Down Syndrome, Autism, and Parent Experiences with the Dually Diagnosed

Master's Thesis

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The Faculty of the Graduate School of Arts and Sciences
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Judith Tsipis, PhD, Advisor

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Master of Science
in
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by
Elizabeth Kellom

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Abstract

Down Syndrome, Autism, and Parent Experiences with the Dually Diagnosed

A thesis presented to the Graduate Program in Genetic Counseling

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Elizabeth Kellom

As of 1999, only 36 instances of individuals with both Down syndrome and Autism Spectrum Disorder (DS-ASD) were described in the medical literature. Many conflating factors presumably delayed or prevented the accurate diagnosis of DS-ASD at the time, though the most striking barrier was a persistent belief in the medical community that the two conditions did not or could not coexist. Extensive work exists delineating the nature and impact of parental stress when a child is born with either Down syndrome or Autism Spectrum Disorder. Within these studies, cohorts of parents of children with either disability are frequently compared to each other, yet investigations into the experience of parents of the dually diagnosed are absent. We interviewed six parents to better understand the diagnostic process and the challenges and joys involved when raising a child with DS-ASD. We coded interviews in ATLAS.ti (version 7.0) and analyzed for themes. We found that parents were usually the ones to suggest their child had autism and most were able to find information about the dual diagnosis from either their doctor or their own Internet research. While the diagnosis of Down syndrome often came as a shock, the autism diagnosis brought a sense of validation to parents who had noticed differences in their child for years. Five of the six parents expressed feelings of alienation from parent or child
groups that focused on solely one diagnosis. The formal diagnosis of autism created access to new services, though utilization varied. This study suggests the importance of accurate, timely diagnosis and appropriate support services for child and family.

Keywords: dual diagnosis – autism – Autism Spectrum Disorder – Down syndrome – DS-ASD – diagnostic odyssey – support groups
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Introduction

In 1999, only 36 instances of individuals with both Down syndrome and Autism Spectrum Disorder (DS-ASD) were described in the medical literature (Capone, 1999; Coleman, 1986). Many conflating factors presumably delayed or prevented the accurate diagnosis of DS-ASD at the time, though perhaps the most striking barrier was a persistent belief in the medical community that the two conditions did not or could not coexist (Froehlke & Zaborek, 2013). Features of autism were instead attributed to the level of cognitive deficit in children with Down syndrome (Froehlke & Zaborek, 2013; Patterson, 1999). The existence of this bias against a dual diagnosis among healthcare providers has never been systematically investigated, but anecdotal reports from disappointed parents support its existence (Froehlke & Zaborek, 2013; Medlen, 1999).

This misconception slowly changed as research into the clinical picture and prevalence of both ASD and DS-ASD progressed (Capone, 1999; Reilly, 2009). In 2016, the Centers for Disease Control’s Autism and Developmental Disabilities Monitoring (ADDM) program reported that approximately 1 in 68 children (1.5%) in the United States has been diagnosed with Autism Spectrum Disorder (ASD). In the past decade, investigators have also evaluated the prevalence of ASD among those diagnosed with Down syndrome and have found it to be significantly higher than that of the general population, though exact numbers vary (Reilly, 2009). Additional research has attempted to refine the diagnostic picture and determine the utility of various questionnaires in making a diagnosis of ASD (Capone, Grados, Kaufmann, Bernad-Ripoll, & Jewell, 2005; Carter, Capone, Gray, Cox, & Kaufmann, 2006; DiGuiseppi et al., 2010; Moss, Richards, Nelson, & Oliver, 2013; Warner, Moss, Smith, & Howlin, 2014). While the medical and research communities are progressing in their acknowledgement and understanding
of DS-ASD, no investigations have looked into the parent experience raising a child with this dual diagnosis.

Extensive work exists delineating the nature and impact of parental stress when a child is born with a disability (McCubbin et al., 1980). Within these studies, cohorts of parents of children with Down syndrome are frequently compared to parents of children with Autism Spectrum Disorder (Esbensen, Bishop, Seltzer, Greenberg, & Taylor, 2010; Siklos & Kerns, 2006), yet these studies do not address those with a dual diagnosis. This may add to the social or medical alienation parents of children with DS-ASD face. If parents do not receive appropriate supports within their community – personal, medical, disability, or otherwise – their mental or physical health can suffer (Cantwell, Muldoon, & Gallagher, 2014; Greenberg, Seltzer, Krauss & Won Kim, 1997). Stressed and struggling parents can also have a negative impact on outcomes for their child with a disability (Robbins, Dunlap, & Plienis, 1991). The sources of stress include achieving an accurate and timely diagnosis, dealing with aspects of the child’s behaviors, and difficulty accessing social, educational, and medical support (Dunn, Burbine, Bowers, & Tantleff-Dunn, 2001).

For parents of children with DS-ASD, the support groups for the wider Down syndrome or autism only communities may not be as helpful (Froehlke & Zaborek, 2013; Medlen, 1999). When mothers of children with disabilities are not utilizing support groups, reasons they may give include “distance from group, family responsibilities, work responsibilities, lack of interest, pessimism that such a group could be useful, and opposition from their spouses” (Shapiro, 1989). In the case of families of children with DS-ASD, additional barriers specific to their situation may decrease participation. For example, the DS- or ASD-only support groups may not deal with the behavioral issues or learning difficulties relevant to parents of children with DS-ASD. In
addition, the abilities and progress of other children in the group may be disheartening for parents of children with the dual diagnosis. Anecdotally this appears to be the case (Capone, 1999; Froehlke & Zaborek, 2013; Medlen, 1999), though no formal studies exist to answer this question.

Several gaps exist in understanding the needs and experiences of parents raising children with DS-ASD. By reaching out through the Down Syndrome-Autism Connection, a nonprofit organization serving such families, we connected to six parents and interviewed them about their family and their experiences raising a child with a dual diagnosis. Questions aimed to illustrate the process of achieving the formal autism diagnosis, their reaction to the diagnosis, the impact of the second diagnosis on the availability of services for their child, and their ability to find information and support for themselves. Our data analysis uncovered themes that demonstrate the importance of appropriate support services for such families and provide insight into the diagnostic process and day-to-day joys and challenges of raising a child with DS-ASD.
Methods

Study Design

This was a qualitative study in which the author conducted semi-structured phone interviews with six parents of children with a dual diagnosis of DS-ASD. Prior to conducting the phone interview, we sent a survey (Appendix D) to the families to assess their demographics, child’s communication abilities and/or other health issues, and the details of how their child was diagnosed. We attached an information sheet (Appendix C) with the survey and asked participants to read it ahead of the interview. The Brandeis University Institutional Review Board approved this study after granting expedited review.

Sample and Recruitment

On our behalf, the president of the Down Syndrome Autism Connection [reference site: http://www.mdsc.org/programs/DownSyndromeAutismConnection.cfm] emailed a recruitment notice (Appendix B) to a group of approximately 30 adult parents (18+) with children aged 10-12 with DS-ASD. All email recipients were members of the Down Syndrome-Autism Connection. Several other parents with children outside the original age range heard about the study by word of mouth and contacted the author asking to participate. To accommodate these parents, we modified the IRB protocol to expand the age range from 10-12 to 5-15 years. In total, we sent out surveys to nine parents who contacted us. Six parents returned completed surveys. Upon receiving the completed survey, we scheduled a phone interview with each parent.
Data Collection Procedure

We acquired verbal consent from each participant prior to beginning the interview by asking if they had read the information sheet sent to them, whether they had any questions, and whether they agreed to participate in the recorded interview. We recorded the interviews with the use of the freeconferencecalling.com service. All interviews used the same interview guide (Appendix E), which consisted of open-ended questions. Each interview was between 23 and 37 minutes long. A transcriptionist from Mulberry Studios transcribed the interviews. All participants received a $25 Amazon.com gift card as a token of appreciation for their contribution. We stored all audio recordings and interview transcripts on box.com, an encrypted site hosted by Brandeis University.

Data Analysis

Prior to importing interview transcripts to ATLAS.ti (v.7.5.17) for analysis, we changed the names of participants and their children. The author performed all thematic analysis and coding, generating a total of 189 codes. We categorized the codes into the following five themes: (1) the journey to the dual diagnosis, (2) the reaction to the dual diagnosis, (3) the benefits of the dual diagnosis, (4) community and coping, (5) challenges and joys.
Results

Family Demographics

We interviewed six participants. All of the parents interviewed were mothers who were members of the support group “The Down Syndrome-Autism Connection.” Their children with DS-ASD children ranged from six to fifteen years old. One was adopted and five of the six were male [Table 1]. We changed all names to protect the privacy of participants.

<table>
<thead>
<tr>
<th>Parent Name</th>
<th>Child Name</th>
<th>Child Gender</th>
<th>Child Age</th>
<th>Adoption Status</th>
<th>Birth Order</th>
</tr>
</thead>
<tbody>
<tr>
<td>Helen</td>
<td>Isaac</td>
<td>Male</td>
<td>11</td>
<td>Not Adopted</td>
<td>2&lt;sup&gt;nd&lt;/sup&gt; of 2</td>
</tr>
<tr>
<td>Charlene</td>
<td>Phillip</td>
<td>Male</td>
<td>12</td>
<td>Not Adopted</td>
<td>2&lt;sup&gt;nd&lt;/sup&gt; of 2</td>
</tr>
<tr>
<td>Deborah</td>
<td>Chris</td>
<td>Male</td>
<td>11</td>
<td>Not Adopted</td>
<td>3&lt;sup&gt;rd&lt;/sup&gt; of 3</td>
</tr>
<tr>
<td>Samantha</td>
<td>Julian</td>
<td>Male</td>
<td>7</td>
<td>Adopted</td>
<td>2&lt;sup&gt;nd&lt;/sup&gt; of 2</td>
</tr>
<tr>
<td>Amber</td>
<td>Dylan</td>
<td>Male</td>
<td>15</td>
<td>Not Adopted</td>
<td>2&lt;sup&gt;nd&lt;/sup&gt; of 2</td>
</tr>
<tr>
<td>Michelle</td>
<td>Jennifer</td>
<td>Female</td>
<td>6</td>
<td>Not Adopted</td>
<td>3&lt;sup&gt;rd&lt;/sup&gt; of 4 (fraternal twin to 4&lt;sup&gt;th&lt;/sup&gt;)</td>
</tr>
</tbody>
</table>

Table 1: Participant Demographics

Three of the parents brought up their own personal experience with the disability community before having a child with special needs. Deborah’s brother passed away from complications of Down syndrome. Samantha volunteered with the Special Olympics and her husband’s aunt has Down syndrome. Michelle worked in her school’s special education classroom for a time. We did not ask participants for their specific location, but in scheduling the
interviews, we learned that four participants called in from Eastern Time, one from Central Time, and one from Mountain Time.

We asked participants to describe their child’s ability to communicate. The methods of communication varied, but only one parent had a child who could speak in some full sentences, which his mother referred to as “semi-verbal.” Other children relied on sign language and gestures while two used a communication device of some kind [Table 2].

Table 2: Communication Abilities

<table>
<thead>
<tr>
<th>Name</th>
<th>Sign Language</th>
<th>Noises</th>
<th>Gestures</th>
<th>Some Spoken Words</th>
<th>Communication Device</th>
<th>Full Sentences</th>
</tr>
</thead>
<tbody>
<tr>
<td>Isaac</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td></td>
<td>Limited</td>
</tr>
<tr>
<td>Phillip</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Chris</td>
<td>+</td>
<td>+</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Julian</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Dylan</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td></td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Jennifer</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td></td>
<td></td>
<td>+</td>
</tr>
<tr>
<td>Total</td>
<td><strong>5</strong></td>
<td><strong>4</strong></td>
<td><strong>5</strong></td>
<td><strong>3</strong></td>
<td><strong>2</strong></td>
<td>~1</td>
</tr>
</tbody>
</table>

In addition to communication abilities, we asked parents about any other health conditions their children currently have or had earlier in life. Previous research has suggested an association between medical complications and the development of autism in a child with Down syndrome (Reilly, 2009). All children were born with a heart defect, ranging from a small hole in the heart not requiring surgery to defects which required multiple surgeries. We asked specifically about a history of epilepsy and leukemia but no parents noted those conditions in their children [Table 3].
Table 3: Other Health Conditions

<table>
<thead>
<tr>
<th>Name</th>
<th>Heart Defect</th>
<th>Partial Agenesis of the Corpus Callosum</th>
<th>Hypothyroidism</th>
<th>Pulmonary Hypertension</th>
<th>Intestinal Blockage</th>
<th>ADHD</th>
<th>Celiac Disease</th>
<th>Sleep Apnea</th>
</tr>
</thead>
<tbody>
<tr>
<td>Isaac</td>
<td>+</td>
<td>+</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Phillip</td>
<td>+</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Chris</td>
<td>+</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Julian</td>
<td>+</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Dylan</td>
<td>+</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Jennifer</td>
<td>+</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>6</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
</tr>
</tbody>
</table>

In interviewing the participants, we addressed five major areas of interest regarding their experience raising a child with DS-ASD. These were (1) their journey to a dual diagnosis, (2) their reaction to receiving the formal dual diagnosis, (3) the benefits of having the formal diagnosis of DS-ASD (4) their support systems prior to and after their dual diagnosis, and (5) the challenges and joys of raising their child.

The Journey to a Dual Diagnosis

Prior to the interview, we asked parents about the diagnostic process for both Down syndrome and autism [Table 4]. Half of the children were diagnosed with Down syndrome at birth and half were diagnosed prenatally. Phillip was suspected of having Down syndrome prenatally and this was confirmed at birth to have Down syndrome. Most children were diagnosed with autism at 7-8 years of age, with the earliest diagnosis at age four and the latest at age fifteen. All children received the formal diagnosis of autism within the past five years. Though we did not ask parents specifically, most parents mentioned first noticing signs of autism, or signs of something being “different,” by ages 3-5. Four out of the six parents were the first to raise the possibility of autism, while psychologists and developmental pediatricians were the ones who made the formal diagnosis.
Table 4: The Dual Diagnosis

<table>
<thead>
<tr>
<th>Name</th>
<th>Timing of Down Syndrome Diagnosis</th>
<th>Age of Autism Diagnosis (years)</th>
<th>Who First Suggested Autism</th>
<th>Who Formally Diagnosed Autism</th>
</tr>
</thead>
<tbody>
<tr>
<td>Isaac</td>
<td>Prenatally</td>
<td>8</td>
<td>Developmental Pediatric</td>
<td>Autism Clinic</td>
</tr>
<tr>
<td>Phillip</td>
<td>Prenatally/At Birth</td>
<td>7</td>
<td>Parent</td>
<td>Developmental Pediatric</td>
</tr>
<tr>
<td>Chris</td>
<td>At Birth</td>
<td>7</td>
<td>Parent/Speech Pathologist</td>
<td>Psychologist</td>
</tr>
<tr>
<td>Julian</td>
<td>At Birth</td>
<td>4</td>
<td>Speech Pathologist</td>
<td>Developmental Pediatric</td>
</tr>
<tr>
<td>Dylan</td>
<td>Prenatally</td>
<td>15</td>
<td>Parent</td>
<td>Psychologist</td>
</tr>
<tr>
<td>Jennifer</td>
<td>At Birth</td>
<td>5</td>
<td>Parent</td>
<td>Psychologist</td>
</tr>
</tbody>
</table>

Parent familiarity with or knowledge of the possibility of co-occurring DS-ASD prior to their own child’s diagnosis was mixed. Most only discovered the possibility after suspecting something in their own child, which led them to research the dual diagnosis on the Internet and/or mention it to their doctor:

“We had heard mixed things from different therapists that were put to Isaac over the years. And I had done enough research about Down syndrome while we were pregnant with Isaac, and then just sort of as he was growing up. So, although we had heard from some professionals that it’s not possible to have both Down syndrome, and autism, I knew that was outdated, and so I knew that it was possible.” – Helen

“I hadn’t even thought about it. Like it didn’t even cross my mind that there would be more than one diagnosis.” - Samantha

“I didn’t know, but I suspected it, so I started Googling things, and then I saw that actually there were people with both diagnoses. I never even thought about it, or even heard about it, actually, beforehand. So, I started doing a little bit of my own research online, and realized that there are children out there with both.” - Michelle

While four of the parents were the first to bring up signs of something different going on with their child, for Samantha, the first mention of autism came from her son Julian’s speech therapist:

“I bit my lip to keep from bawling. I was devastated. I had that feeling of like, ‘Oh, we can’t do one more challenge.’ […] The speech therapist said, ‘Well, it’s clear to me he obviously has autism.’ And I – yeah, I was in shock. It took me by total surprise. I sat there through the whole speech appointment, like ready to burst out into tears.”
The doctors to whom parents raised concerns were generally receptive to their observations and made the appropriate referrals for evaluation. A few parents remarked on the long trail of referrals and long waiting periods for appointments. Two of the parents said that some of their friends and health professionals didn’t agree with their own observations and tried to be reassuring about the concerns they raised:

“Well, [Jennifer] had early intervention. I mentioned it to them, and they didn’t think so. They’re like, ‘No. We don’t see it.’ And I have a couple of family friends that are educators, and they’re like, ‘No, no, no, no. We don’t see it.’ So, everyone that I talked to kept saying, ‘No, no. We don’t see it.’ But I felt like I saw it.” – Michelle

“He was going to speech therapy at the time, and none of the speech therapists ever seemed to clue in that there could be something else involved, but it was this psychological neurologist that was like, ‘There’s something else going on. If he’s not even trying to talk, there’s something else going on.’ […] Even when I went back, and told his speech therapist, she said, ‘I would never have guessed he had autism, and like, I’m maybe just not around him enough.’” – Charlene

Diagnosis was delayed for three children because of their sociability. For Helen’s son Isaac, autism was mentioned as a possibility early on, but his social behavior made it easier to ignore:

“There were some indicators when he was very little, just like 18 months to two years old, where he was an early reader, and very interested in letters, and words, and obsessed with them. And at that time one of the early intervention therapists that worked with him had said, ‘Sometimes this is an indicator for autism.’ But then she said, ‘But yeah, but he’s very social, so I don’t know that that would be the case.’”

Samantha sought a second opinion for the diagnosis of her son, as the formal diagnosis was critical for her to qualify for a state services voucher. She said the following about her family’s first referral for an autism evaluation:

“We ended up going to a psychologist who was a joke. [Laughter] He said, ‘Oh, your kid high fives. He doesn’t have autism.’ I’m like, ‘Who are you? Do you know kids with autism? They high five.’ That means he hasn’t got this? But it was just a joke. He had no experience with the dual diagnosis, and my kid is fairly social. He just lacks all communication skills. He doesn’t know how to communicate. He doesn’t get what communication is.”

Amber went through the diagnostic process twice because of assumptions made about the social nature of her son, Dylan. He was first referred for evaluation at age eight but didn’t receive a formal diagnosis of autism until he was fifteen. Amber had this to say about the experience:
"He’s never had stranger anxiety. So, he was always – he’s always been friendly to so many people. And I think that just – she marked that as being social. And even though he didn’t have eye contact, and didn’t have a lot of words, [...] he still got his point across.”

**Receiving the Dual Diagnosis**

Nearly every parent expressed a sense of relief upon receiving the formal autism diagnosis. We asked them to reflect on differences between learning of the Down syndrome diagnosis and the autism diagnosis. For Helen, the diagnosis of Down syndrome arrived in the context of concern for Trisomy 18 or 13. When comparing the diagnostic experience of Down syndrome with that of autism, she had this to say:

"I think in a lot of ways it was similar in that there was grief, but there was also a sense of relief. So, with the Down syndrome diagnosis there was the grief there is something wrong with your child. And then, the relief that whatever was wrong was not incompatible with life. With the autism diagnosis, there was the compounded grief there’s something else, quote/unquote “wrong” with your kid, but the relief of knowing.”

Helen reflected upon her early sense of guilt about Isaac’s behavior. The autism diagnosis greatly decreased that burden. Helen said:

"[...] My child’s challenging behavior is not necessarily because we’re bad parents, or a bad mom, or he’s a bad kid. He’s got an additional struggle going on, and there may be at the very least understanding that, and beyond that having the additional resources to help address some of those things.”

Charlene’s son screened positive for Down syndrome prenatally but was not diagnosed for a few days after birth:

"It wasn’t so much of a shock, because I already knew there was a chance. I guess, in my case I had kind of a pre-warning, where it’s a bit different for a mother who thought their baby was going to be completely healthy, and then wasn’t.”

Her response to the autism diagnosis was similar in that she’d had time to acclimate to the potential new diagnosis, which eased the shock when it finally came:

"I guess because we had already been raising Phillip for seven years, and like I said it didn’t change who he was, and who we knew him to be, and who we loved, but it just kind of validated what we had kind of known all along that he was different, and at the same time I felt frustrated that it had taken so long that others in the therapy world never saw it.”
Michelle, who had no warning her daughter would be born with Down syndrome, agreed that the buildup of signs made the diagnosis of autism easier to bear:

"Hearing about the Down syndrome, there was no notice, no thought of it. It was like a shock, a major surprise. I took that pretty hard. For my husband and I, it was actually – like we went through a grieving period. It was really hard to take. With the autism, at least I think I was prepared over time, because I kept seeing it, and it was coming to me in little doses, rather than this huge, ‘Oh, she has Down syndrome.’ You know what I mean? I was thinking it for a while, so yeah, I was sad, and my husband was sad, but I guess I had more time to accept it.”

Nearly every parent reiterated in some form the sense of validation upon their child’s diagnosis:

“We knew something else was going on, and it explained a lot, and we were just like, ‘Okay, it’s a little frustrating, because he’s not like a lot of the other kids with Down syndrome. What’s going on?’ [...] A lot of parents are like devastated to get that second diagnosis, but for us it was like, ‘Okay, well, that makes sense.’” - Deborah

“It just confirmed what I was thinking, and I was like, ‘Okay, now I’m not going crazy. Like, I really do see this.’” - Michelle

Samantha and her husband adopted Julian knowing he had Down syndrome. She brought up the clear contrast with receiving the diagnosis of autism:

“When we adopted him, we had no qualms about the Down syndrome diagnosis, and I know other parents totally grieve that diagnosis that the child they thought they were going to have, they grieve that, because of the Down syndrome diagnosis. And all of a sudden I totally got it. Like, ‘Oh, this is the grieving. This is the whole thing that you weren’t expecting.’ It changes your whole outlook on how their life is going to be, how your life is going to be. And so, I kind of went into a major kind of depression, anxiety, like period of time in my life, because I took the diagnosis really hard.”

Though it took multiple tries and many years to get a formal diagnosis for her son, Amber still mentioned a sense of shock as well as relief:

“We went in for the meeting, and she said that she was giving him the diagnosis, it still kind of hit me a little bit different, because like I was expecting it, but then even though you’re expecting it it’s still a shock.”

**Benefits of a Dual Diagnosis**

Every parent mentioned ways in which the formal diagnosis of autism helped their family. Many of the immediate benefits were the relief and validation described previously. In
addition, they noted that their child’s autism diagnosis allowed them access to a whole new set of services and sources of support.

“There is a school for kids with autism in the area that because he has the autism diagnosis, he’s at least eligible for consideration. So, that’s something that if we had not gotten the diagnosis wouldn’t have even come up. [...] So, I don’t know if that’s where he is going to go, but it’s an option we have, and it’s an option I’m kind of hopeful we may get for next year that we wouldn’t have had without the diagnosis.” - Helen

“We started pursuing, once he got the official diagnosis, some other outside resources. So, that gave us some extra outside resources, as far as like some working on potty training, which has just been a forever and ever goal. [...] I’m sure him having the second diagnosis has opened some more ideas to therapies from the school perspective.” – Deborah

“We got ABA therapy for – it was a pilot program at the time for kids age two to seven. By the time we actually got the diagnosis, and got on the program, he was four. So, he got 15 hours of ABA every week for three years until he aged out of the program. And that was heavenly. [...] ABA has made a huge difference, and just knowing that like, okay, he can make progress, even though it’s super-slow.” – Samantha

“[The diagnosis] definitely did help. It actually pushed things right along. Once I had the report from Boston, the school – the public school had – they couldn’t even come close to touching on all the recommendations that were written in the report. [...] We were all in agreement that she’s going to need outside placement.” – Michelle

Though Charlene didn’t take advantage of the autism-specific services in her area, she did put a stop to the speech therapy that was not helping her son. Amber, who received her son’s formal diagnosis of autism about six months ago, felt an earlier diagnosis would have been beneficial:

“I think school would have been different, because I think the school would have had more information to help him. For them he was just a child with Down syndrome. [...] I think they just didn’t really understand how to help him fully. [...] They have a good autism program [...] and I think if they [...] had that diagnosis, it would have been approached a little bit differently with his schooling.”

Community and Coping

All parents interviewed had participated at one time or another in a Down syndrome specific group. As their children got older, some continued their involvement, but to a lesser extent and some stopped going altogether. They gave several reasons for their decreased participation, including: not fitting in, activities not being relevant to their child, logistics and scheduling, and rare negative encounters.
Several parents mentioned a sense of jealousy talking to parents of children with Down syndrome or hurt when watching their child with other children from the playgroup. Samantha hosted her own group for three years but eventually stopped:

“By the end of it, I would just almost cringe going to these dinners, and listen to these moms, and all these wonderful things their kids were doing. And I’m like, ‘That’s not my kid.’ I just didn’t – I didn’t fit in, and then I felt almost that I was always super-negative, because I’d be like, ‘Oh, yeah, my kid dumped this stuff on floor again today.’ Look how crappy my life is. [Laughter] I felt like that’s how I was coming across, like everything I had to say was negative, and then these moms were just like, ‘Oh, my kid’s in ballet now, and she’s so cute.’ And I’m like, ‘There’s no way my kid could be like in a gymnastics class, or…’ – you know? And so, I finally stopped doing it, because it was really hard.”

Michelle recalled one picnic in particular:

“We went to a Down syndrome picnic, and I left crying. [...] ‘We don’t even belong here.’ This was before the autism diagnosis. I said, ‘We don’t even – she doesn’t even fit in with this group.’ I said, ‘It just felt like there’s no place for us.’ [...] Even like the children younger than her were further along than she was. And it was a very disheartening feeling. So, instead of like support, feeling happy, it actually made us feel more left out [...]”

Charlene recalled two specific instances that made her feel directly alienated from the Down syndrome community:

“I was at a potty-training seminar, [...] that the Down syndrome group had put on, and they brought in an outside speaker who had a child with Down syndrome [...] And the mother [...] basically laid it out that if your child isn’t this capable then it’s your fault as a parent.”

On a listserv, she encountered the following:

“A lot of the parents with children that are more typical Down syndrome, if you want to call it that, without the autism side, want to be more inclusive in the classroom, [...] One mom basically indicated that if she walked into a special ed room, and saw a child like Phillip in there, she would not want her child with Down syndrome in that same classroom. And it just hit me like a ton of bricks that here we were in our own special needs community, and we were being prejudiced within the special needs community.”

Several parents mentioned difficulties with programming and the needs of their children not being met. For Helen and Isaac, while the Down syndrome community welcomed them, the timing of the program didn’t suit their family:

“It was just far enough away that the timing in getting to the programming is pretty difficult, but also as he’s gotten older, he’s struggled even more with transitions, and so getting out of the house is more difficult.”
Some parents attempted to reach out to the autism community but few were heavily involved. Helen tried reaching out in the beginning:

“I was finding different groups in the area that have programs for kids with autism, and I would call, or I would e-mail to ask for information, and nobody ever got back to me. And there’s a part of me that wonders if it’s because I said that he has Down syndrome and autism.”

While Charlene didn’t get involved in local autism groups, she does mention a special connection with families of children with autism:

“I’ve actually had more experiences with inclusion when I run into parents that have autistic children that are the more classic autism. In fact, when they see Phillip, and me in like the grocery store, I’ve had a mom more than once, two or three times, a mom has come up to me, and said, ‘Oh, I have a child like that, too.’ And I’m like, ‘Oh, with Down syndrome?’ And they’re like, ‘No, autism,’ because they see the autism mannerisms and actions in Phillip before even his Down syndrome physical features. And that’s happened at least twice. And so, they’re more willing to talk, because they see that, and they understand.”

Samantha felt she couldn’t connect as much with the autism only groups:

“I have a couple of nephews with autism. One of them is super-severe, and one of them is pretty high functioning. [...] Maybe that skewed my thinking, but I just kind of feel like we don’t fit. I wish the DS-ASD diagnosis was like a whole different disability, because it’s not either of them. [...] The groups I really love are the dual diagnosis groups, because they get it, and I just don’t feel like I fit in anymore in either of just the single diagnosis groups.”

Eventually, all parents discovered the Down Syndrome-Autism Connection online Facebook group, which is how they connected to this study. Some parents have gone on to form local networks of their own. What stood out about these DS-ASD groups was the ability of other parents to fully celebrate each child’s small victories. Helen, and several others similarly, illustrated this with the following:

“There is that sense of these are the folks who will understand both the difficulties, and the joys, and why something that in just the Down syndrome group might actually depress other people can be a joyful thing in the Down syndrome/autism group, instead of being like, ‘Oh, your kid is this old, and they’re not potty trained, yet?’ Or, ‘Your kid is this old, and they’re still throwing tantrums?’ [...] Within the Down syndrome/autism group, you can kind of say, ‘Hey, my kid who struggles just did this small, but good thing,’ and they can rejoice in that, instead of not rejoicing.”

When asked what advice they would offer to a parent of a child newly diagnosed with autism, responses varied. However, nearly every parent said something similar to Samantha’s statement:

“Remember that the day after your diagnosis, you have the same kid that you had the day before the diagnosis. And I even told that to myself at the time, because I knew that. I knew that nothing had changed.”
Every parent at some point during the interview framed the autism diagnosis as a way to better understand and help their child. Amber, who went years without a diagnosis, responded:

“\(\text{I would just tell parents not to give up if they suspect that something’s different that there’s something else there, to not give up, and not to just think their doctors know everything, and then move on.}\)"

**Challenges and Joys**

When asked about their biggest challenges, three of the parents brought up struggles with their child’s behavior. Helen expressed this struggle as a sense of uncertainty in how to respond to her son’s behavior:

“\(\text{I can’t figure out how to get him on the bus, and I don’t know how to do this. I don’t know how to support him. I don’t know what the right thing is to do. Is the right thing to force him to get on the bus, and go to school, because he needs to be in school [\ldots]? Or do I need to let him stay home, and calm down? And I never know if what I’m doing is the right thing when he’s having the challenges like that.}\)"

Michelle also mentioned uncertainty in the context of a lack of information:

“\(\text{It doesn’t seem like there’s a lot of information out there. There’s information about Down’s. There’s information about autism. But there isn’t a lot of information about both diagnoses out there. So, sometimes you feel like you’re left to figure it out on your own. And that’s been really hard.}\)”

**Five of the six parents mentioned to feeling physically isolated or unable to leave their house:**

“\(\text{If I don’t have reinforcements with me, like my husband, or my older daughter with me, I can’t always do things. So, sometimes I feel kind of closed off, and a little bit – I won’t say a prisoner in my own house, but sometimes it does feel that way, like I can’t get out to do things, because Jennifer is going to have a hard time. I guess that’s really huge.}\)” – Michelle

“\(\text{We haven’t taken a family vacation since he was probably a year old. [\ldots] You can’t just do normal family things, like go to the zoo. When my husband comes with us we can, and he’s a lot better with him, but just I can’t manage the two kids by myself. And so, I think that’s been the hardest thing is just – and then our house is like a fortress. We have locks everywhere. You can’t just go get a drink, or go the bathroom without undoing locks.}\)” – Samantha

“\(\text{I could sit home all day with him, and not really get him out, and he wouldn’t care, but I want him to be seen as part of the community, so I do push that. [\ldots] He doesn’t care. He’s happy with his iPad sitting here by himself for the most part, but at the same time, when I do get him out there, he’s happy to go swimming at the local pool, or playing on the playground, [\ldots] but I guess the hard part is making sure I don’t isolate him, or myself.}\)” – Deborah
Charlene and Deborah mentioned their children’s inability to talk as their biggest concern:

“If I had one wish in the world it would be that he could talk.” – Charlene

“What’s typical of any parent – [I’m] concerned about his future, and what’s going to happen to him, and who’s going to take care of him. […] It’s hard for me to trust others, because he is nonverbal, and he can’t tell me if there’s something else that happens to him.” – Deborah

When asked about the joys, every mother talked about her child’s personality:

“He’s just a really warm, and affectionate kid. And so, it’s very rewarding to get hugs, and kisses, and he is semi-verbal, and so he can say, ‘I love you, mommy.’ And little things like last night my husband got him a snack, and Isaac said, ‘Thanks, dad, you’re the best.’ And he doesn’t usually say, ‘You’re the best.’” – Helen

“He’s just a fun kid. He’s a practical joker. When you’re in his world, when you’re playing with him, and in his world, he’s a pretty smart kid, and he’s a lot of fun.” – Charlene, about Phillip

“He’s fun. He can be silly. He’s got the best smile, and he’s in his own way very loving, and just a happy go lucky guy, as long as things are going his way. [Laughter] For the most part he’s actually pretty easy compared to some other kids […]” – Deborah, about Chris

“He’s really funny. He will like – if he wants to like do a little funny dance for you, he will like sit you down. [Laughter] And you can’t be doing anything else, and he’ll do his little dance, or his little whatever. And if you try to like sing along, or wiggle with him he’ll like put out his hands, like to tell you to stop. Nope. [Laughter] Like you’re not dancing, I am. Focus over here. I’m center of attention. And then he’ll come and take your hands, and make you clap for him. [Laughter] But he is really funny, which is interesting for a kid who doesn’t talk […]” – Samantha, about Julian

“He’s taught me strength, and good will, and he doesn’t judge. He doesn’t judge people. He doesn’t give up. He just – he wants to be sometimes social, but he just doesn’t understand how to be. You know, I think he just doesn’t have the skills, even though we’ve taught him the skills that he just – he works so hard for things, but he doesn’t – and he doesn’t give up.” – Amber about Dylan

“She’s got a great smile, and a laugh, and it just makes you just slow down a little bit, and just look at the small things. Like we appreciate small victories where you wouldn’t even give it a second thought to a quote/unquote “typical child.” If she has a little victory, we’re on the phone. I’m calling my mother, and we’re all excited, and so, I think we are looking at the more simple things, and appreciating every little step, appreciating who she is, and how she’s developing at her own pace.” – Michelle, about Jennifer
Discussion

In this study, we interviewed six mothers of children with a dual diagnosis of Down syndrome and Autism Spectrum Disorder in order to better understand their experience raising a child with this dual diagnosis. Patterns revealed in the responses illuminate several important psychosocial considerations for healthcare providers and genetic counselors, such as the value of an accurate diagnosis and appropriate support services for parent and child.

Awareness of & Access to Diagnosis

The majority of parents interviewed were the first to raise the possibility of autism to explain their child’s behavior and they were able to access information about the possibility of a dual diagnosis readily through an Internet search. However, parents specifically searched for this information only after they began to suspect their child was developing differently than their peers with Down syndrome. Only Helen reported receiving outdated information from a healthcare provider about the existence of DS-ASD.

Most parents felt their child’s pediatrician, primary care doctor, or geneticist took them seriously when they expressed their concerns. This was in contrast to our suspicion that initial suggestions of autism may be brushed off by healthcare providers based on prior anecdotal evidence from the DS-ASD community (Froehlke & Zaborek, 2013; Medlen, 1999). Zuckerman, Lindly, & Sinche (2015) investigated the phenomenon of passive provider response to parents concerned about their child’s development. The authors considered passive providers to be those who made statements such as “the child might grow out of it,” “it was too early to tell if anything
was wrong,” and “nothing was wrong/child’s behavior was normal.” This study found those with co-occurring ASD and intellectual disability (ID) and/or developmental delay (DD) achieved diagnosis sooner than those with solely ASD. Though the nature of the ID/DD was not given, the authors suggested the closer connection of those with ID/DD to their providers encouraged more expedient referral to specialists who could make a diagnosis. Reasons for a passive response from primary care providers included a lack of access to screening, referral, or diagnostic resources. Though our participants generally felt their providers addressed their concerns appropriately, they bemoaned the multiple visits to specialists and long wait periods for evaluation. It appears this is not a problem specific to those in the DS-ASD community but may be a part of a systemic lack of resources for children with ASD (Miller, 2017).

Half of the parents mentioned the social nature of their child interfering with a timely diagnosis. Kent, Evans, Paul and Sharp (1998), suggested the stereotype of individuals with Down syndrome being good-natured, sociable, and friendly could bias professionals and prevent a full evaluation of social skills. In January 2017, another study found that children with DS-ASD tended to have relatively milder social difficulties than others with ASD, although they demonstrated similar patterns of communication and repetitive behaviors to those typically described in autism (Warner, Howlin, Salomone, Moss & Charman).

**Benefits of the Formal Dual Diagnosis**

A diagnostic odyssey is the period of time characterized by uncertainty, frustration, and isolation from appropriate resources (Carmichael, Tsipis, Windmueller, Mandel, & Estrella, 2015). The participants in this study noted the years-long build-up to an ASD diagnosis was so different from the often surprising, Down syndrome diagnosis. Though feelings were generally mixed, the years they’d spent getting to know their child and their behaviors made the autism
diagnosis itself easier to bear. The formal diagnosis provided a framework for understanding and helping their child as well as a sense of validation for what they had suspected was different for so long.

Once a diagnosis was made, different services and schooling opportunities became available to the participants' children. The diagnosis was incorporated into each child's Individual Education Plans (IEPs) but the way in which services actually changed varied from one child to another. For some, the effect was immediate and profound, such as the school change for Jennifer. For others, it opened opportunities that went unutilized, either because the district was currently meeting their child's needs or, as in Charlene's case, the autism-specific school would not have been appropriate for her child.

**Community & Support**

One definition of community is that of community theory in sociology. It defines community as the amalgamation of four elements (1) membership, as in a feeling of belonging or sharing a sense of personal relatedness; (2) influence, in that the individual makes a difference in the group and the group matters to its members; (3) integration and fulfillment of needs, in which the members' needs are met by the resources brought to them by membership in their group; and (4) shared emotional connection, the belief that members have shared history, places, time, or experience (McMillan and Chavis, 1986). When asked about community and belonging, the responses in this study can be understood in the context of this theory.

All parents had attempted to participate in DS-only community activities such as parent support groups or child playgroups. Many felt they didn't belong, that they couldn't relate to the other parents and that the other parents had a difficult time relating to them and their specific
challenges and successes. Several parents mentioned feelings of longing or jealousy when talking to parents of typically developing children with Down syndrome. Many brought up watching their child alongside their peers with Down syndrome and their feelings of sadness as a parent, wondering what was happening with their child. A few parents mentioned their experiences seemed to “bring the mood down” at gatherings that were supposed to be strengthening, inspiring, or uplifting. Their need to celebrate small things seemed to make others uncomfortable as they lacked a full appreciation of what those victories meant. Parents felt an overall lack of understanding or in rare cases prejudice when it came to their children’s behavior. Few parents had any involvement in the ASD-only community. Because Down syndrome is diagnosed at birth, they may be linked to the DS-only community initially. As the autism becomes more apparent, they would naturally seek out DS-ASD groups rather than ASD-only groups. A few of the mothers did name key individuals in the DS- or ASD-only communities with whom they made emotional connections. Parents who had negative experiences were quick to say there was no animosity between them or the communities of the singly diagnosed, but that these communities were not a good fit for them and their child. Many study participants mentioned they still attend certain gatherings or events, but less frequently than in the past.

The importance of time, place, and experiences as a group in the development of community means many of these mothers participated less and less as leaving the house became increasingly difficult. The DS-ASD community, a primarily online presence through the Facebook group, the Down Syndrome-Autism Connection, is within ready physical access of families. The joys are celebrated by people who understand them and the challenges facing families are normalized within the group. Some parents took advice from other parents while others appreciated the emotional connection to others sharing similar life experiences.
Challenging Behaviors & Lack of Guidance

Child behavior may be the most important determinant in the level of parental stress and care demands (Cnic, Friedrich, & Greenberg, 1983; Floyd & Gallagher, 1997; Hastings & Beck, 2004). Floyd and Gallagher compared parent groups of school-age children with disabilities, either intellectual, physical, or behavioral. They found the mere presence of behavioral problems was more relevant to parent stress levels than the particular type of disability. In our study, many parents commented on their child’s difficult behaviors when asked about challenges. Their child’s behavior had far-reaching effects in both the family dynamic and emotional well-being of the parent and child. Physical isolation was one of the main points brought up by five of the six mothers as the child’s behavior often made it difficult to leave the house. Additionally, many mothers felt they were unable to control their child if they were by themselves. Interventions benefit parental well-being when they successfully improve the skills or reduce the problem behaviors of children with disabilities (Hastings & Beck, 2004).

At least two parents in our study directly commented on a lack of information and guidance in regards to handling their child’s behavior. Parent perception of their ability to parent, also referred to as self-efficacy, is reduced among parents of children with greater levels of behavior problems, though the causal direction is less clear (Paczkowski & Baker, 2007). Several parents communicated the importance of doing what was right by their family, whether it be the amount of therapy their child received or the behaviors on which they compromised (such as riding the bus to school or eating more than just a select few meals). They seemed to wish there were better solutions or more information about behaviors to anticipate, but ultimately stood fast in their choices on behalf of their family.
In 1983, Wikler, Wasow, and Hatfield investigated the evolution of strength as a part of the experience of mothers of children with disabilities. They found 75% of their 27 respondents felt being a parent of a child with a disability had strengthened them. Amber expressed a similar sentiment when asked about the joys of raising her son, stating he “taught [her] strength.” Additionally, the authors of the aforementioned study recommended practitioners adopting a strengths perspective ask parents about their child’s unique traits and how they have benefitted the family. When asked about joy, every parent in our study immediately made the connection between their child’s unique personality and the happiness created by getting to know them. They acknowledged the challenges, but as echoed by the advice these mothers gave to other parents, their child’s personality did not change the day after diagnosis. The diagnosis of autism was simply a way to understand and help them.
Conclusions

We interviewed parents of children with DS-ASD about their experience with raising their child and the diagnostic process. Contrary to expectations, most participants had ready access to information about the existence of the dual diagnosis and their medical providers took their concerns seriously. This may indicate a welcome sign of increasing awareness of the dual diagnosis in the medical community. Though parents could access information about the dual diagnosis, several still lamented a lack of information in regards to symptomology, managing difficult behaviors, and planning for the near and distant future.

The formal diagnosis brought relief and validation for many parents after years of built up frustration, isolation, and suspicion that something was different about their child. These feelings are common to those who undergo a diagnostic odyssey for themselves or their child. The diagnosis opened doors to services though the overall utilization of said therapies or school changes varied. In general, parents integrated the diagnosis into their lives as a way to better understand and help their child reach their full potential.

All parents had at least attempted to participate in DS-only parent and child groups. The participation in those groups faded over the years for a variety of reasons. Many parents brought up logistic issues as far as the distance or timing of the groups. Their child’s behavior made getting to the various events or gatherings time-consuming and draining. The group activities themselves often felt irrelevant to their child’s needs or outside their capabilities or interests. Many parents brought up a sense of jealousy or discomfort around parents of children with DS-only. While small victories felt like a success to their family, relaying this joy to the DS-only
community seemed uncomfortable or awkwardly received. Few parents had much if any involvement in the ASD-only community. Only rarely were parents met with outright unwelcome behavior or comments from either DS- or ASD-only groups. The online platform of the DS-ASD community provides readily available support to parents who feel “trapped” in their own home.

The insights gained by this investigation are of particular use to genetic counselors who are trained in the psychosocial support of children and families with a genetic condition such as Down syndrome and/or autism. This study emphasizes the importance of specific, relevant information for parents as well as that of appropriate supports and expectations. Parents felt most supported when their child’s successes were celebrated with understanding and enthusiasm, and their child’s struggles were not met with uncomfortable questions or judgment.

Limitations & Future Directions

We interviewed only six parents for this study so our participants may not represent the views of all parents of children with DS-ASD. In addition, the potential for selection bias exists as all participants were recruited through the Down Syndrome Autism Connection. Because of this, we expected we would not hear from parents who were unable to find support resources. Half of our participants were linked to the same geographic medical community, which could have influenced the ease of diagnosis and access to support. Additionally, this study interviewed only mothers; the experiences of fathers may differ. Further investigations could include a survey-based quantification of parent experience. Another alternative could involve replicating this study in cohorts of other genetic syndromes. Fragile X syndrome is one example of a genetic syndrome in which the increased prevalence of autism is better known within the community (National Fragile X Foundation). Alternatively, individuals with William’s syndrome are thought
to have endearing, overly friendly personalities with excellent expressive language abilities (William Syndrome Association). Investigation into the parent experience with either of these conditions co-occurring with ASD could yield valuable comparisons.
References


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Appendix A: The Down Syndrome-Autism Connection Permission Letter

Dear Elizabeth,

As current President of the Board of Directors for the Down Syndrome-Autism Connection, I am pleased to collaborate with you on your thesis project involving data accessed from survey and interview based responses from our members. All members participating do so willingly and on a volunteer basis.

Again, I am happy to collaborate with you on this project.

Sincerely,

Jeanne Doherty
President- The Down Syndrome-Autism Connection
Appendix B: The Down-Syndrome Autism Connection Recruitment Language

Dear Friends,

I'm writing to tell you about a research study looking into the experiences of parents, such as yourself, who are raising a child with a diagnosis of both Down syndrome and autism (DS-ASD). This investigation was proposed by a young woman named Elizabeth Kellom. Elizabeth is a genetic counseling student in a graduate program at Brandeis University and will be completing this research in order to earn her Master's degree. She has volunteered with our parent group on several occasions to provide childcare for children with Down syndrome and autism.

The goal of her study is to better understand the experiences of parents raising children with DS-ASD. If you would like to participate, you would be asked to fill out a short survey about your family and child. Afterwards, at a time convenient to you, Elizabeth will interview you by phone to answer some more in-depth questions. This should take between 30 - 45 minutes. All information shared will be completely confidential.

As thanks for participating, you would be given a $25 Amazon gift card upon completion of the interview.

If you would like to participate or have any questions, please contact Elizabeth at (781)652-1534 or by email at ekellom@brandeis.edu.

We can all help by sharing our stories. We can potentially help future clinicians by giving a voice to all those living with DS-ASD and those who love them.

Thank you.

Warm Regards,

Jeanne Doherty
Board President
The Down Syndrome-Autism Connection
Appendix C: Informed Consent Document

Information Sheet

We ask that you read the information sheet before the interview. You will be asked if you understood all the information in the information sheet, whether you have any questions, and if you voluntarily agree to participate in this study, before we begin the interview.

Purpose of the Study: The aim of this study is to learn more about the experiences of parents, such as yourself, who are raising a child with Down syndrome and autism (DS-ASD).

Study Procedures: You will be asked to fill out a short survey about your family and child. Afterwards, you will be asked to participate in a recorded phone interview, lasting between 30 - 45 minutes. Mainly, you will be asked questions about your experience finding a diagnosis for your child, your support systems, and the challenges and joys of raising a child with DS-ASD.

Benefits: There is no direct benefit to you. However, information is power and we hope the testimonies provided by yourself and other parents will empower future parents and educate clinicians and others who serve you and your family.

Risks: Your participation in this study presents no more than minimal risk. It is possible that talking about your experiences could bring up some distressing emotions or memories. If you become uncomfortable, you can skip a question or stop the interview entirely.

Privacy and Confidentiality: All records containing identifying information, such as names, email address and telephone numbers will be kept strictly confidential during the study. All study related documents and materials (including interview transcripts and audio files) will be kept in a secure location accessible only to the Principal investigator and student researcher. Transcripts, interview notes, and audio files will be labeled with a coded ID number, which will be assigned to you upon enrollment into the study. If you are quoted or referred to in any written or oral reports of this study, you will be given an alternate name. You will never be referred to by your name or any other identifying information.

Cost and Compensation: There will be no cost to you to participate in the study other than the time it takes to conduct the survey and interview. As a token of our appreciation, we will send you a $25 Amazon e-gift card upon completion of the interview.

Contact Information: If you have any questions about the study, please contact the student researcher, Elizabeth Kellom at ekellom@brandeis.edu. If you are having difficulty reaching her, or if you encounter problems related to the study, you may also contact the Principal Investigator for this project, Judith Tsipis, at tsipis@brandeis.edu. 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.
Appendix D: Demographic Survey

Thank you for your willingness to participate in this study. Your input is valuable!

Prior to our interview, please look over the informational sheet provided to you in this email. Then answer the following questions about your family and your child with Down Syndrome and Autism. Feel free to skip any question you do not wish to answer. When you have finished the survey, please email it back to me and we can set up a time and date for a telephone interview.

1. Current Age of child: ______
2. Gender of child: ______
3. Is your child adopted? ______
4. Age at Down Syndrome Diagnosis
   1. Prenatally
   2. At birth
   3. Infancy
   4. After 1 year
5. Age at Autism Diagnosis: ______
6. Who first suggested your child may have autism?
   1. pediatrician/primary doctor
   2. geneticist
   3. family member
   4. friend
   5. childcare provider
   6. Myself and/or the other parent
   7. Other: __________
7. Who formally diagnosed your child with autism?
   1. pediatrician/primary doctor
   2. geneticist
   3. psychologist
   4. autism specialist
   5. other: ________
8. How does your child communicate? Check all that apply.
   1. Sign Language
   2. Noises
   3. Gestures
   4. Some Spoken Words
   5. Full Spoken Sentences
   6. Communication Device (iPad, tablet, etc.)
   7. Other: _______________
   [total children]

10. Does your child with Down syndrome have, now or previously, any of the following major medical problems? Check all that apply.
   i. Heart Defect
   ii. Epilepsy
   iii. Leukemia
   iv. Other: ________________
   v. None of the Above

Thank you very much!
Appendix E: Interview Guide

1. Tell me a little about when you found out [---] had Down Syndrome. What were some of your thoughts at that time?

2. Prior to receiving the second diagnosis, did you know that it was possible for children with Down Syndrome to also have autism? Were you able to get information about it through your doctor or the internet? What groups, resources?

3. I’d like to hear about the period of time when the suspicion of autism first arose - how did you react to whoever first raised the possibility? If you raised the concern first, how did people react to your concern?

4. Tell me about your experience after receiving the diagnosis of autism. How was it similar or different from receiving the Down Syndrome diagnosis?

5. Did the second diagnosis of autism open new doors for services? What were they?

6. Did you participate in any Down Syndrome only parent groups? How did they work for you? Helpful, harmful, irrelevant - in what way? After the autism diagnosis, did you seek out any autism only groups?

7. Do you feel a sense of community with either group, or do you find your situation too removed from both?

8. What’s been the most difficult thing about raising a child with DS-ASD?

9. What’s been your most joyous or rewarding moment while raising [---]?

10. If you could tell a parent in your shoes one thing, what would it be?