The Medical Care of Children Born with Ambiguous Genitalia from the Parent Perspective

Master’s Thesis

Presented to

The Faculty of the Graduate School of Arts and Sciences
Brandeis University
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David Rintell, Ed.D., Advisor

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by

Nancy Herrig

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ABSTRACT

The Medical Care of Children Born with Ambiguous Genitalia from the Parent Perspective

A thesis presented to the Genetic Counseling Program

Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Nancy Herrig

Guidelines for the clinical management of individuals born with disorders of sexual development (DSD) were established with the publication of the 2006 DSD Consensus Statement. The purpose of this study was to explore the experiences of parents of children born with ambiguous genitalia due to DSD and to examine their perceptions of the medical care they received in relation to the DSD Consensus Statement. We conducted semi-structured telephone interviews with eight parents whose children were born with ambiguous genitalia who were between the ages of three months and ten years. Five main themes emerged from the interviews: 1) communication with healthcare professionals (HCPs), 2) experience and expertise of HCPs, 3) coordination of care, 4) sources of psychosocial support, and 5) role of the Internet for information. While parents reported generally positive experiences with regard to the experience and expertise of their HCPs, they reported an almost equal number of negative as positive experiences regarding communication. Only two parents reported receiving their medical care through an established specialized multidisciplinary team. Most parents assembled their own medical team with variable success in coordinating the care among the different
specialists. Psychosocial care provided by mental health staff with expertise in DSD was available only to parents who received their care through established multidisciplinary teams. The Internet played an important role for most parents by providing peer support, guidance in decision-making, and education. Our findings suggest that increased availability of established multidisciplinary teams and better communication between parent and HCPs could improve care and relieve stress placed on families.

Key Words: ambiguous genitalia, consensus statement, disorders of sexual development, DSD, gender assignment, genital surgery, healthcare professional, Internet, multidisciplinary team, psychosocial, qualitative, support groups
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INTRODUCTION

The clinical management of children born with ambiguous genitalia is often a complex challenge for healthcare professionals (HCPs). For parents, the birth of a child with ambiguous genitalia is a stressful experience, often the gender of the child is uncertain and parents may be concerned for the future physical, psychological and sexual health of their child. Difficult medical decisions must often be made by parents such as selection of sex of rearing, genital surgery, or hormone therapy.

Ambiguous genitalia is a rare condition that occurs between 1 and 2 per every 1000 live births (Blackless et al., 2000) and falls under the umbrella diagnosis of disorders of sexual development (DSD). DSD refers to a wide range of “congenital conditions in which development of chromosomal, gonadal, or anatomical sex is atypical” (Lee, Houk, Ahmed, & Hughes, 2006). Until recently, there were no practical clinical guidelines to provide HCPs with standardized diagnostic and management approaches in the care of patients with DSD or guidance as to how to effectively support parents and patients through the informed consent decision-making process. At a 2005 meeting in Chicago, a group of fifty experts reached a consensus on the optimal care for children with DSD and published a Consensus Statement on the Medical Management of DSD (Lee et al., 2006). Since then progress has been made in implementing these clinical guidelines (Pasterski, Prentice, & Hughes, 2010).
While much research has been conducted on the clinical management of children born with DSD, previous research with parents and caregivers focused mainly on the psychological impact of having a child born with ambiguous genitalia and was quite limited. Duguid et al. (2007) found that some parents experienced clinical levels of stress (19%) and reduced coping ability (13%). Crissman et al. (2011) reported on a qualitative study of health-related quality of life as reported by parents who serve as proxy informants for their children with DSD who had undergone genital surgery. The study found that parents experienced particular stress in two areas: Uncertainties regarding diagnosis and optimal management and conflicts between maintaining privacy versus disclosing the condition to access social support. Parents expressed concerns about their child’s future physical, social, and sexual development.

Sanders, Carter, and Goodacre (2011) identified three elements that influence parents search for harmony regarding their child’s genital ambiguity and genital surgery: shock, protection, and anxiety. Fedele et al. (2010) investigated the impact on caregivers of having a child with a DSD, as well as how the decision of whether or not to have early genital surgery on the child impacted the mental health and parenting characteristics of primary caregivers. Their study found that some parents experienced higher levels of stress and maladaptive parenting characteristics when they chose not to proceed with genital surgery at all or when they chose to proceed with genital surgery very early in life.

Understanding the psychological impact of having a child with ambiguous genitalia is important for genetic counselors and other HCPs as they care for families.
The clinical management of families can also affect on how they cope with the diagnosis and treatment. Five general concepts of care were identified in the Consensus Statement (Lee et al., 2006) for optimum clinical management:

1) Gender assignment must be avoided before expert evaluation in newborns.
2) Evaluation and long-term management must be performed at a center with an experienced multidisciplinary team.
3) All individuals should receive a gender assignment.
4) Open communication with patients and families is essential, and participation in decision-making is encouraged.
5) Patient and family concerns should be respected and addressed in strict confidence.

The goal of this study is to assess the parents' perceptions of the medical care their children received in relation to the Consensus Statement (Lee et al., 2006) on the management of DSD. Increased understanding of parent’s experiences can help genetic counselors and other HCPs provide better care and support for these families. It is our hope that the experiences shared by the parents in this study will also be helpful to other parents of children affected by DSD.
METHODS

This study was approved by Brandeis University's Institutional Review Board.

Study Population

We recruited participants for this study through national support organizations and online support groups including the AIS-DSD Support group for Women and Families, the Magic Foundation, and the Congenital Adrenal Hyperplasia Education and Support Network. We distributed recruitment notices, which were specific to the organization (Appendices A & B), to administrators of the different support organizations so that they could distribute them to their membership. We encouraged interested individuals to contact us directly via email. We then conducted a brief telephone interview (Appendix C) to assess eligibility. Inclusion criteria included: Individuals must be a parent of a child born with ambiguous genitalia, must be 18 years of age or older, must be fluent in English, and the child born with ambiguous genitalia must be between the age of 3 months and 10 years as of January 1, 2012.

Eight respondents who expressed interest were eligible for study participation and we scheduled interviews at their convenience. We obtained informed consent (Appendix D) from each participant prior to the telephone interviews. We offered the participants a $25 gift card as a gesture of appreciation for their time.

Demographics

A total of seven interviews were completed of which one was held with both the
mother and father and six with the mother only. Of the seven interviews conducted for the study, six were of parents residing in the United States and one was of parents residing outside the United States. The children of the participants ranged in age from four months to 9.9 years. Three of the children were adopted and for two of these children the ages are approximate. At the time of adoption, the three children ranged in age from 1.5 to 3.3 years. Table 1 shows further information about the participants and their children. Table 2 summarizes the characteristics of the parents interviewed. The characteristics of the children are summarized in Table 3. Table 4 summarizes the DSD diagnoses of the children of the participants.

**Interviews**

We developed a semi-structured interview guide with open-ended questions (Appendix E) that were based on the clinical management guidelines detailed in the DSD Consensus Statement (Lee et al., 2006). We structured the interview to follow the parents’ experiences in chronological order to explore the different aspects of their child’s medical care such as birth, testing, diagnosis, gender selection, gathering information, interactions with HCPs, consideration of surgery, and psychosocial support. We also asked parents what had been the most and least helpful aspects of their experience and what would have made this process better. The interview guide was used to conduct in-depth telephone interviews with eligible parents. Though we used the same interview guide for each participant for consistency, some questions were adapted based on the details of each parent’s experiences and the completeness of his or her answers. The overall content remained consistent between interviews. The mean interview
duration was 71 minutes (range 47 to 105 minutes).

**Data Collection & Analysis**

The interviews were audio recorded and transcribed by a confidential transcriptionist. All records containing identifying information collected from the study participants were kept confidential. Transcriptions of the interviews were imported into Atlas.ti (version 6.2), a software package for quantitative analysis of textual data. The data was analyzed using grounded theory (Glaser & Strauss, 1999). The transcripts were analyzed line by line and open coding was used to identify statements made by parents that were related to feelings, thoughts or experiences related to their child’s medical care. The codes were grouped into main content areas and the most frequently occurring codes were further grouped into themes and sub-themes that emerged from the data (Figure 1).
Figure 1. Summary of main content areas, themes and sub-themes
<table>
<thead>
<tr>
<th>Interview</th>
<th>Participant</th>
<th>Age of child at interview (years)</th>
<th>If adopted, age of child at adoption (years)</th>
<th>Age of child at diagnosis (years)</th>
<th>Gender of rearing</th>
<th>DSD diagnosis</th>
<th>Genital surgery?</th>
</tr>
</thead>
<tbody>
<tr>
<td>P1</td>
<td>mother &amp; father</td>
<td>3.5</td>
<td>1.5</td>
<td>2.5 years</td>
<td>M</td>
<td>5α-RD-2</td>
<td>yes</td>
</tr>
<tr>
<td>P2</td>
<td>mother</td>
<td>10 (approx.)</td>
<td>3.5 (approx.)</td>
<td>4 years (approx.)</td>
<td>F</td>
<td>OT-DSD</td>
<td>yes</td>
</tr>
<tr>
<td>P3</td>
<td>mother</td>
<td>3 (approx.)</td>
<td>1.5 (approx.)</td>
<td>2 years (approx.)</td>
<td>M</td>
<td>5α-RD-2</td>
<td>no</td>
</tr>
<tr>
<td>P4</td>
<td>mother</td>
<td>1.5</td>
<td>-</td>
<td>1 year</td>
<td>F</td>
<td>5α-RD-2</td>
<td>no</td>
</tr>
<tr>
<td>P5</td>
<td>mother</td>
<td>8</td>
<td>-</td>
<td>first week of life</td>
<td>F</td>
<td>CAH</td>
<td>yes</td>
</tr>
<tr>
<td>P6</td>
<td>mother</td>
<td>3</td>
<td>-</td>
<td>first week of life</td>
<td>F</td>
<td>CAH</td>
<td>yes</td>
</tr>
<tr>
<td>P7</td>
<td>mother</td>
<td>0.5</td>
<td>-</td>
<td>first week of life</td>
<td>F</td>
<td>CAH</td>
<td>no</td>
</tr>
</tbody>
</table>

Table 1. Demographics of participants (N=8) and their children (N=7). 5α-RD-2, 5α-reductase-2 deficiency; OT-DSD, ovotesticular DSD; CAH, congenital adrenal hyperplasia

<table>
<thead>
<tr>
<th>Parenting Role</th>
<th>Number</th>
<th>Percentage</th>
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<tbody>
<tr>
<td>Mother</td>
<td>7</td>
<td>87.5</td>
</tr>
<tr>
<td>Father</td>
<td>1</td>
<td>12.5</td>
</tr>
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</table>

<table>
<thead>
<tr>
<th>Age of Parents at Interview (years)</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>30 - 39</td>
<td>4</td>
<td>50</td>
</tr>
<tr>
<td>40 - 49</td>
<td>3</td>
<td>37.5</td>
</tr>
<tr>
<td>50 - 59</td>
<td>1</td>
<td>12.5</td>
</tr>
<tr>
<td>Adoptive parents</td>
<td>4</td>
<td>50</td>
</tr>
<tr>
<td>Birth parents</td>
<td>4</td>
<td>50</td>
</tr>
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</table>

Table 2. Summary of demographics of parent participants (N=8)
<table>
<thead>
<tr>
<th>Gender of rearing</th>
<th>Number</th>
<th>Percentage</th>
</tr>
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<tbody>
<tr>
<td>Boys</td>
<td>2</td>
<td>28.6</td>
</tr>
<tr>
<td>Girls</td>
<td>5</td>
<td>71.4</td>
</tr>
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</table>

<table>
<thead>
<tr>
<th>Age of children at interview (years)</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 - 2.9</td>
<td>4</td>
<td>57.1</td>
</tr>
<tr>
<td>3 - 6.9</td>
<td>1</td>
<td>14.3</td>
</tr>
<tr>
<td>7 - 10</td>
<td>2</td>
<td>28.6</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Child by adoption</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>3</td>
<td>42.9</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Child by birth</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>4</td>
<td>57.1</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Time until diagnosis (from birth or adoption)</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>First week of life</td>
<td>3</td>
<td>42.9</td>
</tr>
<tr>
<td>6 months</td>
<td>2</td>
<td>28.6</td>
</tr>
<tr>
<td>1 year</td>
<td>2</td>
<td>28.6</td>
</tr>
<tr>
<td>&gt; 1 year</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Children who have had genital surgery</th>
<th>Number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>4</td>
<td>57.1</td>
</tr>
</tbody>
</table>

Table 3. Summary of demographics of children of participants (N=7)

<table>
<thead>
<tr>
<th>DSD diagnosis</th>
<th>N (%)</th>
<th>Gender of Rearing</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Boy</td>
</tr>
<tr>
<td>46, XX DSD</td>
<td>4 (57.1)</td>
<td>0 (0)</td>
</tr>
<tr>
<td><em>Congenital adrenal hyperplasia</em></td>
<td>3 (42.9)</td>
<td>0</td>
</tr>
<tr>
<td><em>Ovotesticular DSD</em></td>
<td>1 (14.3)</td>
<td>0</td>
</tr>
<tr>
<td>46, XY DSD</td>
<td>3 (42.9)</td>
<td>2 (66.7)</td>
</tr>
<tr>
<td><em>5α-reductase-2 deficiency</em></td>
<td>3 (42.9)</td>
<td>2</td>
</tr>
</tbody>
</table>

Table 4. Summary of DSD diagnoses of children of participants (N=7)
RESULTS

The interview discussions focused on five main content areas: (1) communication with HCPs, (2) experience and expertise of HCPs, (3) coordination of care, (4) sources of psychosocial support, and (5) role of the Internet for information. Using codes to categorize the participants' responses, we identified themes and sub-themes representing the participants' feelings, thoughts, and experiences. Figure 1 summarizes the content areas and the most frequently occurring themes and sub-themes expressed by participants.

Pediatric endocrinologists and urologists are the primary HCPs who provide medical care for children who are born with DSD. Unless otherwise specified, parents refer to specialists in these areas when describing a HCP.

1. Communication with HCPs

Two major themes emerge from the discussion with parents about communication with their child's HCPs.

Theme: Interactions with HCPs

Each parent individually reported an almost equal number of positive versus negative interactions with HCPs regarding communication.

Positive interactions with HCPs. When parents recounted positive interactions, HCPs were described with the following attributes: kind, reassuring, empathetic, accessible, caring and supportive. Of the positive communication attributes, listening and
sensitivity emerged as the ones most important to parents:

“[The doctors have] all been sensitive, and if not knowledgeable, very willing to learn, especially like with her pediatrician. I send him stuff all the time. He really wants to learn. [She] is pretty rare in this area, but they’ve all been willing to listen to me and what I’ve learned.” (P4)

Another parent related the following view held by their endocrinologist:

“I always tell my doctors and everybody that I work with that you always listen to the parents. The parents know their children, and if they question something, always go back and look again.” (P5)

Some parents had to find new HCPs for a variety of reasons such as family relocation, relocation of the HCP or dissatisfaction with the current HCP. Many parents described the challenges they encountered when finding a new HCP. One parent described the following positive experience at a new clinic:

“I started to cry the first time I went to an appointment with them and all three of them walked in the room. And right away all of them gave me their cell phone numbers... they were there for me, let me tell you.” (P6)

**Negative interactions with HCPs.** When describing negative interactions with HCPs, parents frequently used words such as insensitive, opinionated, unresponsive, uninformed, unkind and directive. A parent related the following negative interaction during their first visit to a new urologist:

“The new guy we went to see last time... was the typical surgeon personality. Cold, short even, ‘What are you doing here? You get an ultrasound every time you go. I want you to go back down for an ultrasound now. Unfortunately, I’m not going to be able to see you but a very limited time now, because you should have known to do that.’ You know, talked down to us, did not even look [my child] in the eye. We won’t be going back to him.” (P2)

Communication with parents is especially important shortly after the birth of a child born with ambiguous genitalia when the gender of the child is not known. The
choice of language used by HCPs can deeply affect parents as they are coping with the uncertainty of their child's health. One mother shared this experience with a doctor and nurse that occurred shortly after the birth of her child.

"I swear one of them referred to her as It. That could have been in my postpartum brain, but that crushed me." (P4)

Clarity and timeliness of information from HCPs. Many parents reported wanting clear, understandable explanations as well as timely follow-up care. One family shared this experience with a HCP that occurred shortly after the birth of their child:

"[Right after the birth they said,] ‘There is a good chance your baby is a girl, but your baby could also be a boy, and so we’re just going to try to refer to the baby as the baby... We’ll do a chromosome test, and by tomorrow you should know whether your baby is a boy or a girl.’ And that was fine, I could understand that." (P7)

However, communication broke down with this family. Two days passed and the parents were not given the results of the testing:

"This was the hardest part for me... nobody came to me... It wasn’t until very late on Sunday evening when finally I kind of broke down. I was sleeping [in the NICU] after I was discharged and I said to the nurse, ‘They said they were going to say something, and no one told me anything.’ And she told me, ‘As far as I know, she’s a girl. They did the testing, it came back that she’s two X, but I can’t give you that official word. I’ll have the doctor come over and give you that.’ The doctor on duty, it wasn’t even the doctor I was dealing with, it was just the doctor who was there that night, [she said], ‘Your baby is a girl. She has two X’s. Then finally I said, ‘Phew. Now I can stop avoiding people saying the baby, and just kind of now refer to her officially as a girl.’" (P7)

Directiveness. During some interactions parents encountered HCPs who were directive in communicating information about medical care options. One parent described the following conversation with the urologist:

"The urologist said: ‘Certain things are going to have to be fixed. Because obviously if female was the end result, they would have to go create a vagina and
everything. He doesn’t have one. And if male was the end result, then there are going to have to be—’ And I was just like, ‘No, there doesn’t have to be any of these things. A person can grow up without these things.’ We had already decided that unless there was a medical necessity we were not going to do anything surgical any time, not at this age at all.” (P3)

**Theme: Parent as Advocate**

Many parents related that they became advocates for their child’s medical care. One parent described the difficulty they encountered in trying to find a diagnosis for their son before surgery to repair a more severe form of hypospadias:

“They just wanted to operate [on] him, and they didn’t want to test, and we had to have a lot of conversation with [the doctor]. Finally the endocrinologist said, ‘It’s okay. We will test it, but I think we won’t find anything.’” (P1)

Testing revealed the child had a DSD. Another parent described the process of becoming their child’s advocate in the following way:

“I was a first-time mom, you live and you learn... I just was so into trusting anything the doctors said. But I don’t want it to sound like I don’t trust doctors, because I do, but at the same time I trust myself too.” (P6)

2. **Experience and Expertise of HCPs**

Two major themes emerged from the discussion with parents regarding the experience and expertise of HCPs in caring for children with DSD.

**Theme: Prior Experience with Child's Condition**

Most families (five out of seven) had to travel an hour or more from home to find HCPs with expertise in caring for children with DSD. For the parents interviewed, travel time to their current urologists and endocrinologists ranged from 0.5 to 3 hours with a mean of 1.6 hours. One family who had recently changed their child’s medical care reported:
“Well [the hospital] is going to be more like three hours for us. It’s a goodly ways, but it’s worth it to me. [The hospital] has always had a good reputation in the field, so I’m hoping at least I’m opening the door to people that have some more familiarity and expertise.” (P2)

Most parents were satisfied with the experience and expertise of their child's endocrinologist and urologist. However, DSD are rare and it can be difficult for parents to find a HCP who has experience with their child’s condition.

“He told me that in his 25 years of practice as an endocrinologist he’s never treated a patient with it, never had a case of it, so zero experience, and the same for Dr. [urologist].” (P3)

Two of the families who had children with CAH reported that their children had experienced multiple adrenal crises requiring hospitalizations or emergency room (ER) visits. For these families, the experience and expertise of the endocrinologist was crucial for the health and safety of their children. One of the parents described the following serious crisis:

“Her potassium skyrocketed... and her sodium was almost nonexistent... the first night we were there [in the hospital] our endocrinologist looked at us and said, ‘I don’t know how to get this down. I don’t know how to bring her potassium down... I have tried everything that I know.’ And it was about 3:00 in the morning, and he goes, ‘I’ve got one more thing I can try. I’ve got feelers out to different people to see what they would suggest.’ So, then we started the kayexalate, which pulled the potassium out of the formula. We got her potassium down... we were in and out [of the hospital] for the first six months. So, I think I figured it out we were in the hospital for like six weeks out of the first four months, and then another two weeks for the next two months. So that was the first six months of her life.” (P5)

The parent of the other child with CAH who has experienced multiple adrenal crises reported that her new endocrinologist has established a specific protocol for the parents to follow that has reduced trips to the ER.
“I really appreciate the protocol that my doctor now has vs. before. [Before] I would have a letter from them that said, ‘If vomiting but looking good--’... It was just so non-specific I didn’t ever really know what to do. Where now, this particular doctor that I have, they have a very specific protocol to follow, so then you’re like, ‘Okay, it’s time to give the shot’... and they make sure you know how to give the shot... that has just taken away so much stress from me, because now I don’t have to bring them to the ER... now that I just have such a protocol, like, ‘Okay this is when I have to give the shot,’ then I don’t have to sit there and think, ‘Should I? Shouldn’t I? ER? Not ER?’” (P6)

Parents encountered problems with HCPs who were inexperienced in treating their child's condition during ER visits or with doctors and nurses who did not have experience treating children in adrenal crisis:

“If we go to Urgent Care, we’ve had issues where it’s like, [I say] ‘She needs IV fluids.’ [They say] ‘Why would she need that?’ [I say] ‘Well, look at her. She’s dehydrated and she’s not moving.’ And they’re like, ‘Well, it’s not a big deal.’ [I say] ‘Well, see, this will kill her.’ So then we have to explain what it is.” (P5)

Sometimes parents received conflicting information from HCPs with limited experience as described by one mother:

“[My child’s] weight is kind of low, and so the pediatrician is like, ‘Well, she’s failure to thrive.’ And the endocrinologist is like, ‘She’s fine.’ Because a lot of kids that have this deal with growth and things like that and that’s part of it. And so sometimes I feel kind of bad.” (P6)

**Theme: Surgical Decision-making**

Most parents reported that the decision of whether or not to choose genital surgery for their child as the most difficult. Some parents expressed concern that they were making the right decision and for putting their child through the trauma of surgery. One mother who made the decision to proceed with genital surgery for her child shared her thoughts:

“I know it’s going to be a controversial decision, and at some point I’m fully prepared to have [my child] say, ‘Why did you do it?’ On the other hand, I also
know if I hadn’t done it I probably might would have gotten the same thing. ‘Why didn’t you do it and give me a—’ I know with adolescents and young adults you’re going to catch it either way... I’m prepared. We made a decision that I think has been in [my child’s] best interest. Can you always second guess? Would it maybe have turned out just as well? Maybe. I mean, I don’t know. We just did the best we could... And I also know that I think the person will come around to knowing, to accepting that this is their life and that their parents made the best decisions they could with what they were given at the time.’” (P2)

For parents who have made the decision to have genital surgery for their child, the expertise and reputation of the surgeon, as well as the type and timing of the surgery are important considerations. One mother described why she chose a particular surgeon:

"That's exactly why I went to [this hospital] for the urology, because it was number one, the doctors were the top one percentile in the area, the surgery is extremely amazing" P7

Another parent related how the urologist explained the importance of the timing of the surgery for hypospadias.

“She really explained [to] us why, because... the older he gets the more complicated it is to do the surgery... It’s more difficult to heal. Maybe now when he’s small he has two surgeries and possible when he’s eight or nine it’s more like four. She really explained this very well.” (P1)

In the Consensus Statement (Lee et al., 2006), the recommendation regarding surgical invention is that the emphasis in all cases should be on functional rather than strictly cosmetic appearance. In keeping with the guidelines, this was the recommendation that was given to a parent by the urologist:

“[The] next step is to put her under anesthesia and confirm that the anatomy of my daughter’s vagina and her bladder and those things are all in the right place so that there isn’t any medical need for surgery, And he said he believes that if everything is as he sees it to be or believes it to be, it’s not going to be necessary for her to actually have a major surgery to do anything, it would all kind of be cosmetic” (P7)

Another parent reported receiving the opposite recommendation from a urologist:
“To him, I was coming off as being close-minded to surgical intervention. And I asked him… ‘Well what argument can you give me for doing [surgery] at age two or age 18 months or whatever?’ And he said, ‘Well, it’ll heal faster.’ … Well I’ve read enough to know that that’s not necessarily true for kids this age, that they need to be at least potty trained before you consider it, because the healing time is fairly lengthy. And he said, ‘Well, because in school other kids are going to see them going to the bathroom and they’re going to get teased in the locker room.’ And I said, ‘Well, he’s going to be going to a very small private school that doesn’t have communal bathrooms and also doesn’t have sports teams or locker rooms, so that’s not an issue.’ And it was just like he didn’t give me anything except social stuff.” (P3)

3. Coordination of Care

Two major themes emerged in response to questions regarding coordination of medical care among the different specialties.

Theme: How Coordination is Achieved

Most parents reported seeing individual specialists for their child's medical care. Sometimes a specialist would recommend another specialist. However, most parents assembled a multidisciplinary team on their own:

“We’ve kind of hodgepoded this team together for her… from the [online] parent groups, there was a big stress on getting a team together, so I always felt like we didn’t have a team for her, really, until we found the geneticist. And so I felt more like I was doing everything I could once we had that team put together.” (P4)

Several parents reported that they would have preferred a clinic that had a multidisciplinary team to help coordinate their child's care but there is none in their area.

Ongoing communication with the child's pediatrician is an important aspect of the child’s medical care. Some parents reported that their child's pediatrician was not a significant part of the team while others had pediatricians who were very involved:

“I had found our pediatrician long before the endocrinologist because you have
to get set up when you’re pregnant. So she came in while we were in the hospital after the birth. She was there when the endocrine doctor was there, and so she got to hear a lot of what he said. She worked very closely with him. She had never heard of it. Well, she had heard of it, she had never seen it, had never treated anybody with it. So she was basically learning on the fly. We had releases for each doctor to be able to communicate with each other. And they did, and they did very well. Our pediatrician had no problem calling our endocrine doctor saying, ‘Hey, I don’t know, does she need to see you for this or is this something that we can handle?’ So they worked very closely together in the beginning.” (P5)

Two families reported receiving their care through an established multidisciplinary team with expertise in DSD that included pediatric specialists in endocrinology, urology, genetics, and psychology/psychiatry. Both of these families had previously received care at different clinics that did not have multidisciplinary teams.

“We were happy that we made the transfer so we could work with more professional people. They listened to us and were very close as a team... We had an appointment with the psychologist, endocrinologist, and urologist... when we made an appointment with one of them they planned it together. Even the psychologist asked, ‘If you want, I can lead the conversation if you have trouble asking questions, or I can help you.’” (P1)

One parent had many years of experience seeing individual specialists before finding care through an established multidisciplinary team explained the advantages of the team approach:

“It’s the doctor, a nurse, and then the psychologist. They all come to all the... appointments... I had to take [my child] to the urologist, and they just wanted to check her out post-surgery or whatever, and he’s part of it. There’s just a team of doctors. I like it, because they agree on everything before they see the patient, so that way patients don’t see them and get five different answers from five different doctors.” (P6)

Theme: Diagnosis

For the three families in the study who had daughters with CAH, diagnosis was made during the first week of life. The time until diagnosis for the other four children
ranged from 6 months to one year. Two of these families reported that the delay occurred because they had not been referred to a geneticist. Involvement of the geneticist sometimes happened upon recommendation of either an endocrinologist or urologist:

“But he was the first one at six months to want us to see a geneticist. No one had mentioned doing that at that point. So then we met with the geneticist pretty quickly after that, and she sent off the blood samples to see if we could get a definite diagnosis.” (P4)

4. Sources of Psychosocial Support

Two major themes emerged from the discussions of psychosocial support.

Theme: Availability of Specialized Psychosocial Care

Psychological support services with expertise in DSD were not available to parents unless they received care through an established multidisciplinary team that included this specialty. Two parents reported receiving their care through teams that included psychosocial support. Only one family reported ongoing involvement with a psychologist as part of their child’s medical care and shared this description of the experience:

“Very good. The first time...we talked about when [our child] goes to school they have to go to the shower ... How we do that and what to tell other children... And she told us then about surgery, if we don’t want to have the surgery. If it’s not necessary we don’t have to do it... Also [the psychologist] knows [our child]. Every time we go he also sees the psychologist, so they get a balance. Later on when he’s older he’ll maybe speak with her alone, he’ll ask questions, how to do things.” (P1)

Two parents reported that the only psychological support they received was shortly after the birth of their child when a social worker visited them briefly in the hospital. Parents who did not use specialized psychological support gave a variety of reasons: It had not been available, was not needed or not easily accessible. One parent
reported that their endocrinologist recommended a specialist for psychological support who was 900 miles away. Two parents reported that while they had not received specialized psychological care in the past they might be interested in the future:

“[My child] is really approaching the age I would like to find somebody. We are in a very small town.... I would love to find somebody... but to find somebody that specialized or had some knowledge of the psychiatry of this would be great. I don’t even really know where to begin.” (P2)

A third parent reported that before they made any decision about genital surgery for their young child they would like speak with some with expertise in this area:

“I will say I certainly would like to use the... clinic to talk to a psychiatrist or some form of therapist to discuss this.” (P7)

**Theme: Other Sources of Psychosocial Support**

All parents reported they received support from a variety of sources.

**Role of the Internet for psychosocial support.** Most reported the Internet as an important resource for psychological support. For a parent, having a child with a DSD can be an isolating experience because of the difficulty of discussing their child's condition with family and friends. Most parents interviewed valued the Internet for the ability to find and connect with other parents whose children have the same or similar conditions. They reported that it is helpful to talk to other parents who understand what they were experiencing.

“We have a group of 10 parents all with adoptive children with DSD... we met [on] the Internet... Once a year we come together with them, and the rest we do on the Internet.” (P1)

However, two parents reported that there are negative aspects of the Internet and support groups. One of the parents described a negative experience with an online
support group when she shared information about her child’s surgery:

“One [group] is very, very judgmental in its anti-surgery stance... that’s a little bit of the danger of those groups. They tend to take one side or the other... unless you go out and open your own group for the other, it doesn’t have a balanced approach in my mind... I’ve been pretty open about [our surgery], and nobody has been hateful towards me about it, but you can tell there’s disapproval there... I’ve been contacted by others that [say], ‘We just don’t feel comfortable at all speaking up about it.’ And what I find is... on the groups there’s probably just about as many people that have [chosen surgery] as that haven’t, they’re just not the vocal ones.” (P2)

**Role of family and community for psychosocial support.** Parents reported that they received strength and support from people in their lives such as spouses and parents as well as their faith and churches:

“My faith. I know God is good, and he watches over my children, and I need not fear.” (P6)

When parents find a community of parents who have the same rare condition as their child it can play an important support role in their lives. Parents with experience also gain from helping other parents:

“I just talked to a mom, her daughter just had surgery last week. If I was a little bit closer I would have driven down there and just been with her because I know what we had gone through, and I never knew anybody who had gone through it. But still at the same time we were texting, and she would ask me, ‘Well, after did you do this or did you do this?’” (P6)

5. **Role of the Internet for Information**

All parents reported that the Internet was an important resource for information. Parents described belonging to online organizations and support groups that provided information that helped them in three areas: education, decision-making, and finding care.

“I don’t know that any physician could sit down and fill you in on this subject or
educate you 100%. If you didn’t do it on your own, ... I have a little bit of a medical background with being an RN. I think for the average person it’s got to be very, very daunting, and certainly very complex.” (P2)

Information gained through the Internet led one parent to question their child's clinical diagnosis, which eventually led to a change of diagnosis after further testing:

“I had been reading a lot, and from the way she looked I didn’t feel she was complete androgen [insensitivity], because they generally look completely normal female, and so I had been kind of struggling with that, because it made me nervous to think that she might be partial, because then you go back into the, ‘Well, should you be raising her male then?’ And so we talked to her endocrinologist. I think he was nervous about the partial diagnosis too, because... he kept just saying, ‘Well she’s nearly complete.’” (P4)

Through online support several parents reported learning from other parents what to expect in the future which helped them to feel better prepared. The parent of a young child with CAH shared this experience:

“I think that really it wasn’t until I read other mothers’ experiences-- And I know each child reacts differently, some it takes a lot more to go into crisis than others. But some parents say teething can put their kids into crisis. And that’s like, ‘Oh my gosh, this could be-- Babies teethe all throughout the year.’ And that seemed overwhelming at first, but I really feel like I have a better handle on it now.” (P7)

Several parents reported that they had been looking for adults with their child’s condition through the Internet. One parent who is raising their child as boy has only found adults who were raised as women with their son’s condition:

“Maybe years back when they were young it was easier to raise them as girls, and they did not have maybe the surgeons who could do the operations. That’s why maybe we only speak [to] women... I spoke to three adult women, and two of them [feel like] a boy, and they have poor relationships with women. So, that was also good to hear, that they feel as a boy.” (P1)

Parents report using the Internet and online support groups as resources to help make decisions. One parent, who is in the process of gathering information before
making a decision about surgery for her daughter, is using the Internet to contact adults with her daughter's condition:

“I’m trying to get the opinions of other women that have CAH whose parents opted to have surgery on them before they could make a decision of their own. So I’m really looking for that point of view, because I want to know if there are a lot of women out there or a few women out there or any women out there who feel their parents made the decision for them that they would not have made for themselves.” (P7)
DISCUSSION

In this study, we sought to examine the experiences of parents of children born with ambiguous genitalia due to a DSD and to report their perceptions of the medical care they received in relation to the Consensus Statement on the management of DSD (Lee et al., 2006).

Communication with HCPs

Communication emerged as the most important aspect of the parent's experience with their child's HCP. Effective communication is the foundation of the HCP-parent-child relationship. According to a technical report in Pediatrics (Levetown, 2008), the communication skills of the HCP influences all aspects of patient care from diagnosis to developing a treatment plan. Skillful communication correlates with improved health outcomes, better adaptation of the family to a challenging diagnosis, increased treatment compliance, better patient understanding and improved psychological and social outcomes. In contrast, poor communication can lead to lifelong anger and regret for the family as well as poorer outcomes that may have legal implications for the HCP.

Parents in this study described positive experiences with HCPs who listened and who were caring, empathetic and respectful of them and their children. This finding is consistent with previous studies (Beach et al., 2005; Bendapudi, Berry, Frey, Parish, & Rayburn, 2006; Nobile & Drotar, 2003) Bendapudi et al. (2006) in a survey of patients identified seven ideal behavioral themes. The ideal physician is confident, empathetic,
humane, personal, forthright, respectful and thorough. Our findings also showed that when parents were unsatisfied with the communication skills of a HCP they would often seek out a new HCP.

Each parent reported an equal number of positive and negative interactions with HCPs. Parents described these experiences as contributing to becoming an advocate for their child's healthcare. When the relationship between the parent and HCP was one of mutual respect parents regarded themselves as partners in their child's care.

**Experience and Expertise of HCPs**

Parent reported that they were willing to seek expert care that was distant from home if none was unavailable in their community. Some participants related that their HCPs had no experience with their child's condition, however, this did not impact their ability to receive expert care from this provider. Other parents described that they encountered HCPs who had extensive experience with their child's condition but they were not satisfied with the quality of the care from these providers. Therefore experience alone was not an indicator of quality for these parents. These findings are supported by a recent study of members of multidisciplinary DSD teams and DSD support groups. Streuli, Kohler, Werner-Rosen, and Mitchell (2012) identified four categories of professionalism needed for the responsible treatment and support of children and families affected by DSD: child-oriented professionalism, family-oriented professionalism, shared-decision-oriented professionalism and quality-oriented professionalism. Professionalism refers to skills and related conduct, aims, and qualities that ultimately characterize expert and ethical HCPs.
For the three parents of children who have CAH, two encountered HCPs outside of their specialists (pediatricians or emergency room personal) who did not understand various aspects of the care of a child with CAH. This can be a life threatening circumstance for a child in adrenal crisis. These experiences demonstrate the need for training of emergency room providers and pediatricians as well as the development of protocols for children in adrenal crisis. Auchus et al. (2010) recommended that parents of children with CAH be provided with tools, such as pamphlets or laminated cards, that allows them to effectively advocate for themselves or their children in emergency situations.

All parents expressed the need to gather information before making a decision regarding genital surgery. The expertise of the HCPs is crucial in providing parents with the most up-to-date information particularly when there may be no clear "best-choice". Many parents described the desire to understand the advantages and disadvantages of the different options and being supported by their HCP in whatever decision they make. Karkazis, Tamar-Mattis, and Kon (2010) proposed a shared decision-making approach regarding decisions about genital surgery for children born with DSD. The authors detail six sequential steps to address parents' emotional and informational needs and to allow for reflection and transparency during the decision-making process.

**Coordination of Care**

According our findings, access to an experienced multidisciplinary DSD team as recommended by The Consensus Statement (Lee et al., 2006) was not available to most parents interviewed. Several parents reported forming their own teams with variable
success at coordination of information and care between the specialties. In this approach the parent becomes the team coordinator. While parents reported success with this strategy it is limited when compared to a model of care that has a fully functioning multidisciplinary team. According to the Consensus Statement (Lee et al., 2006) the responsibilities of the team include: Ongoing communication with the family's primary care physician, education of other health care staff in the appropriate initial management of affected newborns and their families, development of a plan for clinical management with respect to diagnosis, gender assignment, and treatment options before making recommendations. The two families who eventually found care through a multidisciplinary team expressed relief and satisfaction with this approach.

Two parents reported that they experienced a delay in the diagnosis of their child's condition because they had not been referred to a geneticist from the outset. Neither of these families received their care through a DSD team. Timely, accurate diagnosis is essential for children with DSD because in many cases families are making surgical or treatment decisions that may have lifelong implications for the child. The wrong decision based on incomplete information may lead to physical and psychological problems for the child in the future (Warne, 2008).

Genetic counseling can play an important role on a multidisciplinary team. Parisi et al. (2007) reporting on the experience of the DSD team at their clinic, states that the genetic counselor is usually the initial point of contact for the team and also coordinates the team. The genetic counselor contacts the appropriate specialists and coordinates care for the families. By the nature of their training, genetic counselors are well suited to help
families to understand the complexities of the information they encounter and to provide psychosocial support when needed.

**Psychosocial Support**

The birth of a child with a DSD is a stressful event for families. Parents may be concerned about uncertainty of their child's gender as well as the child's future psychological and psychosexual development. As reported in other studies, some families interviewed were able to relieve some of the stress through the support of trusted family and friends, however, not all families utilized these resources because they wished to protect their child's privacy and viewed disclosure as too risky (Crissman et al., 2011).

Specialized psychosocial care with expertise in DSD was only available to two families in the study. Several other parents expressed interest in seeking psychosocial care but none was offered in their area. According to the Consensus Statement (Lee et al., 2006) psychosocial support should be an integral part of caring for families affected by DSD to promote positive adaptation. Psychosocial support may be ongoing or only utilized at certain times during the course of a family's care. Initially, support may primarily be for the parents but as the child grows up they should have access to this care for their own needs and concerns.

There is a need for training of HCPs who understand the psychosocial needs of patients and their families affected by DSD. This role may be filled by a number of different specialists such as: psychologists, psychiatrists, social workers or genetic counselors.
Role of the Internet

While HCPs were the first choice for information for most of the participants, all emphasized the significant importance of the Internet as a resource. A report from The Pew Internet Research Institute (Fox, 2011) indicates that there has been an explosion of health-related information on the Internet. The Pew study found that 80% of American Internet users look online for health information. The study also reported that 70% of American Internet users who are also caregivers look online for health information and are more likely than other adults to use social network sites to gather and share health information and seek support.

In the current study, the discussions with parents of the role of the Internet for information and support reflected findings in previous studies of parents who had children with rare genetic conditions. Gundersen (2011) found that in addition to filling a need to obtain and share information, exchanging information online helped parents feel more secure in their role as caregiver. The parents in the current study discussed the process of adjusting and coping with their child's condition. Initially parents described the stress and shock they experienced. As they gathered information from all sources, including the Internet, they were able to comprehend and manage their situation and reduce their stress. Parents also discussed the value of knowing that there are other children like their own. Having a child born with ambiguous genitalia is a rare occurrence, that combined with a parent's natural desire to protect their child's privacy often prevents parents from learning about other children in their community with the condition. The Internet draws upon a wide geographic area as well as culturally and
economically diverse communities. It can also provide anonymity for parents and the reassurance that their child's privacy is protected.

Another area of discussion with parents involved the importance of learning about their child's condition so they could become an advocate for their child. Parents described gathering information from the Internet and bringing it to their HCPs. A study by McMullan (2006) found that HCPs can respond to the 'Internet informed' patient in one or more ways of three ways: 1) HCP feels threatened by the information the parent brings and responds defensively by asserting their 'expert opinion' (HCP-centered relationship), 2) the HCP and patient collaborate in obtaining and analyzing the information (patient-centered relationship), and 3) the HCP will guide patients to reliable health information websites (Internet prescription). One of the concepts of optimum care of the Consensus Statement (Lee et al., 2006) is that parents' participation in decision-making is encouraged. When HCPs embrace the 'Internet informed' parent this allows parents to become informed partners in the decision-making process.

Parents also discussed negative aspects of the Internet. Sometimes information is inaccurate or may not apply to their child. Parents may have differing opinions and express them strongly. If HCPs are familiar with the specific support groups on the Internet they can help parents navigate through the abundance of information to find organizations that provide accurate medical information and a supportive community as well as caution parents about the problems they may encounter.
**Study Strengths and Limitations**

This was a small qualitative study designed to explore the experiences of parents of children born with ambiguous genitalia due to DSD and to examine their perceptions of the medical care they received in relation to the Consensus Statement (Lee et al., 2006). The participants generously shared their experience so that others can have a better understanding of what it means to have a child born with a DSD. However, the sample size was small, consisting of only eight parents during seven interviews. A larger sample size would allow for expansion of the analysis and data saturation. One of the interviews included both parents while six were only with the mother. We did not notice a difference in the amount of information shared during the interview with both parents but it is not possible to quantitatively verify differences. Recruitment through online support organizations likely caused sampling bias. It is possible that parents who are active in these groups may have specific kinds experiences or characteristics that result in them being more willing to participate in this study. In addition, demographic information such as race, socioeconomic status, and level of education was not collected. Therefore, the results of this study may not represent the views and experiences of all parents who have children born with ambiguous genitalia.

A strength of the study is that participants were from a diverse geographical area, and included parents from across the United States and outside the United States. In addition, there was a diversity of types of DSD of the children of the participants.

One confounding factor was that four of the eight participants were adoptive parents. While none of the adoptive children had a diagnosis of a DSD before their
adoption, all of the adoptive parents were aware that their child had ambiguous genitalia before proceeding with the adoption. In contrast, all of the non-adoptive parents learned of their child's condition at birth. Although the adoptive parents may not have appreciated the complexities of their child's condition at the time of the adoption, this difference in the two parent groups may have impacted their experiences and influenced the challenges these families encountered.

Another limitation is that all seven interviews were conducted over the phone to offer greater access for participation. While this maintained consistency, it may have influenced the way the interviewer built rapport and trust with the participant in the absence of eye contact and other non-verbal communication. On the other hand, the confidentiality afforded by the telephone may have allowed participants to express feelings they otherwise may not have explored.

Finally, as with all qualitative studies, there was the limitation of recall bias. Because interviews were conducted at varying intervals after some of the events described, it is possible that the participants' memories and emotions have changed with time. Also, it is important to consider the subjectivity and personal bias of the author when coding the interviews and interpreting the data.

**Future Research**

Continued research in this area should explore the experiences of young people growing up with DSD. A deeper understanding of their experiences will inform HCPs and parents in all aspects of care. In addition, a longitudinal study of families from the birth or adoption of a child would allow for comparison at specific periods of time during
the course of a family's experience. Further studies are also needed to identify areas for improvement in the care of children with CAH.
CONCLUSION

This study explored the experiences of parents of children born with ambiguous genitalia in relation to the guidelines given in the Consensus Statement (Lee et al., 2006). Although, full implementation of the guidelines was not available for most participants, the study found that families have access to high quality care for their children with DSD. The parents' narratives indicated that it is important to improve care to relieve the stress placed on these families.

Parents often expressed a need for better communication with their HCP. HCPs who listened, were caring, and demonstrated empathy and respect were more highly regarded and families were more likely to remain under their care. Conversely, parents had lower regard and less confidence in HCPs who were directive or insensitive. Specialized training of HCPs to develop effective communication skills can improve the health outcome of patients and result in better adaptation by the family.

Multidisciplinary teams were not accessible for most parents. Those that received their care through these teams expressed the most satisfaction with their care indicating that greater access is needed. In the absence of a team, parents usually reported coordinating care for their child and acting as the team leader.

Specialized psychosocial care is not accessible for most families. This could be addressed by offering training rotations for psychologists, social workers and genetic
counselors. In addition, HCPs should be aware of the support groups that are available for the specific conditions. They should familiarize themselves with what type of information is provided and the accuracy of that information. These are important resources that parents will seek out on their own. By acknowledging their importance, the HCP can help parents understand the positive and negative aspects of these groups.

Parents want to make the best possible decisions so that their children will have happy and fulfilling lives. The findings in this study reinforce the importance of taking steps to increase adherence to the guidelines to provide the best care and support for these parents and their children.
REFERENCES


Gundersen, T. (2011). 'One wants to know what a chromosome is': the internet as a coping resource when adjusting to life parenting a child with a rare genetic


Appendix A: Recruitment Notice

Are you the parent of a child who was born with ambiguous genitalia?

I am a graduate student in the Genetic Counseling Program at Brandeis University and I am seeking volunteers to participate in a research project. The goal of this study is to explore the experiences of parents of children who are born with ambiguous genitalia. Participation in this research study is open to all individuals:

- who are the parent of a child born with ambiguous genitalia who is currently between the ages of 3 months and 10 years as of January 1, 2012
- who are 18 years of age or older
- who are fluent in English (open to international participants)

What will be involved if I participate?
Participation in this study is voluntary. Following a brief phone conversation to assess eligibility, participants will be asked to take part in an audio-recorded phone interview that will last approximately one hour. I hope to have all phone interviews completed by February 13, 2012. All participants will receive a $25 Amazon gift card as a token of appreciation for their time and expertise.

How will my privacy be protected?
All identifying information for the participants will be kept confidential and will be destroyed after completion of the study. Identifying details will be changed to protect the privacy of all participants.

If you are interested in participating in this study, please contact me by email, Nancy Herrig at nherrig@brandeis.edu by January 27, 2012.

If you know of anyone who might be interested in participating please feel free to pass this information on!

I appreciate your willingness to participate in this study and look forward to hearing from you.
Sincerely,

Nancy Herrig, Student Researcher/Genetic Counseling Graduate Student
Dr. David Rintell, Principal Investigator/Faculty Advisor
Brandeis University
Waltham, MA
Appendix B: CAH Recruitment Notice

Are you the parent of a child who was born with ambiguous genitalia due to congenital adrenal hyperplasia?

I am a graduate student in the Genetic Counseling Program at Brandeis University and I am seeking volunteers to participate in a research project. The goal of this study is to explore the experiences of parents of children who are born with ambiguous genitalia.

Participation in this research study is open to all individuals:

- who are the parent of a child born with ambiguous genitalia who is currently between the ages of 3 months and 10 years as of January 1, 2012
- who are 18 years of age or older
- who are fluent in English (open to international participants)

What will be involved if I participate?

Participation in this study is voluntary. Following a brief phone conversation to assess eligibility, participants will be asked to take part in an audio-recorded phone interview that will last approximately one hour. I hope to have all phone interviews completed by February 13, 2012. All participants will receive a $25 Amazon gift card as a token of appreciation for their time and expertise.

How will my privacy be protected?

All identifying information for the participants will be kept confidential and will be destroyed after completion of the study. Identifying details will be changed to protect the privacy of all participants.

If you are interested in participating in this study or would like more information, please contact me by email, Nancy Herrig at nherrig@brandeis.edu by Jan. 27, 2012.

If you know of anyone who might be interested in participating please feel free to pass this information on!

I appreciate your willingness to participate in this study and look forward to hearing from you.

Sincerely,
Nancy Herrig, Student Researcher/Genetic Counseling Graduate Student
Dr. David Rintell, Principal Investigator/Faculty Advisor
Brandeis University,
Waltham, MA USA
Appendix C: Phone Intake and Eligibility Screening Tool

Name:
Location:
Phone Number:
1. What is your age?
2. What is your child’s date of birth?
3. Was your child born with ambiguous genitalia?

If yes, has your child received a diagnosis? If yes, what is their diagnosis?
(If parent not eligible, I will explain why and thank them for their time)
4. I would like to review the informed consent form with you now. (Appendix D).
5. I will send you the informed consent form that we have just discussed. Please read it over and if you wish to participate sign the participant’s statement on the last page and return it to me. Once I have received statement I will sign it and send you a copy. If you have any questions about the form please do not hesitate to contact me. What is the most convenient method for me to send the form to you (mail, email, fax)?

You may fax, email, or mail the signed form to me before our interview. Which method would you prefer?
6. When would be a convenient time to schedule our interview?

Date:  Time:  Participant’s time zone:

Now I just have a couple of questions before our interview
7. At what hospital was your child born?
8. Is this the same hospital where your child continues to receive care?

-If no, where does your child currently receive care?

Thank you for your time
Appendix D: Informed Consent Form

BRANDEIS UNIVERSITY DEPARTMENT OF BIOLOGY GENETIC COUNSELING GRADUATE PROGRAM
Informed Consent to Participate in Research
An Assessment of the Current Medical Care of Children Born with Ambiguous Genitalia from the Parent Perspective

Principal Investigator: David Rintell, Ed.D.
Student Researcher: Nancy Herrig

INTRODUCTION
David Rintell is a licensed clinical psychologist. He is also a faculty member for the Genetic Counseling Graduate Program at Brandeis University. Nancy Herrig is a Master’s candidate in the Genetic Counseling Program at Brandeis University. This is a research study to explore the recent experiences of parents of children born with ambiguous genitalia.
You are being invited to participate in this study because you have a child who was born with ambiguous genitalia. 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 and 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 explore the experiences of parents of children between the ages of 6 months and 10 years old who were born with ambiguous genitalia that was diagnosed at birth. We hope to better understand the current medical care being delivered to these families and the experiences of families from the parent perspective. It is our hope that the experiences shared by parents in this study will be useful in educating medical professionals in this area as well as helpful to other parents of children with this condition.

PROCEDURES TO BE FOLLOWED
You will be asked to participate in an audio-recorded telephone interview lasting approximately one hour. During this interview, you will be asked questions regarding your experiences as a parent of a child born with ambiguous genitalia.

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, Dr. David Rintell, a licensed clinical psychologist, is available by telephone to talk with you (617-734-6778).

**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 experiences of parents of children who are born with this condition.

**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 digital audio files) will be kept in a secure location accessible only to the student researcher, and any databases containing identifiers will be password-protected using a password known only to the student researcher. Transcripts, interview notes, and digital 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 the study, you will be given an alternate 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**
There is no payment for your participation. You will receive a $25 Amazon gift card 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, Nancy Herrig, at nherrig@brandeis.edu or (781) 771-6041. You may also contact the Brandeis University Faculty Sponsor for this project, Dr. David Rintell, at rintell@brandeis.edu or (617)-734-6778. 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 Nancy Herrig 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.
Please indicate your willingness to be audio-recorded by initialing here: __________

Date __________  Participant’s Signature __________

Participant’s Name (Please Print) __________

Date __________  Student Researcher’s Signature __________
Appendix E: Interview Guide

1. I know we spoke briefly about this on the phone, but can you describe when you first learned of your child’s condition?
   Probes: Who told you?
   - What was the initial explanation you received?
   - What were you told about the gender of your child?
   - What were your thoughts after receiving this information?

2. Do you have a family history of any conditions affecting reproductive, urological or sexual function?
   Probes: Urologic abnormalities, neonatal deaths, amenorrhea, infertility, consanguinity
   - Which family members and what conditions do they have?

3. After this initial period, what Dr.(s) or other medical professionals met with you?
   Probes: Did your hospital have a clinic for specifically for children with conditions affecting sexual or reproductive function?
   - Did you meet with a team, one person or different people individually?
   - Did you meet with a genetic counselor or geneticist?
   - What was your experience with the different medical personnel?

4. What testing was done for your child?
   Probes: physical exams, blood tests, ultrasound, laparoscopy

5. What were you told when the results of the tests returned?
   Probes: How long did it take to receive the test results?
   - What diagnosis did you receive for your child’s condition?
   - What were you told about any risks associated with this condition?
   - What were your thoughts at this point?

6. Has your family received psychosocial support?
   Probes: Who provided the support?
   - Please tell me what this experience was like?
   - Do you plan on continuing with this support for you and your child?

Now let’s talk about the decisions you had to make regarding your child’s condition.

7. What information were you given regarding choosing your child’s gender?
   Probes: Who did you discuss this with?
   - What information or recommendations were you given?
   - If your child was diagnosed with a specific condition, what were you told about gender assignment in other children with this condition?
   - What were your thoughts after receiving this information?

8. What decision was made regarding your child’s gender?
   Probes: Please describe what this experience was like

9. Were surgical options discussed with you?
   Probes: Who did you discuss this with?
What information were you given, i.e. functional or cosmetic outcomes, risks, timing, or possible need for multiple surgeries?
What recommendations were made?
What were your thoughts after receiving this information?
10. What decision did you make regarding surgery?
   Probes: Has the surgery taken place?
   Please describe what this experience was like.
11. Were hormone replacement options discussed with you?
   Probes: Who did you discuss this with?
   What information were you given, i.e. outcome & risks?
   What recommendations were made?
   What were your thoughts after receiving this information?
12. What decision did you make regarding hormone therapy?
   Probes: Has your child begun hormone therapy?
   Please describe what this experience was like.
Now, a few follow-up questions regarding the different decisions that you had to make
13. Was the medical staff accessible during this time to address questions and concerns?
   Probes: Who was most helpful?
   Who was least helpful?
   Would you recommend this hospital (or team or specific Dr.) to other parents of children with this condition?
14. Did you use other resources outside the hospital to assist you in making these decisions?
   Probes: What were these resources?
   How were these resources helpful?
15. In looking back at this period of time, how do they view it today?
   Probes: What could have made this process better?
   Would you make the same decisions today?
   What would you do differently?
16. What or whom has help you the most through this experience?
17. Is there anything that we have not talked about that you think is important to know?