMENT
You will receive a $25 gift certificate to Amazon.com for participation in the research study as a gesture of appreciation for your time and expertise.

COST
There will be no cost to you to participate in the study, other than the time it takes to conduct this interview.

WHOM TO CONTACT
If you encounter any problems related to study participation or have questions about the study, you may contact the Student Researcher, Ilana Chilton, at ichilton@brandeis.edu or (913) 269-0069.

You may also contact the Principal Investigator for this project, Beth Rosen-Sheidley, at Sheidley@brandeis.edu or (781) 736-2336.

If you have questions about your rights as a research study subject, contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at (781) 736-8133.
PARTICIPANT’S STATEMENT

I have read this consent form and have discussed with Ilana Chilton the procedures described above. I have been given the opportunity to ask questions, which have been answered to my satisfaction. I understand that any questions that I might have will be answered verbally or, if I prefer, with a written statement.

I understand that my participation is voluntary. I understand that I may refuse to participate in this study. I also understand that if, for any reason, I wish to discontinue participation in this study at any time, I will be free to do so.

If I have any questions concerning my rights as a research subject in this study, I may contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8133.

I have been fully informed of the above-described study with its risks and benefits, and I hereby consent to the procedures set forth above.

I understand that as a participant in this study my identity and data relating to this research study will be kept confidential.

I consent to an audio recording of my interview. ___________________________________________ Participant’s Initials

______________________  ____________________________
Date  Participant’s Signature

______________________  ____________________________
Date  Student Researcher Signature
APPENDIX D
Interview Guide

Participant ID: __________
Date/Time: _______________________

I. Introduction and Informed Consent:
   i. Explain the presence and purpose of recording equipment and note-taking:
      • I would like to audiotape our interview so that I don’t miss anything. I may also take a few notes during the interview. Afterward, the interview will be transcribed by a confidential transcription service and your name will not be associated with it. Does that sound ok?
   ii. Briefly review the aims of the study and purpose of interviews:
      • I want to understand what it is like being a parent of two children with autism and what it was like getting each of those diagnoses.
   iii. Review the signed informed consent:
      • Let the participant know that he/she can take a break or terminate the interview at any time for any reason

II. Background Information:
   Age: _______ State of residence: _______ Occupation: __________________
   Ethnicity:
   _____ African-American    _____ Asian-American    _____ Caucasian
   _____ Hispanic/Latino      _____ Native American  _____ Other
   Education:
   _____ high school diploma    _____ some college    _____ bachelors degree
   _____ some graduate work     _____ master’s degree  _____ doctoral degree
   _____ vocational or professional certificate and/or associates degree

Tell me a little bit about who is who in your family (acquiring pedigree)
   a. Prompts: who lives in the house, names and ages of immediate family members, specific diagnoses for each affected child.

III. Experience receiving the first diagnosis of an ASD in your family:
I’d like to switch gears and discuss your experience receiving a diagnosis of an ASD in your first child. I’m going to start by asking you about what it was like before you learned your child had an ASD.

   1. Think back to the time leading up to the diagnosis.
      a. What was it that first led you to be concerned about _____? How old was he/she?
      b. How much time went by between when you first became concerned and when you received a diagnosis? If there was a time lapse – Why?
      c. How would you describe your feelings leading up to the first diagnosis?
         i. Prompts: confused, guilty, isolated, relief, etc.
2. Let’s talk about when you received the official diagnosis.
   a. Who did you receive the diagnosis from?
   b. Tell me about this experience [with the provider].
      i. Prompts: was it personable (cold/warm)? Supportive? Quick?
   c. How did you feel when he/she was giving you the diagnosis?
      i. Prompts: relief, confusion, despair, blame, guilt, doubt, etc.
   d. I imagine there might have been a follow-up plan, can you tell me about that?
      i. Prompts: EI, schooling, follow-up medical appointments
   e. Tell me about things you wish would have been different.
      i. Prompts: delivery of news, timeliness of diagnosis, content, support

3. What impact did this diagnosis have on you and your family?
   a. Tell me about your family dynamics before and after the diagnosis.
      i. Prompt: Changes in family dynamics? family activities? dynamics with spouse? Did you move? Did you change schools?
      ii. What was the most notable change?
   b. Did there come a time when you (and your family) felt better adjusted?
   c. What was your way of getting information about ASD? When?
      i. Prompts: support group, informational brochures, online, physicians, school
   d. How did your feelings/family coping change over time, how would you describe yourself now?
      i. Prompts: anger, guilt, acceptance, understanding

IV. Risk perception, etiological beliefs, and family planning:
1. What do you view as the cause(s) of ASDs?
   a. Prompts: genetic influences, vaccinations, complications during birth, illness, diet, infection, don’t know, etc.
   b. If participant mentions inherited factors, ask about genetic testing:
      i. Who did you meet with?
      ii. What was this experience like for you?
      iii. What were the results?
2. What do you view as the risk/odds associated with having a second child with an ASD?
3. How did you come to decision to have another child?
   a. Did you think about the chances that another child might have an ASD?
   b. What were the factors that played into your feelings about the risk of having a second child with an ASD?
i. Prompts: physicians, GCs, online information, friends/family, support groups, personal feelings

Q. If you could give one statement of advice to health care professionals delivering the news of a second diagnosis in a family, what would you say?

IV. Wrap-up:
We have reached the end of the tape-recorded portion. Before I stop recording, do you have anything else you would like to add? Was there anything you thought I was going to ask but didn’t?

Closing comments:
   i. Thank the participant and ask them how the interview experience was for them.
   ii. Remind them that Beth Rosen-Sheidley is available as a resource if they experience any emotional distress following the interview
   iii. Reassure participant of confidentiality of responses