Surgical Fetal Intervention: Assessing the Current Practices of Genetic Counselors

Master’s Thesis

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ABSTRACT:

The option of surgical fetal intervention (SFI) for the treatment of fetal anomalies is relatively new. It is generally recommended for anatomic problems that cause ongoing damage to either the developing fetus or the mother. It is important that parents considering SFI receive counseling from a medical professional before making a decision; prenatal genetic counselors are in an ideal position to do this. This study was designed to assess prenatal genetic counselors’ current knowledge, experience, and comfort level with SFI, and to compare two groups of counselors: those who work in a fetal intervention setting and those who do not. We recruited prenatal genetic counselors through the National Society of Genetic Counselors (NSGC) Listserv who had a minimum of one year of clinical experience and saw patients at least eight hours per week to take a quantitative survey. The survey consisted of multiple choice, Likert scale, and open-ended questions. We collected the survey data in SPSS and completed ANOVA and correlation analysis. We received 82 completed surveys: 28 from fetal intervention counselors and 54 from general prenatal counselors. All respondents were aware of SFI and 84% reported experience counseling for SFI. However, we found a wide range of knowledge levels from questions on details about the surgeries and the use of SFI for specific fetal anomalies. Those counselors who worked in a fetal intervention setting had significantly higher overall knowledge and comfort scores, but not experience scores. We found that 61.3% of respondents first learned about SFI in their genetic counseling training, but 41.5% felt their program did not prepare them well to counsel for it. Our findings indicate that there is a need for genetic counseling programs to increase the
amount of time they spend teaching about SFI. It also indicates a need for educational materials and learning opportunities for practicing genetic counselors.
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I. INTRODUCTION

Fetal intervention can be defined as any therapeutic intervention for the purpose of correcting or treating a fetal anomaly or condition. Fetal intervention can be divided into the broad categories of non-invasive and invasive techniques. Non-invasive fetal interventions, those that do not involve direct contact with the fetus or its environment, primarily include chemical interventions such as insulin given to diabetic mothers or tocolytic drugs to prevent premature labor. Invasive techniques, or fetal surgeries, are in utero operative treatments of a fetus. They range from inserting a needle into the fetal environment, for procedures such as fetal blood transfusions, to open fetal surgery to correct a fetal lesion, such as a myelomenigocele.

Dr. Liley in New Zealand performed the first successful documented invasive fetal intervention in 1963 with a blood transfusion into the peritoneum of a hydropic fetus with severe Rh disease (Liley, 1963; Redwine, 1993). By the 1980s researchers had begun to experiment with techniques for opening and closing a gravid uterus without disrupting the pregnancy or jeopardizing the mother’s health and reproductive potential (Harrison, 1993). Such attempts at open fetal surgery met with limited success until 1989, when an absorbable staple was developed for a surgical stapling device. This allowed surgeons to open the uterus while preventing both excessive bleeding and separation of the fetal membranes from the uterine wall (Bond, 1989). The most significant limitation to fetal surgery since the advent of the absorbable stapling device has been the risk of
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preterm labor and delivery after the surgery (Kunisaki, 2008).

Today there are three major techniques used for surgical fetal intervention (SFI): minimally invasive fetal surgeries (MIFS), open fetal surgery (OFS), and fetoscopic surgery (FSS). Minimally invasive procedures involve a percutaneous approach, developed in the 1980s, typically used to drain “space-occupying fluid-filled structures” such as the pleural space or bladder of the fetus (Kunisaki, 2008). Under ultrasound guidance, a needle is inserted through the maternal abdomen and into the fluid filled structure. A small wire is then inserted though the lumen of the needle and the needle is removed. A catheter can then be placed over the wire and used to drain the space whenever it is needed. Minimally invasive fetal surgery is generally an outpatient procedure (Kunisaki, 2008).

Dr. de Lorimier and Dr. Harrison of the University of California, San Francisco, pioneered open fetal surgery in the late 1970s and early 1980s. Open fetal surgery is performed under ultrasound guidance with careful monitoring of both the mother and the fetus during the procedure. First, the mother is put under anesthesia and an incision is made in the lower abdomen to expose the uterus. Next, the uterus is opened using the absorbable stapling device and the fetus is externalized. Once the surgical repair of the fetus is completed, the uterus and then the maternal abdominal wall are closed. OFS requires a 3-7 day hospitalization and cesarean delivery of the current and all future pregnancies. Preterm labor and delivery is a significant risk with OFS (Kunisaki, 2008).

Fetoscopy was developed in the 1970s as a diagnostic technique, but lost momentum when ultrasound techniques were refined in the 1980s. However, in the late 1990s, with the invention of smaller lightweight surgical instruments, fetoscopy regained
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popularity as a less invasive approach for fetal intervention. The technique requires a relatively small (1-2mm) incision in the maternal abdomen, through which a probe fitted with a camera and surgical instruments is inserted. Surgeons can thus gain access to the uterus to repair the fetal anomaly (Kunisaki, 2008). Like OFS, fetoscopic surgery is always performed under ultrasound guidance, and has replaced OFS for some, but not all, fetal problems (Kunisaki, 2008).

Over 20 facilities in the United States alone offer fetal intervention services. The most common fetal anomalies corrected through fetoscopic surgery include twin-twin transfusion syndrome (TTTS), twin reversed arterial perfusion (TRAP) sequence, and amniotic band syndrome (ABS) (Senat, 2004; Kunisaki, 2008). Congenital cystic adenomatoid malformation (CCAM) of the lung, fetal sacrococcygeal teratoma (SCT), and some congenital heart defects and urinary tract obstructions can be treated by OFS (Eber, 2007; Redwine, 1993; Kunisaki, 2008). Additionally, clinical trials are currently ongoing for the prenatal treatment of fetal anomalies such as myelomeningocele (MMC) by open fetal surgery (Kunisaki, 2008). In some cases, open fetal surgery takes place at the time of delivery via the ex-utero intrapartum (EXIT) procedure. During an EXIT procedure, a caesarean section is performed exposing only the top half of the fetus. Surgery is then performed while the fetus is still connected to the mother and receiving oxygen through the umbilical cord. When surgery is completed, the fetus is fully delivered. EXIT is most frequently used in the case of severe airway obstructions (Marwan, 2006), in which case the intervention involves securing an airway with intubation or tracheostomy before full delivery.

The diagnosis and assessment of birth defects has greatly improved over the past
fifty years because of enhancements in ultrasound techniques as well as the development of fetal MRI, amniocentesis, and fetoscopy techniques (Redwine, 1983; Coakely, 1984). However, it is important to note that when detected prenatally, the majority of birth defects are still best treated with medical therapy during the pregnancy or with surgery immediately after birth (Kunisaki, 2008). Fetal anomalies that are recommended for surgical fetal intervention are generally anatomic problems that cause ongoing damage to the developing fetus, are associated with a significant risk for fetal demise, or pose significant risk to the mother.

When a fetal anomaly is first detected, parents are often unprepared for the diagnosis (Baker, 1998). It is important at this point that the parents receive counseling from a medical professional so that they are able to move forward with a complete understanding of their situation and the options available to them (Baker, 1998). Genetic counseling is a subspecialty of medicine that “deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family” through a process of open communication (Baker, 1998). Genetic counselors help individuals and their families to comprehend medical and genetic facts relevant to a genetic disorder, as well as make fully informed decisions regarding how they want to deal with the genetic disorder within their family (Baker, 1998). The philosophy of genetic counseling includes a non-directive approach to patient education with complete disclosure of all available information. Genetic counselors also promote a psychosocial dimension in counseling, helping individuals and families to recognize their priorities, beliefs, and fears in order to make informed decisions (Hansen, 2004; Baker, 1998). Additionally, genetic counselors help affected individuals and their families to adjust as best they can to living with a
disorder and to accept the possibility of recurrence in other family members (Baker, 1998).

A genetic counselor is a master’s level professional. The American Board of Genetic Counselors (ABGC) currently recognizes thirty-three accredited graduate programs in the United States. Training in these programs includes four academic semesters or six academic quarters learning genetic, medical, and technical information as well as counseling skills and ethics. In addition, genetic counseling students are required to complete clinical training during their graduate education to support the development of practice-based skills.

Prenatal medicine is one of the major areas in which genetic counselors work. In this area, a genetic counselor’s practice is not limited to genetic conditions. They are also called upon to counsel on issues that may have little or no genetic basis, such as teratogenic exposures, history of pregnancy loss, or the diagnosis of birth defects (Baker, 1998). In most clinical situations an individualized approach that considers the indication, time of gestation at genetic counseling referral, and maternal age is needed. When counseling for the prenatal diagnosis of a birth defect or genetic disorder, genetic counselors give information to help parents both understand and acknowledge the diagnosis and make choices about the course of action they are going to take (Agnieszka, 2007). A prenatal genetic counseling session is an important interaction on which life-or-death decisions may be based. Generally, parents are presented with three options after the diagnosis of a fetal anomaly: terminate the pregnancy, continue the pregnancy while monitoring the fetus, or actively intervene with the pregnancy to treat the fetus (Agnieszka, 2007). If the parents choose to continue the pregnancy, they can choose
either to keep the baby once he or she is born or to give the baby up for adoption. A prenatal genetic counselor will often stay in touch with the parents throughout the pregnancy, and for some time after the baby is born (Hansen, 2004).

Prenatal genetic counselors are in an ideal position to discuss SFI options with parents for whom it is a possibility; yet SFI has never been studied within the field of genetic counseling. Over the past 45 years SFI has gone from a promising idea to a successful practice. Decades of research and trials have established it as a safe and ideal solution for some fetal lesions. However, despite a profusion of publications on both the surgical techniques and the ethics of SFI, little has been published regarding the decision making process of the parents when presented with the option of fetal surgery. Nor is there data regarding the manner in which medical professionals present this choice to parents. Therefore this study was put forth with the following aims:

- To assess prenatal genetic counselors’ current knowledge of SFI
- To assess prenatal genetic counselors’ experience and comfort levels with SFI
- To compare the knowledge, experience and comfort levels of two groups of prenatal genetic counselors, those who work in a fetal intervention setting and those who do not
- To learn about the counseling practices of genetic counselors in a fetal intervention setting
- To ascertain what might be done to help genetic counselors learn about and counsel for this intervention option in the future
II. METHODS

Recruitment.

In order to gather information on the current practices of prenatal genetic counselors regarding SFI, we recruited participants through the National Society of Genetic Counselors (NSGC) to take a quantitative survey (Appendix C). We posted two recruitment notices (Appendices A and B) on the NSGC’s General, Prenatal, and Fetal Intervention and Therapy Listservs. The first recruitment notice (Appendix A) was directed towards prenatal genetic counselors. The second recruitment notice (Appendix B) was specifically focused towards a subset of prenatal genetic counselors who work in or with a center that offers fetal intervention services. This subset of counselors was also recruited via word of mouth. Both recruitment notices had a link to direct participants to an online survey hosted by Survey Monkey (www.surveymonkey.com).

To be eligible for this study, a participant had to be a genetic counselor currently working in a prenatal genetic counseling clinical setting where they saw patients at least one full day, or the equivalent of one full day, per week. A participant was also required to have at least one full year of prenatal clinical experience prior to participating in the study.

A subset of the participants was prenatal genetic counselors that worked directly in or with a center that offered fetal intervention. This subset of counselors was recruited for their specific expertise in fetal intervention (Appendix B). At the end of the survey,
participants who indicated they worked directly in or with a fetal intervention center were invited to participate in an additional 20 to 30-minute telephone interview. Participants were asked to email the primary investigator directly upon completion of the survey if they were interested in being interviewed. Our target number of interviews was between two to six.

**Data Collection.**

We offered participants an anonymous online survey (Appendix C) hosted by Survey Monkey (www.surveymonkey.com) for a period of three weeks. Survey questions were primarily quantitative, and included multiple choice, Likert scale and three open-ended questions. The survey consisted of five sections:

1) Participant information/demographics
2) General knowledge about surgical fetal intervention
3) Experience counseling for surgical fetal intervention
4) Comfort level counseling for surgical fetal intervention
5) Counseling practices of genetic counselors in a fetal intervention setting.

The first four sections were administered to all eligible participants. Section 1 ascertained basic information about the participants, including the state in which they practiced and how long they had been practicing. It also established whether or not they were affiliated with a fetal intervention center. Section 2 assessed prenatal genetic counselors’ general knowledge regarding SFI, including their understanding of different surgical techniques as well as which fetal anomalies are currently treated with SFI. It also asked how counselors had first learned about SFI, and if they were exposed to the topic
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during their genetic counseling training. Section 3 was used to determine *how often* genetic counselors are called upon to discuss SFI with their patients. This section also addressed how genetic counselors prepared for sessions in which SFI may need to be discussed, the topics or issues they typically discussed during a session, and the resources they used as professionals or offered to their patients. Section 4 explored how comfortable counselors felt discussing SFI options and if they felt it was their role. It also assessed their interest in educational materials or seminars.

Prenatal genetic counselors who were not affiliated with a fetal intervention center were finished with the survey after completing Sections 1-4. Section 5 was only presented to counselors who indicated that they were affiliated with a fetal intervention center in Section 1. Section 5 assessed the role of a genetic counselor who is affiliated with a fetal intervention center.

At the end of section 5, the participants were invited to participate in a brief telephone interview regarding their personal experiences working as prenatal genetic counselors in a fetal intervention setting. The interviews were to be conducted using a semi-structured approach based on an interview guide (Appendix E) comprised of qualitative open-ended questions. The interview guide ensured that the overall interview content would remain consistent between all of the interviews.

**Data Analysis**

We completed statistical analysis of all survey responses using SPSS to determine knowledge level, comfort level and most common practices among participants and to compare the responses from the two groups of prenatal genetic counselors – those who
were affiliated with a fetal intervention center and those who were not. Each participant was given a score for Knowledge Level, Experience Level, and Comfort Level (the scoring is explained below). We then used a one-way analysis of variance (ANOVA) to compare the three scores between groups 1 and 2. We also used correlation analysis to determine if there was a significant relationship between different responses.

**Knowledge level:**

We measured knowledge of SFI by scoring participants’ answers to questions on both SFIs and a selection of fetal anomalies that can be treated with SFI. Table 1 shows the four multiple choice knowledge questions that we asked regarding the technical details of 4 common SFIs – minimally invasive fetal surgery (MIFS), fetoscopic surgery (FSS), open fetal surgery (OFS), and ex utero intrapartum therapy (EXIT) – and the correct responses. All 4 questions had multiple correct answers. Participants were given 1 point for each correct response and lost one point for each incorrect response. For example, minimally invasive fetal surgery had 3 incorrect and 2 correct answers, so the lowest possible score a participant could get was -3 and the highest possible score was 2. If participants indicated that they did not know what the procedure was, they were given a 0 for that question, neither gaining nor losing points for their overall knowledge score.
Table 1. SFI Knowledge Questions

<table>
<thead>
<tr>
<th>Question</th>
<th>Correct Answers</th>
</tr>
</thead>
<tbody>
<tr>
<td>Below you will find a list of statements about fetal surgery. Please indicate which of the following statements apply to MINIMALLY INVASIVE FETAL SURGERY. (Select all that apply).</td>
<td></td>
</tr>
<tr>
<td>a) The fetus is observed via ultrasound during the procedure.</td>
<td>X</td>
</tr>
<tr>
<td>b) The fetus is observed via a fiberoptic telescope.</td>
<td></td>
</tr>
<tr>
<td>c) The fetus is exposed.</td>
<td></td>
</tr>
<tr>
<td>d) The pregnancy is allowed to continue to term when the surgery is complete.</td>
<td>X</td>
</tr>
<tr>
<td>e) The fetus is required to be delivered by cesarian section.</td>
<td></td>
</tr>
<tr>
<td>f) I do not know what this procedure is.</td>
<td>X</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Question</th>
<th>Correct Answers</th>
</tr>
</thead>
<tbody>
<tr>
<td>Below you will find a list of statements about fetal surgery. Please indicate which of the following statements apply to FETOSCOPIC SURGERY. (Select all that apply).</td>
<td></td>
</tr>
<tr>
<td>a) The fetus is observed via ultrasound during the procedure.</td>
<td>X</td>
</tr>
<tr>
<td>b) The fetus is observed via a fiberoptic telescope.</td>
<td></td>
</tr>
<tr>
<td>c) The fetus is exposed.</td>
<td></td>
</tr>
<tr>
<td>d) The pregnancy is allowed to continue to term when the surgery is complete.</td>
<td>X</td>
</tr>
<tr>
<td>e) The fetus is required to be delivered by cesarian section.</td>
<td></td>
</tr>
<tr>
<td>f) I do not know what this procedure is.</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Question</th>
<th>Correct Answers</th>
</tr>
</thead>
<tbody>
<tr>
<td>Below you will find a list of statements about fetal surgery. Please indicate which of the following statements apply to OPEN FETAL SURGERY. (Select all that apply).</td>
<td></td>
</tr>
<tr>
<td>a) The fetus is observed via ultrasound during the procedure.</td>
<td>X</td>
</tr>
<tr>
<td>b) The fetus is observed via a fiberoptic telescope.</td>
<td></td>
</tr>
<tr>
<td>c) The fetus is exposed.</td>
<td></td>
</tr>
<tr>
<td>d) The pregnancy is allowed to continue to term when the surgery is complete.</td>
<td>X</td>
</tr>
<tr>
<td>e) The fetus is required to be delivered by cesarian section.</td>
<td></td>
</tr>
<tr>
<td>f) I do not know what this procedure is.</td>
<td>X</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Question</th>
<th>Correct Answers</th>
</tr>
</thead>
<tbody>
<tr>
<td>Below you will find a list of statements about fetal surgery. Please indicate which of the following statements apply to EX UTERO INTRAPARTUM THERAPY (EXIT). (Select all that apply).</td>
<td></td>
</tr>
<tr>
<td>a) The fetus is observed via ultrasound during the procedure.</td>
<td>X</td>
</tr>
<tr>
<td>b) The fetus is observed via a fiberoptic telescope.</td>
<td></td>
</tr>
<tr>
<td>c) The fetus is exposed.</td>
<td></td>
</tr>
<tr>
<td>d) The pregnancy is allowed to continue to term when the surgery is complete.</td>
<td>X</td>
</tr>
<tr>
<td>e) The fetus is required to be delivered by cesarian section.</td>
<td></td>
</tr>
<tr>
<td>f) I do not know what this procedure is.</td>
<td>X</td>
</tr>
</tbody>
</table>

Once participants answered the four questions on the types of SFI a definition was provided for each surgical procedure to be referred to for the remainder of the survey.

Table 2 shows the definitions that were provided.
We measured knowledge of treatment by SFI by scoring participants’ answers to 2 sets of multiple-choice questions on fetal anomalies and 2 open-ended questions on fetal intervention centers. Table 3 lists the 12 fetal anomalies we addressed along with their incidences in the general population and the SFI used to treat them (Bianchi, 2000). The first set of multiple-choice questions asked participants if they were aware SFI was available for each anomaly. They were given 1 point for each answer of “Yes” and zero for each answer of “No,” with a possible score range of 0-12 points. The second set of multiple choice questions asked participants if they knew which of the four SFIs previously presented was used to treat each anomaly. Participants were given 1 point for each correct answer and lost 1 point for each incorrect answer; if they left the question
blank they were given a zero. Therefore the lowest possible score a participant could get if they answered everything incorrectly was -12, and the highest possible score they could achieve if they knew the correct SFI used to treat each anomaly was 12. For some of the anomalies there was more than one answer accepted.

Table 3. Fetal Anomalies

<table>
<thead>
<tr>
<th>Condition</th>
<th>Incidence</th>
<th>SFI</th>
</tr>
</thead>
<tbody>
<tr>
<td>Myelomeningecele (MMC)/Spina Bifida (SB)</td>
<td>0.41-1.43/1,000 (1,000 commonly quoted)</td>
<td>OFS</td>
</tr>
<tr>
<td>Cervical Teratoma (CT)</td>
<td>Rare (~150 congenital cases described)</td>
<td>EXIT/ OFS</td>
</tr>
<tr>
<td>Congenital high airway obstruction syndrome (CHASOS)</td>
<td>Rare (~16 congenital cases described)</td>
<td>OFS</td>
</tr>
<tr>
<td>Congenital cystic adenomatoid malformation (CCAM)</td>
<td>~1/25,000</td>
<td>MIFS/ OFS</td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome (HLHS)</td>
<td>1/10,000</td>
<td>MIFS</td>
</tr>
<tr>
<td>Congenital diaphragmatic hernia (CDH)</td>
<td>1/3,000-1/5,000</td>
<td>MIFS/ EXIT</td>
</tr>
<tr>
<td>Urinary tract obstruction (UTO)</td>
<td>1/4,000</td>
<td>MIFS</td>
</tr>
<tr>
<td>Sacrococcygeal Teratoma (SCT)</td>
<td>1/35,000-1/40,000</td>
<td>OFS</td>
</tr>
<tr>
<td>Twin-twin transfusion syndrome (TTTS)</td>
<td>1/10,000-9/10,000</td>
<td>FSS</td>
</tr>
<tr>
<td>Twin reversed arterial perfusion syndrome (TRAP)</td>
<td>1/35,000</td>
<td>MIFS/ FSS</td>
</tr>
<tr>
<td>Amniotic band syndrome (ABS)</td>
<td>1/1,200</td>
<td>FSS</td>
</tr>
<tr>
<td>Pleural Effusions (PE)</td>
<td>1/1,500</td>
<td>MIFS</td>
</tr>
</tbody>
</table>

(Data from Fetology, 2000)

The 2 open-ended knowledge questions asked participants to list any fetal intervention centers that they were aware of, first in the area where they practiced and second in the United States in general. One point was awarded for each question if the participant was able to correctly name at least one center.

The total knowledge score was a combination of all the knowledge questions (Table 4). The lowest possible score, given if a participant answered every question incorrectly, was -20; the highest possible score, given if a participant answered every question correctly, was 38.
Table 4. Breakdown of Knowledge Score

<table>
<thead>
<tr>
<th>Knowledge Category</th>
<th>Range</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>min</td>
</tr>
<tr>
<td>MIFS</td>
<td>-3</td>
</tr>
<tr>
<td>FSS</td>
<td>-2</td>
</tr>
<tr>
<td>OFS</td>
<td>-1</td>
</tr>
<tr>
<td>EXIT</td>
<td>-2</td>
</tr>
<tr>
<td>Aware of anomaly</td>
<td>0</td>
</tr>
<tr>
<td>Correct SFI for anomaly</td>
<td>-12</td>
</tr>
<tr>
<td>Aware of local program(s)</td>
<td>0</td>
</tr>
<tr>
<td>Aware of other program(s)</td>
<td>0</td>
</tr>
<tr>
<td>TOTAL knowledge score</td>
<td>-20</td>
</tr>
</tbody>
</table>

**Experience Level:**

We measured participant experience with SFI by giving 1 point for each fetal anomaly the participant had counseled for in regards to SFI. The lowest possible score a participant could get was zero, given if they had never counseled for any of the anomalies listed. The highest score they could get was 12, given if they had experience counseling for each of the 12 anomalies listed.

**Comfort Level:**

We measured comfort with SFI by scoring participants’ responses to 3 sets of Likert scale questions. Table 5 shows the questions asked and the Likert scale used. The questions asked participants how comfortable they were discussing the 12 fetal anomalies in general, SFIs for the 12 fetal anomalies, and each of the four SFIs.

The lowest possible score a participant could get if they answered “1 – Not at all comfortable” for each question was 28, and the highest possible score a participant could get if they answered “4 – Very comfortable” for each question, was 112.
Table 5. Breakdown of Comfort Scores

<table>
<thead>
<tr>
<th>Question Set</th>
<th>Likert Scale</th>
<th>Points min</th>
<th>Points max</th>
</tr>
</thead>
<tbody>
<tr>
<td>How comfortable are you discussing the following fetal anomalies? (n=12)</td>
<td>1-Not at all comfortable, 2-Somewhat uncomfortable, 3-Somewhat comfortable, 4-Very comfortable</td>
<td>12</td>
<td>48</td>
</tr>
<tr>
<td>How comfortable are you discussing the available fetal interventions for the following fetal anomalies? (n=12)</td>
<td>1-Not at all comfortable, 2-Somewhat uncomfortable, 3-Somewhat comfortable, 4-Very comfortable</td>
<td>12</td>
<td>48</td>
</tr>
<tr>
<td>How comfortable are you discussing the following surgical fetal interventions? (n=4)</td>
<td>1-Not at all comfortable, 2-Somewhat uncomfortable, 3-Somewhat comfortable, 4-Very comfortable</td>
<td>4</td>
<td>16</td>
</tr>
</tbody>
</table>

We also asked participants to rate a series of statements (Table 6).

Table 6. Statements on SFI

Please use the scale below to rate the following statements. (n=7)

1-Strongly Disagree
2-Disagree
3-Neither agree nor disagree
4-Agree
5-Strongly agree

Statements:

All parents of a fetus that has been diagnosed with a fetal anomaly that is potentially correctable by surgical fetal intervention should be referred to a genetic counselor.

It is important for a prenatal genetic counselor to recognize a fetal anomaly that is potentially correctable by surgical fetal intervention.

It is important for a prenatal genetic counselor to be knowledgeable about the different surgical fetal interventions that are currently available.

It is important for a prenatal genetic counselor to discuss surgical fetal intervention as an option with all parents of a fetus with an anomaly that is potentially correctable by surgical fetal intervention.

I would be interested in new educational material on surgical fetal intervention.
I would be interested in an online seminar on surgical fetal intervention.
I would be interested in a short educational course on surgical fetal intervention.
II. RESULTS

Demographics.

We received 101 survey responses. We excluded 19 responses from the analysis because a majority of survey questions were unanswered and neither knowledge nor comfort scores could be calculated. Therefore a total of 82 responses were counted. All respondents identified themselves as prenatal genetic counselors who had practiced in a clinical setting for greater than 8 hours a week and had been working as prenatal genetic counselors for longer than 1 year.

Respondents were between 24 and 54 years of age with a mean age of 33.8 years old. Reported year of graduation ranged between 1980 and 2008 with the mean year of graduation at 2000. The majority of respondents reported they had been practicing as prenatal genetic counselors for between 1 and 9 years, with 36.6% (30/82) reporting between 1 and 4 years, and 29.3% (24/82) reporting between 5 and 9 years. The majority of respondents reported counseling patients in a clinical setting between 17 and 40 hours a week, with 25.6% (21/82) reporting 17-24 hours per week, 20.7% (17/82) reporting 25-32 hours per week, and 21.9% (18/82) reporting 33-40 hours per week. The two most frequently reported NSGC Regions of practice were 2 and 4, at 29.6% and 30.6% respectively.

Figures 1 through 4 show how the demographics of the study participants compared to the demographics of the NSGC general membership according to the NSGC Professional Status Survey 2008 Analysis.
Figure 1: Demographics: Years Practicing as a Genetic Counselor

![Bar chart showing years of practice as a genetic counselor.](image1)

Figure 2: Demographics: Year of Graduation from Genetic Counseling Program

![Bar chart showing year of graduation.](image2)
Figure 3. Demographics: Age

![Bar chart showing age demographics for NSGC Membership and Study Participants.](image)

Figure 4. Demographics: Region

![Bar chart showing region demographics for NSGC Membership and Study Participants.](image)
Respondents were divided into two groups based on their answer to the question, “Are you directly involved with fetal intervention services? (In other words do you work in a setting where surgical fetal intervention is offered?).” Of the respondents, 34.1% (28/82) responded, “Yes,” and were classified as “group 1 – fetal intervention counselors” and 65.9% (54/82) responded, “No,” and were classified as “group 2 – general prenatal counselors.” There was no significant difference between groups 1 and 2 for any of the demographic questions.

**Knowledge Scores.**

Table 7 shows the responses to the knowledge questions on four SFIs – minimally invasive fetal surgery (MIFS), fetoscopic surgery (FSS), open fetal surgery (OFS), and ex utero intrapartum therapy (EXIT). Respondents’ total scores for these four questions ranged from -8 to 12. The scores for each individual surgery were:

- Minimally invasive fetal surgery: RANGE -1 to 2, AVERAGE 1.41
- Fetoscopic surgery: RANGE -2 to 3, AVERAGE 1.87
- Open fetal surgery: RANGE -1 to 4, AVERAGE 2.18
- EXIT: RANGE -1 to 3, AVERAGE 1.57.

The SFI that the fewest respondents were familiar with was minimally invasive fetal surgery with 15.2% of respondents reporting that they did not know what the procedure was. A slightly smaller percentage reported that they were not familiar with fetoscopic surgery (14.8%), open fetal surgery (7.4%) and EXIT (13.6%).

We compared knowledge scores for each of the four SFIs between group 1, fetal intervention counselors, and group 2, general prenatal counselors, using ANOVA. We
found a significant difference in score between the two groups for both minimally invasive fetal surgery and fetoscopic surgery ($F (1, 77) = 7.85, p = 0.006; F (1, 79) = 8.31, p = 0.005$). We found a borderline significant difference between scores for EXIT ($F (1, 79) = 3.79, p = 0.055$). We found no significant difference between the two groups for open fetal surgery ($F (1, 79) = 0.98, p = 0.325$). Table 8 summarizes the average scores by group.

Table 7. Responses to SFI Knowledge Questions
The percentage of participants aware that a SFI is available for each fetal anomaly is shown in Figure 5. The responses are split by into group 1, counselors affiliated with a fetal intervention center, and group 2, counselors who are not affiliated with a fetal intervention center. The most common fetal anomalies that participants indicated they were aware a SFI is available for were:

- Myelomeningocele/ spina bifida (100%)
- Twin-twin transfusion syndrome (98.5%)
- Congenital diaphragmatic hernia (83.3%)
- Urinary tract obstruction (88.1%)

Participants were least aware that there are interventions available for:

- Pleural effusions (51.5%)
- Amniotic band syndrome (36.8%)
- Hypoplastic left heart syndrome (32.4%).

Using an ANOVA we found group 1, counselors affiliated with a fetal intervention center, scored significantly higher than group 2, general prenatal counselors, when we
asked participants if they were aware that a SFI was available to treat each fetal anomaly \((F (1,66)=8.08, p=0.006)\). Table 8 summarizes the average scores by group.

Group 1 also scored significantly higher when we asked participants which SFI(s) was used to treat each fetal anomaly \((F (1,99)=4.98, p=0.028)\). Figure 6 shows the distribution of responses for selecting the correct SFI for each anomaly. Most respondents knew the correct available SFI for the following conditions:

- Myelomeningocele/ spina bifida (79.4%)

- Twin reversed arterial perfusion syndrome (67.7%)

- Twin-twin transfusion syndrome (67.7%).

Few respondents knew the correct available SFI for:

- Congenital high airway obstruction syndrome (17.6%)

- Hypoplastic left heart syndrome (5.9%)
Congenital high airway obstruction syndrome (1.5%).

Knowledge scores for the two open-ended questions on participants’ awareness of centers offering fetal intervention services ranged from 0 to 2. We compared the two groups of counselors using ANOVA based on participant’s knowledge of both local and national centers. The majority of counselors affiliated with fetal intervention centers were able to name both a local and national center: 95.7% (22/23) local and 91.3% (21/23) national. Fewer general prenatal counselors were able to name centers: 66.7% (30/45) local and 82.2% (37/45) national. Fetal intervention counselors scored significantly higher than general prenatal counselors on the local level, but not on the national level ($F (1, 66) = 7.70, p = 0.007; F (1, 65) = 0.66, p = 0.419$). The most common answers by both groups combined for local fetal intervention centers were:

- Children’s Hospital of Philadelphia (n=13)
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- Cincinnati Children’s Hospital (n=13)
- Children’s Hospital Boston (n=5)

The most common answers for national fetal intervention centers were:

- Children’s Hospital of Philadelphia (n=32)
- Cincinnati Children’s Hospital (n=21)
- University of California San Francisco Children’s Hospital (n=21)
- Children’s Hospital at Vanderbilt (n=17)

The total knowledge scores for all participants ranged from 4 to 32 points out of a potential range of -20 to 38 (Table 8). Using ANOVA to compare the total knowledge scores between groups, we did not find a significant difference ($F (1,66)=0.57, p=0.453$). When we only took the participants correct answers (positive points) into account we found group 1, fetal intervention counselors, scored marginally higher then group 2, general prenatal counselors ($F (1,44)=4.33, p=0.043$).

Experience with SFI.

The majority of respondents (62.7%) reported that they counsel one or more patients each year regarding SFI, but less than one patient each month. “One or more patients each year” was the most common response by both counselors who are affiliated with a fetal intervention center and counselors who are not. We found no statistical difference between the groups. Thirteen participants reported that they had never counseled a patient regarding SFI; these thirteen participants did not complete section 3 of the survey.
Table 9 shows the fetal anomalies that participants reported having discussed with a patient, specifically with regards to SFI. The four fetal anomalies most frequently counseled for in both groups were:

- Myelomeningocele/ spina bifida
- Congenital diaphragmatic hernia
- Urinary tract obstruction
- Twin-twin transfusion syndrome

The five fetal anomalies with the highest incidence in the general population according to Fetology are:

- Myelomeningocele/ spina bifida (~1/1,000)
- Amniotic band syndrome (1/1,200)
- Pleural effusions (1/1,500)
- Congenital diaphragmatic hernia (1/3-5,000)
- Urinary tract obstruction (1/4,000) (Bianchi, 2000).

The four fetal anomalies participants had counseled for the least were:

- Amniotic band syndrome
- Cervical teratoma
- Congenital high airway obstruction syndrome
- Hypoplastic left heart syndrome

The four fetal anomalies with the lowest incidence in the general population according to Fetology are (Bianchi, 2000):

- Congenital high airway obstruction syndrome (rare; ~16 congenital cases described)
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- Cervical teratoma (rare; ~150 congenital cases described)
- Sacrococcygeal teratoma (1/35-40,000)
- Twin reversed arterial perfusion syndrome (1/35,000)

For each of the 12 fetal anomalies addressed group 1, counselors affiliated with a fetal intervention center, reported experience counseling for them more frequently then group 2, counselors who are not affiliated with a fetal intervention center. Using ANOVA we found a group 1 reported a significantly greater amount of experience counseling for sacrococcygeal teratoma, twin-twin transfusion syndrome, amniotic band syndrome, and pleural effusions ($F(1,65)=5.69, p=0.020; F(1,65)=10.57, p=0.002; F(1,65)=4.37, p=0.041; F(1,65)=4.47, p=0.004$). We found no significant difference between groups for the remaining fetal anomalies.

The experience score overall ranged from 1 to 10 points out of a possible 12 with an average of 3.63. Fetal intervention counselors had an average of 4.29 points with a range of 2 to 10. General prenatal counselors had an average of 3.21 points with a range of 1 to 10. Fetal intervention counselors had a significantly higher total experience score then general prenatal counselors ($F(1,99)=8.88, p=0.004$).
Table 9. Fetal Anomalies Counseled For

<table>
<thead>
<tr>
<th>Question: for which of the following fetal anomalies have you discussed surgical intervention surgery with a patient? (Please check all that you have counseled for)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Responses:</strong></td>
</tr>
<tr>
<td><strong>Overall</strong></td>
</tr>
<tr>
<td>MMC/SB</td>
</tr>
<tr>
<td>CT</td>
</tr>
<tr>
<td>CHAOS</td>
</tr>
<tr>
<td>CCAM</td>
</tr>
<tr>
<td>HLHS</td>
</tr>
<tr>
<td>CDH</td>
</tr>
<tr>
<td>UTO</td>
</tr>
<tr>
<td>SCT*</td>
</tr>
<tr>
<td>TTTS*</td>
</tr>
<tr>
<td>TRAP</td>
</tr>
<tr>
<td>ABS*</td>
</tr>
<tr>
<td>PE*</td>
</tr>
</tbody>
</table>

*Statistically significant difference between groups
Bold Most common responses

SFI Resources.

We asked participants which resources they used to prepare to counsel for SFI. The most common responses in both group 1 and group 2 were:

- Websites (61.4%)
- Other medical professionals (61.4%)
- Written material (59.6 %)
- Other genetic counselors (47.4%)

The most frequently reported websites were those for the MOMS trial and for hospitals where SFI is offered. The other medical professionals most frequently approached, other then genetic counselors, were perinatologists, pediatric surgeons and nurses.

We also asked participants if they offer resources to patients after a genetic counseling session for SFI. Most participants, 91.2%, reported that they do offer resources to patients. The most common responses both group 1 and group 2 were:

- Referral to a maternal fetal medicine specialist/pediatric surgeon (94.3%)
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- Websites (64.2%)
- Referral to a genetic counselor who works directly with fetal intervention (62.3%)
- Written pamphlets (54.7%)

Some of the least frequent responses were:
- Support groups (22.6%)
- Family referrals (5.7%)
- Educational groups or services (1.9%)

Discussion Points for SFI.

Table 10 shows a number of discussion points that may be addressed when counseling for SFI. Participants rated each discussion point on a scale of 1 to 4, from “not at all important” to “very important.” The majority of participants reported that each point was important. There was no significant difference between groups.

Table 10. Importance of Points of Discussion

<table>
<thead>
<tr>
<th>Question: How important do you feel it is to discuss the following when you counsel for surgical fetal intervention?</th>
<th>Not at all important</th>
<th>Somewhat unimportant</th>
<th>Somewhat important</th>
<th>Very important</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient's family history</td>
<td>1.70%</td>
<td>13.00%</td>
<td>56.90%</td>
<td>27.60%</td>
</tr>
<tr>
<td>Patient's personal medical history</td>
<td>0.00%</td>
<td>1.70%</td>
<td>27.60%</td>
<td>70.70%</td>
</tr>
<tr>
<td>The etiology of the fetal anomaly</td>
<td>0.00%</td>
<td>1.70%</td>
<td>13.80%</td>
<td>84.50%</td>
</tr>
<tr>
<td>Any genetic syndromes known to be associated with the fetal anomaly</td>
<td>0.00%</td>
<td>0.00%</td>
<td>3.40%</td>
<td>96.60%</td>
</tr>
<tr>
<td>An overview of the type of fetal surgery that is recommended</td>
<td>0.00%</td>
<td>1.70%</td>
<td>10.30%</td>
<td>87.90%</td>
</tr>
<tr>
<td>The technical details of the fetal surgery</td>
<td>6.90%</td>
<td>6.00%</td>
<td>43.10%</td>
<td>41.40%</td>
</tr>
<tr>
<td>The risk to the fetus</td>
<td>1.70%</td>
<td>6.00%</td>
<td>8.60%</td>
<td>89.70%</td>
</tr>
<tr>
<td>The risk to the mother</td>
<td>1.70%</td>
<td>6.00%</td>
<td>8.60%</td>
<td>89.70%</td>
</tr>
<tr>
<td>The benefits of having fetal surgery vs. postnatal management</td>
<td>1.70%</td>
<td>1.70%</td>
<td>5.20%</td>
<td>91.40%</td>
</tr>
<tr>
<td>Termination options</td>
<td>0.00%</td>
<td>1.70%</td>
<td>19.00%</td>
<td>79.30%</td>
</tr>
<tr>
<td>Adoption</td>
<td>0.00%</td>
<td>17.20%</td>
<td>41.40%</td>
<td>41.40%</td>
</tr>
<tr>
<td>Recurrence risks</td>
<td>0.00%</td>
<td>5.20%</td>
<td>37.90%</td>
<td>56.90%</td>
</tr>
</tbody>
</table>

**Bold** Most common responses
Comfort Scores.

Table 11 shows the most common responses, separated by group, to 3 sets of Likert scale questions assessing participants’ level of comfort with the 12 fetal anomalies and 4 SFIs addressed in this survey. In the first set, participants in both groups expressed that they were either “somewhat comfortable” or “very comfortable” with all of the fetal anomalies except congenital high airway obstruction syndrome and twin reversed arterial perfusion syndrome. Of note these are two of the least common fetal anomalies addressed in this study. For each of the 12 fetal anomalies counselors from group 1, those affiliated with a fetal intervention center, reported an equal or greater level of comfort compared to group 2, counselors who are not affiliated with a fetal intervention center. Using ANOVA we found fetal intervention counselors were significantly more comfortable with congenital high airway obstruction syndrome, hypoplastic left heart syndrome, congenital diaphragmatic hernia, urinary tract obstruction, twin-twin transfusion syndrome, and pleural effusions ($F(1,62)=5.61, p=0.021$; $F(1,62)=5.64, p=0.021$; $F(1,62)=6.81, p=0.011$; $F(1,62)=4.21, p=0.030$; $F(1,62)=4.88, p=0.031$; $F(1,61)=7.64, p=0.008$). Fetal intervention counselors also scored significantly higher on their total fetal anomaly comfort score ($F(1,62)=4.05, p=0.48$).

In the second set of Likert scale questions, participants reported the greatest level of comfort discussing the available SFI for:

- Myelomeningocele/ spina bifida
- Congenital diaphragmatic hernia
- Urinary tract obstruction
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- Twin-twin transfusion syndrome.

Similar to the first set myelomeningocele/ spina bifida, congenital diaphragmatic hernia, and urinary tract obstruction are three of the most common fetal anomalies addressed in this study. For each of the 12 fetal anomalies group 1, fetal intervention counselors, reported an equal or greater level of comfort compared to group 2, general prenatal counselors. Using ANOVA we found group 1 was significantly more comfortable with myelomeningocele/ spina bifida, congenital diaphragmatic hernia, urinary tract obstruction, twin-twin transfusion syndrome, and pleural effusions ($F (1,62)=4.38$, $p=0.040$; $F (1,62)=5.95$, $p=0.018$; $F (1,61)=8.84$, $p=0.004$; $F (1,62)=6.95$, $p=0.011$; $F (1,62)=6.50$, $p=0.013$). There was no significant difference between groups for the overall comfort score in this set.

In the third set, the participants taken as a whole were “somewhat comfortable” discussing each of the four SFIs. The majority of group 1, fetal intervention counselors, reported being “very comfortable” discussing minimally invasive fetal surgery, and the majority of group 2, general prenatal counselors, reported being either “somewhat uncomfortable” or “not at all comfortable” with fetoscopic surgery. Using ANOVA we found group 1 was significantly more comfortable with minimally invasive fetal surgery and fetoscopic surgery ($F (1,62)=10.34$, $p=0.002$; $F (1,61)=6.98$, $p=0.010$). There was no significant difference between groups for the overall comfort score in this set.
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Table 11. Comfort Responses

<table>
<thead>
<tr>
<th>How comfortable are you discussing the following fetal anomalies?</th>
<th>How comfortable are you discussing the available fetal interventions the following fetal anomalies?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Overall</td>
<td>Group 1</td>
</tr>
<tr>
<td>MMC/SR*</td>
<td>4</td>
</tr>
<tr>
<td>CT</td>
<td>3</td>
</tr>
<tr>
<td>CHAOS</td>
<td>2</td>
</tr>
<tr>
<td>CCAM*</td>
<td>3</td>
</tr>
<tr>
<td>HLHS*</td>
<td>4</td>
</tr>
<tr>
<td>CDH*</td>
<td>4</td>
</tr>
<tr>
<td>UTO*</td>
<td>3</td>
</tr>
<tr>
<td>SCT</td>
<td>3</td>
</tr>
<tr>
<td>TTTTS*</td>
<td>3</td>
</tr>
<tr>
<td>TRAP</td>
<td>2</td>
</tr>
<tr>
<td>ABS</td>
<td>4</td>
</tr>
<tr>
<td>PE</td>
<td>3</td>
</tr>
</tbody>
</table>

*Statistically significant difference between groups

The majority of participants reported that they either “agree” or “strongly agree” with each of the 7 statements on SFI presented in section 4 (Table 6). There was no statistically significant difference between group 1 and 2.

Counseling in a Fetal Intervention Setting.

The following results are exclusively from group 1, counselors affiliated with a fetal intervention center. When asked which professionals they work with in a fetal intervention setting, greater than 50% of participants reported working with perinatologists, other genetic counselors, and cardiologists (Figure 7). About 69% reported that they have patients referred to them from other genetic counselors.
When asked which professionals addressed some important discussion points with a patient, the discussion points most commonly allotted for genetic counselors in a fetal intervention setting were:

- The patient’s family and personal medical history
- The etiology of the fetal anomaly
- Possible genetic syndromes associated with the fetal anomaly
- Termination and adoption options
- Recurrence risks.

The discussion points most commonly addressed by a perinatologist in a fetal intervention setting were:

- The recommended fetal surgery, including the technical details of the surgery
- The risks and benefits of the procedure to the fetus
- The risks and benefits of the procedure to the mother.
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Open-Ended Questions.

We asked participants who were affiliated with a fetal intervention center to summarize their perception of the role of genetic counselors in SFI. Thirteen participants responded. Their quotes regarding the current roles of genetic counselors in a fetal intervention setting are listed in Table 12. There were not enough responses to find any statistically significant themes. We also gave participants the chance to state anything else that they felt should have been addressed in the survey. None had any suggestions.

Table 12. Participant’s views on the Role of Genetic Counselors in SFI

<table>
<thead>
<tr>
<th>Responses</th>
</tr>
</thead>
<tbody>
<tr>
<td>To fully discuss the pros/cons and all options</td>
</tr>
<tr>
<td>Very center specific, team player with other specialties to provide the genetic background and information about testing</td>
</tr>
<tr>
<td>Contact and support for patient, translating complex medical information into more patient-friendly terms, organization, ensuring that appropriate testing is being performed and that the patient is aware of other potential genetic risks</td>
</tr>
<tr>
<td>To discuss the fetal anomaly and any potential recurrence risks</td>
</tr>
<tr>
<td>Sometimes overlooked</td>
</tr>
<tr>
<td>It is different at every center and ever changing. When I first began here, I was more of a coordinator. Now I am more of a member of the team, seeing patients as a counselor</td>
</tr>
<tr>
<td>Depends on the center. Some GC’s do counseling about the procedures and associated risks/benefits. Other places, the MFM discussed the procedures</td>
</tr>
<tr>
<td>Obtain family history, discuss birth defect (etiology, association with chromosomal/genetic anomalies and syndromes), offer genetic testing, offer resources and support to family</td>
</tr>
<tr>
<td>Very similar to our role in other settings - information providers, support resource, aid in decision-making</td>
</tr>
<tr>
<td>Very important</td>
</tr>
<tr>
<td>To make the parents fully aware of the etiology of the birth defect, the recurrence risk, the benefits and limitations of surgical intervention, a resource for the family, allow for referral to other appropriate health care professionals</td>
</tr>
<tr>
<td>I believe the GC should serve as a common denominator among all the specialists in the group so the patient/family has someone to build a relationship/rapport with</td>
</tr>
<tr>
<td>We only do minimally invasive and EXIT on a regular basis. The GC tends to be involved with the minimally invasive, but while we’ve seen the EXIT patients are not involved with the counseling for the procedure as this falls more into the delivery management arena</td>
</tr>
</tbody>
</table>
Education in Fetal Intervention.

When asked where they first learned about SFI participants reported:

- During their genetic counseling training (61.3%)
- After graduate school (46.3%).

When asked to what extent their graduate school program for genetic counseling addressed SFI participants reported:

- Part of a lecture (46.3%)
- Not at all (30.5%).

Nearly half of participants (41.5%) felt that their graduate school program for genetic counseling did not prepare them very well to counsel for SFI.

Figure 8 shows the distribution of the years in which participants graduated from their genetic counseling master’s programs. We performed correlation analysis between the times that a participant reported first learning about SFI and his or her year of graduation. The two variables were significantly related ($r=-0.111$, $p=<0.029$); the more recently a participant graduated the more likely they were to have learned about SFI in their genetic counseling training. We also performed correlation analysis between a participant’s year of graduation and the extent that the participant’s graduate school program taught about SFI. We found a significant correlation ($r=0.361$, $p=<0.001$); the more recently a participant graduated the more their program taught about SFI. There was no significant correlation between when a participant first learned about SFI and their total knowledge or comfort scores.
Interviews.

At the end of the survey, participants who indicated they worked directly in or with a fetal intervention center were asked to email the primary investigator directly upon completion of the survey if they were interested in being interviewed. Our target number of interviews was between two to six. Only one participant volunteered for the interview. As there was not a high enough response, the interview portion of this study was not completed.
IV. DISCUSSION

Assessing the Knowledge of Prenatal Genetic Counselors.

Answering questions about fetal intervention is difficult because it is a fairly new field that is evolving quickly and constantly. Also, it is not easy to determine what the correct response is to a question on SFI as there may be more than one procedure that can be used to treat the same fetal anomaly and different fetal intervention centers may have different approaches. This study demonstrates that prenatal genetic counselors are generally aware of SFI; all 82 respondents had learned about SFI before taking this survey and all but 13 had counseled for SFI at least once before. Additionally, no participants received a negative overall knowledge score. Still, a wide range of knowledge levels was demonstrated.

Of the four types of SFI that this study addressed – minimally invasive fetal surgery, fetoscopic surgery, open fetal surgery, and EXIT – participants were generally most knowledgeable about minimally invasive fetal surgery. There was a greater range of knowledge levels for the other 3 procedures. This could be because minimally invasive fetal surgery is the most common form of SFI. Approximately 85% of all participants were aware of each of the 4 procedures. Interestingly, the most participants reported they were informed of open fetal surgery, which is the SFI done the least often. This could reflect the uniqueness of open fetal surgery, or the media hype and shock factor associated with this highly invasive procedure where the pregnancy is still allowed to
When we asked participants which fetal anomalies could be treated by SFI, the most common responses were myelomeningocele/spina bifida, congenital diaphragmatic hernia, urinary tract obstruction and twin-twin transfusion syndrome. There could be a few different reasons why these fetal anomalies are the most well known. For one, myelomeningocele/spina bifida, congenital diaphragmatic hernia and urinary tract obstruction are three of the most common fetal anomalies addressed in this study and are therefore more likely to be seen in a clinical setting. Additionally, both
myelomeningocele/ spina bifida and congenital diaphragmatic hernia have highly publicized, ongoing clinical trials for SFI options. Finally, the treatment of twin-twin transfusion syndrome by fetoscopic surgery has also received some publicity in recent years.

The fetal anomalies participants were least aware of were pleural effusions, amniotic band syndrome, and hypoplastic left heart syndrome. These are not the least common fetal anomalies addressed in this study. However, the surgical treatment of amniotic band syndrome and pleural effusions is not highly publicized, and SFI for hypoplastic left heart syndrome is fairly new and uncommon.

When we asked participants which SFI was available to treat each of the fetal anomalies, the three for which the most respondents knew the correct answer were myelomeningocele/ spina bifida, twin reversed arterial perfusion syndrome, and twin-twin transfusion syndrome. This could once again be explained in part by the amount of publicity for fetal intervention with these anomalies. The three fetal anomalies for which the least respondents knew the correct available SFI were congenital high airway obstruction syndrome, hypoplastic left heart syndrome, and congenital high airway obstruction syndrome. The numbers could also reflect the frequency at which SFI is performed for these fetal anomalies. There was a notable trend for participants to score low for the same anomalies in each set of knowledge questions addressing fetal anomalies. This is logical, as a participant who did not know that SFI is available to treat a fetal anomaly would not have known which SFI is used for that fetal anomaly.

Participants were given the opportunity to list fetal intervention programs that they were aware of both in their region of practice and on a national level. Only a few of
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over 20 facilities in the United States alone who offer SFI for at least 1 fetal anomaly were listed, but the responses show which programs are well known in the genetic counseling community. It is important for genetic counselors to be aware of fetal intervention options nationally, not just regionally. With the help of the internet patients are increasingly able to identify fetal intervention programs outside of their regions on their own and request information from their counselors.

Assessing the Knowledge of Prenatal Genetic Counselors by Group.

We broke participants into two groups, those prenatal genetic counselors who work in a fetal intervention setting (group 1) and those who do not (group 2). We found that group 1 consistently scored higher on the knowledge questions, though not always with statistical significance.

Fetal intervention counselors, group 1, scored significantly higher than general prenatal counselors, group 2, on knowledge of minimally invasive fetal surgery and fetoscopic surgery, but not open fetal surgery or EXIT. Fetal intervention counselors scored significantly higher overall than general prenatal counselors when asked which fetal anomalies they were aware could be treated with SFI and which SFI was used to treat each fetal anomaly. Still, participants who were affiliated with a fetal intervention center only knew the correct SFI for an average of about 3 fetal anomalies. It is possible that this reflects an increased knowledge only of the fetal anomalies that are treated at the participant’s clinic.

Counselors affiliated with a fetal intervention center were significantly more knowledgeable about fetal intervention programs in their area. However, as every
member of this group of counselors was able to name at least the center they worked with and they only had to name one fetal intervention center to receive a point in this category, this is not surprising. This difference does not indicate a higher awareness of other programs in their area. There was no significant difference between groups for knowledge of programs in the United States in general.

There was not a significant difference between each group’s total knowledge score when both correct and incorrect answers were taken into account. However, the group of counselors who were affiliated with a fetal intervention center, group 1, did significantly better when only correct answers were compared. This implies that fetal intervention counselors are more likely to respond correctly than general prenatal counselors, but just as likely to respond incorrectly. It could reflect the fact that counselors in group 1 often work in fetal intervention centers that treat only a small selection of fetal anomalies or offer only one or two types of SFI. Therefore these counselors are more likely to be knowledgeable about the surgeries and fetal anomalies they work with, but have the same knowledge level as other prenatal counselors for those surgeries and anomalies they do not counsel for regularly.

Assessing the Experience of Prenatal Genetic Counselors.

The majority of respondents in both groups, those who work in a fetal intervention setting and those who do not, reported that they counsel one or more patients each year regarding SFI, but less than one patient each month. It cannot be determined how many patients the respondents counsel for on average each year based on this survey; the potential range is between 1 and 11 patients. There was no statistical
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difference between the groups. This shows that even prenatal genetic counselors who work in a fetal intervention setting are not counseling solely for SFI. Genetic counselors are not always involved when SFI is indicated. Also, SFIs are still rare procedures that are only recommended under very specific conditions where the risk to both the fetus and the mother is not too high and all other options have been considered.

Less than 10% of all respondents reported having counseled a patient regarding SFI for cervical teratoma, congenital high airway obstruction syndrome, hypoplastic left heart syndrome, and amniotic band syndrome. When responses were broken down by group the percentage only exceeded 10% in group 1, fetal intervention counselors, for hypoplastic left heart syndrome. This could be a good indicator of which fetal anomalies are treated with SFI the least often. The fetal anomalies that were most commonly counseled for were myelomeningocele/ spina bifida, twin-twin transfusion syndrome, congenital diaphragmatic hernia, and urinary tract obstruction. This could reflect both a higher frequency of SFI for these anomalies and a higher level of publicity. The group of counselors affiliated with fetal intervention centers scored significantly higher in counseling experience for sacrococcygeal teratoma, twin-twin transfusion syndrome, amniotic band syndrome and pleural effusions. Their total counseling experience score was also significantly higher. Yet, once again, fetal intervention counselors only had experience counseling for SFI with an average of 3 fetal anomalies. This would support the hypothesis that prenatal genetic counselors who work in a fetal intervention setting are only more knowledgeable about the fetal anomalies they have experience with in their clinic.
SFI Resources.

When asked what resources they use to prepare to counsel for SFI, over 50% of participants reported that they use some combination of written materials, websites, and the advise of other medical professionals. There was no significant difference between fetal intervention counselors and general prenatal counselors. Counselors who work in a fetal intervention setting were notably more likely to consult other genetic counselors.

The majority of respondents reported that they offered some kind of resource to their patients after counseling for SFI. Over 50% reported referring patients to one or more of the following: perinatologists, genetic counselors who work directly with fetal intervention, websites, or written pamphlets. Few respondents reported referring patients to the following resources: other families who had been in similar situations, support groups, or educational services. Both the SFIs and the fetal anomalies discussed are very rare, so it is not surprising that family referrals are rare and that few support groups exist to help patients in these particular circumstances. Also, most educational material on SFI is directed at medical professionals. These data show that there is likely a need for patient oriented services, both supportive and educational.

Assessing the Comfort Level of Prenatal Genetic Counselors.

The majority of all participants expressed that they were either “somewhat comfortable” or “very comfortable” counseling for all of the fetal anomalies except congenital high airway obstruction syndrome and twin reversed arterial perfusion syndrome. Both of these fetal anomalies are very rare; only ~16 cases of congenital high airway obstruction syndrome have ever been reported congenitally and twin reversed
arterial perfusion syndrome has an incidence of 1/35,000. These were not the only rare anomalies discussed in this study though. Counselors affiliated with a fetal intervention center, were statistically more comfortable counseling for congenital high airway obstruction syndrome, hypoplastic left heart syndrome, congenital diaphragmatic hernia, urinary tract obstruction, twin-twin transfusion syndrome, and pleural effusions. This could indicate that some of these anomalies are more frequently referred to fetal intervention services than the others.

When asked to rate their comfort level discussing available SFI for each anomaly, fetal intervention counselors reported being on average “somewhat comfortable” with nine and “very comfortable” with one, urinary tract obstruction. The majority of the general prenatal counselors were only “somewhat comfortable” with four anomalies, and they were not “very comfortable” with any. Both groups were “very uncomfortable” with hypoplastic left heart syndrome and amniotic band syndrome. The overlap in comfort levels between groups shows which fetal anomalies are best known in the context of SFI (myelomeningocele/ spina bifida, congenital diaphragmatic hernia, urinary tract obstruction, and twin-twin transfusion syndrome) and which fetal anomalies are not (hypoplastic left heart syndrome and amniotic band syndrome).

Most respondents reported being on average “somewhat comfortable” discussing open fetal surgery and EXIT. There was a significant difference between the groups’ comfort levels discussing minimally invasive fetal surgery and fetoscopic surgery. In each case, counselors affiliated with fetal intervention centers were significantly more comfortable than those who were not. Fetoscopic surgery was the only SFI that general prenatal counselors were uncomfortable with. However, these self-reported comfort
levels were not consistent with the wide range of knowledge levels found when participants were questioned on the details of each SFI.

Group 1, fetal intervention counselors, scored significantly higher than group 2, general prenatal counselors, for total comfort scores when discussing the 12 fetal anomalies in general, but no significant difference was found for total comfort discussing the 4 SFIs or the SFI options for each fetal anomaly. This would support the conclusion that prenatal genetic counselors who work in a fetal intervention setting are most comfortable counseling about the anomalies and SFIs that they encounter in their clinical practice.

Counseling in a Fetal Intervention Setting.

In general, centers that offer SFI have a multidisciplinary approach. Participants who were affiliated with a fetal intervention center reported working with other genetic counselors, general obstetricians, perinatologists, general surgeons, geneticists, radiologist, cardiologists, and nurses. Of all these options, the most frequently reported professionals were perinatologists, other genetic counselors, and cardiologists.

When asked which professionals addressed the discussion points first listed in the experience section of the survey (Table 10) that affect a patient’s decision whether or not to pursue SFI, the most frequent response was always either a genetic counselor or a perinatologist. Genetic counselors most often discuss the patient’s family and medical history, as well as the relevant genetic information related to the fetal anomaly and alternative options for the pregnancy. Notably, these are points that prenatal genetic counselors frequently discuss whether or not SFI is being considered. Discussion of the
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SFI itself, including risks, benefits, and technical details is reportedly most often conducted by the perinatologist.

**Role of Genetic Counselors in SFI.**

When we asked participants to summarize what they felt the role of genetic counselors was in SFI, there were a number of roles mentioned. Three participants stated that the genetic counselor’s role depends on the center where they work, for example, “It is different at every center and ever-changing.” One genetic counselor reinforced the conclusion that counseling in a fetal intervention setting is comparable to most other genetic counseling positions when they summarized their role as, “very similar to in other settings – information providers, support resource, aide in decision-making.” Multiple respondents mentioned discussing information about the fetal anomaly as well as genetic testing risks and options. The most frequently mentioned point was being a resource and support for the patient. Two respondents also stated that discussion of the surgeries is not typically the genetic counselor’s role.

Regardless of which group they fell into, a majority of participants also indicated they agreed that it is important for prenatal genetic counselors to be knowledgeable about the different SFIs that are currently available. Participants indicated that they felt it was important for a prenatal genetic counselor to discuss SFI as an option with all parents of a fetus with an anomaly that is potentially correctable by SFI. Also, a majority of patients reported that they “strongly agreed” with a statement that “it is important for a prenatal genetic counselor to recognize a fetal anomaly that is potentially correctable by SFI.”
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Education in Fetal Intervention.

About 60% of participants reported that they first learned of SFI in their genetic counseling training, but about 40% also reported that they did not feel that their genetic counseling master’s program prepared them well to counsel for SFI. The majority of respondents reported that SFI was only addressed in part of one lecture in their training, and about 30% indicated that it was not addressed at all. These facts indicate that genetic counselors’ knowledge about SFI is currently gained primarily from experiences after graduate school, even when it is first mentioned during their training.

There was a significant relationship between participants’ years of graduation from their genetic counseling master’s programs and both when they first learned about SFI and also the extent to which their program taught SFI. We found that the more recently a participant graduated, the more likely they were to have learned of SFI during their training and the more time their program likely spent teaching about SFI. This is logical as SFI is still a fairly new field that is continuously changing and growing.

Limitations.

The small number of participants (n=82) in this study made finding statistically significant comparisons and correlations difficult. This limitation became more evident when the total number of participants was divided into two groups (n=28 and 54), and made comparing responses based on participant’s region of practice impossible. There could be differences in awareness and knowledge of fetal intervention depending on where a genetic counselor practices. Additionally, some of the statistical analysis could
have been affected by the difference in group sizes, with group 2, general prenatal counselors, having twice as many respondents as group 1, fetal intervention counselors.

Given the study’s title, which was posted on both recruitment notices, the participants were most likely genetic counselors who had heard of SFI before, and had some degree of interest in the subject. Consequently, it is possible that this group does not accurately represent the knowledge, experience, and comfort levels of all prenatal genetic counselors. Also, the titles of the four SFIs – minimally invasive fetal surgery, fetoscopic surgery, open fetal surgery, and ex utero intrapartum therapy – are fairly descriptive of the procedures themselves and could have lead to an over estimation of participant knowledge. Also, in the knowledge section when asking which SFI was used to treat each anomaly, the survey only allowed participants to select one answer. For a number of the fetal anomalies however more than one SFI would have been correct. While multiple correct answers were accepted, counselors could have been confused when they were not allowed to choose all of the correct answers.

The question, “Are you directly involved with fetal intervention services? (In other words do you work in a setting where surgical fetal intervention is offered?),” has a lot of room for interpretation. For example, neither involvement nor fetal intervention setting were well defined. A participant was classified as either group 1 or group 2 based on their answer to this question. Finally, not all women carrying a fetus with an anomaly that is treatable with SFI see a genetic counselor. Therefore this study reflects the genetic counselor’s experience with SFI, but not the patient’s.
Future Directions.

Future studies could attempt to collect a greater number of responses for more accurate and detailed results. Genetic counselors who work with fetal intervention are still a small group, but as the field grows more genetic counselors are likely to work with SFI. Studies could also assess the practices of perinatologists with SFI or the experiences of patients. Plus, a majority of participants reported that they would be very interested in new educational materials on SFI as well as an online seminar or short course. There are also a limited number of supportive and educational resources available for the patient, and a low level of awareness about what is available. Support groups and educational material such as patient friendly websites could be developed with information available at fetal intervention centers.
V. CONCLUSION

The role of genetic counseling in a fetal intervention setting appears to be the same as it is in most other genetic counseling settings. Fetal intervention counselors report discussing familiar points such as the patient’s family and medical history, as well as the genetic information related to the fetal anomaly and alternative options for the pregnancy. Multiple respondents mentioned discussing risks and options for genetic testing, and the most frequently mentioned role of a fetal intervention counselor was being a resource and support for the patient.

The prenatal genetic counselors who completed this survey are generally aware of SFI as an option for their patients as well as a selection of the fetal anomalies that can be treated with SFI. Most counselors in both fetal intervention and general prenatal settings have some amount of experience counseling for SFI. In fact, the majority of both groups counsel between one and eleven patients a year. This shows that even prenatal genetic counselors who work in a fetal intervention setting are not counseling solely for SFI. It also shows that since both groups of counselors encounter SFI equally as often in their clinical practice, it is equally important for both groups to be knowledgeable about the topic.

Fetal intervention counselors only had experience counseling on SFI for an average of 4 fetal anomalies. It seems that working in a setting where fetal intervention is offered often only exposes a counselor to a portion of SFI techniques and anomalies.
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Also, though they had higher overall levels of knowledge as well as experience, fetal intervention counselors only had increased knowledge for an average of 3 fetal anomalies. Many fetal intervention centers only treat a limited number of fetal anomalies and only offer one or two types of SFI. A possible explanation is that counselors who work in fetal intervention setting are most knowledgeable about the SFIs used in their center and the fetal anomalies treated in their center. If this is the case, then it seems that genetic counselors are currently gaining their knowledge of SFI primarily through clinical experience.

Further evidence of this potential explanation comes from the two groups total knowledge scores. Fetal intervention counselors are more likely to respond correctly to knowledge questions than general prenatal counselors, but just as likely to respond incorrectly. It could indicate that counselors who work in a fetal intervention setting are more knowledgeable about SFIs and fetal anomalies that they work with, but that they know just as much, or as little, as general prenatal counselors concerning any other SFIs and fetal anomalies.

The more recently a participant graduated from their genetic counseling master’s programs, the more likely they were to have learned of SFI during their training and the more time their program likely spent teaching about SFI. Still, the majority of respondents reported that SFI was only addressed in part of one lecture in their genetic training, and about 30% indicated that it was not addressed at all. This supports the conclusion that prenatal genetic counselors currently seem to gain most of their knowledge on SFI though their experiences after graduating from their genetic counseling programs. This indicates that though genetic counseling graduate programs
are beginning to address SFI more frequently, they still need to increase the extent to which they teach about SFI. It also indicates a need for more educational materials and learning opportunities for practicing genetic counselors.

Similarly, there is a need for more educational opportunities and resources for patients who are considering SFI. Given that both the SFIs and the fetal anomalies addressed in this study are very rare, it is not surprising that family referrals are rare and that few support groups exist to help patients. These data show that there is likely a need for patient oriented services, both supportive and educational. An important part of genetic counseling is acting as a support for patients, and therefore genetic counselors would be well suited to helping create and provide resources for parents who are considering SFI.
REFERENCES


Appendix A: Recruitment Notice A

Are You A Prenatal Genetic Counselor?

I am a graduate student in the Brandeis University Genetic Counseling Program, and am seeking volunteers to take part in a research project exploring prenatal genetic counselors’ experiences with and opinions regarding surgical fetal intervention.

You are eligible to participate if you:

• Have worked for at least 1 year as a genetic counselor in a clinical prenatal setting
• Currently see prenatal patients at least one full day a week (or the equivalent of one day per week)

The study involves an online, anonymous survey that will take approximately 30 minutes of your time.

If you are interested, please click the link below. Thank you for your time!

http://www.surveymonkey.com/s.aspx?sm=v0CmljbqATSmGEFVpqieQ_3d_3d

Sincerely,

Caitlin Melley

Brandeis University
Genetic Counseling Masters Candidate
Appendix B: Recruitment Notice B

Are you a prenatal genetic counselor who works in or with a center that offers fetal intervention?

I am a graduate student in the Brandeis University Genetic Counseling Program, and I am seeking volunteers to take part in a research project exploring the experiences of prenatal genetic counselors’ who work directly with fetal intervention. The project focuses on surgical fetal interventions.

You are eligible to participate if you:

• Have worked for at least 1 year as a genetic counselor in a clinical prenatal setting
• Currently see prenatal patients at least one full day a week (or the equivalent of 1 full day per week)
• Currently work in or with a center that offers fetal intervention services.

The study involves an online, anonymous survey that will take approximately 30 minutes of your time. Respondents will also have the option to participate in a brief telephone interview. All interviews will be kept confidential and each interview participant will receive a $25 gift card to Amazon.com as compensation.

If you are interested, please click the link below. Thank you for your time!

http://www.surveymonkey.com/s.aspx?sm=v0CmljbqATSvmGEFVpqieQ_3d_3d

Sincerely,
Caitlin Melley
Brandeis University
Genetic Counseling Masters Candidate
Appendix C: Survey Document

Dear Genetic Counselor,

Thank you very much for agreeing to take part in this research study.

This study is open to prenatal genetic counselors currently working in a clinical setting. Participants must have at least one full year of clinical experience in the prenatal setting and see patients at least one full day a week (or the equivalent of one full day).

Your participation is completely voluntary. You may choose to skip any questions or stop taking the survey at any time. The answers that you provide will remain anonymous and confidential. The procedures in place to ensure your confidentiality have been approved by the Brandeis University Human Subjects Review Board.

The purpose of this research study is to assess knowledge and current practices regarding surgical fetal intervention in a prenatal genetic counseling setting. You will be asked questions related to the following topics:

- Your general knowledge about surgical fetal intervention
- Your clinical experiences with specific genetic counseling situations
- Your comfort level with specific genetic counseling situations

If you have any questions, please do not hesitate to contact me at camelley@brandeis.edu.

Sincerely,

Caitlin Melley

MS Candidate, Genetic Counseling
Brandeis University
Surgical Fetal Intervention: Assessing the Current Practices of Genetic Counselors  
Principal Investigator: Caitlin Melley

Section 1. Background Information

1) Are you currently employed as a prenatal genetic counselor?
   a) Yes.
   b) No.

(if a participant answers “No” to this question they will be redirected to a page thanking them for their time and informing them that they are not currently eligible to participate in this study)

2) What year did you graduate from your genetic counseling master’s program?_______

3) How many years have you been practicing as a prenatal genetic counselor?
   a) Less than 1 year.
   b) 1-4 years.
   c) 5-9 years.
   d) 10-14 years.
   e) 15-19 years.
   f) More then 19 years.

(if a participant answers “Less than 1 year” to this question they will be redirected to a page thanking them for their time and informing them that they are not currently eligible to participate in this study)

4) How much time do you spend counseling patients in a clinical setting each week?
   a) Fewer than 8 hours.
   b) 8-16 hours.
   c) 17-24 hours.
   d) 25-32 hours.
   e) 33-40 hours.
   f) Greater than 40 hours.

(if a participant answers “Fewer than 8 hours” to this question they will be redirected to a page thanking them for their time and informing them that they are not currently eligible to participate in this study)

5) How old are you currently?
   20-24, 25-29, 30-34, 35-39… 70-74, 75+

6) What state do you currently practice in?
   AL, AK, AZ, AR, CA… WI, WY
7) Are you directly involved with fetal intervention services? (Do you work in a setting where surgical fetal intervention is offered?)
   a) Yes.
   b) No.

(if a participant answers “no” to this question they will be directed to the version of the survey without section 5; if a participant answers “yes” to this question they will be directed to the version of the survey with section 5. In both versions of the survey sections 1-4 will be identical)

[end of page]
Section 2. General knowledge about surgical fetal intervention

1) How did you first learn about fetal intervention surgery?
   a) During undergraduate education.
   b) During genetic counseling training.
   c) During a presentation at a professional meeting.
   d) Through genetic counseling clinical practice.
   e) From a journal article.
   d) From a patient.
   e) From a personal family member/friend.
f) Other: _____________

2) To what extent did your graduate school program for genetic counseling address surgical fetal intervention?
   a) Not at all.
   b) Part of a lecture.
   c) 1 full lecture.
   d) 2 or more lectures.
   e) 1 full course (semester).
   f) More then 1 full course.

3) How well do you think your graduate school program prepared you to counsel for surgical fetal intervention?
   a) Very well
   b) Somewhat well
   c) Not very well
   d) Not at all.
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4) Below you will find a list of statements about fetal surgery. Please indicate which of the following statements apply to MINIMALLY INVASIVE FETAL SURGERY. (Select all that apply).

<table>
<thead>
<tr>
<th>Statement</th>
<th>answer</th>
</tr>
</thead>
<tbody>
<tr>
<td>The fetus is observed via ultrasound during the procedure.</td>
<td>X</td>
</tr>
<tr>
<td>The fetus is observed via a fiberoptic telescope.</td>
<td></td>
</tr>
<tr>
<td>The fetus is exposed.</td>
<td></td>
</tr>
<tr>
<td>The pregnancy is allowed to continue to term when the surgery is complete.</td>
<td>X</td>
</tr>
<tr>
<td>The fetus is required to be delivered by caesarian section.</td>
<td></td>
</tr>
<tr>
<td>I do not know what this procedure is.</td>
<td></td>
</tr>
</tbody>
</table>

5) Below you will find a list of statements about fetal surgery. Please indicate which of the following statements apply to FETOSCOPIC SURGERY. (Select all that apply).

<table>
<thead>
<tr>
<th>Statement</th>
<th>answer</th>
</tr>
</thead>
<tbody>
<tr>
<td>The fetus is observed via ultrasound during the procedure.</td>
<td>X</td>
</tr>
<tr>
<td>The fetus is observed via a fiberoptic telescope.</td>
<td>X</td>
</tr>
<tr>
<td>The fetus is exposed.</td>
<td></td>
</tr>
<tr>
<td>The pregnancy is allowed to continue to term when the surgery is complete.</td>
<td>X</td>
</tr>
<tr>
<td>The fetus is required to be delivered by caesarian section.</td>
<td></td>
</tr>
<tr>
<td>I do not know what this procedure is.</td>
<td></td>
</tr>
</tbody>
</table>

6) Below you will find a list of statements about fetal surgery. Please indicate which of the following statements apply to OPEN FETAL SURGERY. (Select all that apply).

<table>
<thead>
<tr>
<th>Statement</th>
<th>answer</th>
</tr>
</thead>
<tbody>
<tr>
<td>The fetus is observed via ultrasound during the procedure.</td>
<td>X</td>
</tr>
<tr>
<td>The fetus is observed via a fiberoptic telescope.</td>
<td></td>
</tr>
<tr>
<td>The fetus is exposed.</td>
<td>X</td>
</tr>
<tr>
<td>The pregnancy is allowed to continue to term when the surgery is complete.</td>
<td>X</td>
</tr>
<tr>
<td>The fetus is required to be delivered by caesarian section.</td>
<td>X</td>
</tr>
<tr>
<td>I do not know what this procedure is.</td>
<td></td>
</tr>
</tbody>
</table>
7) Below you will find a list of statements about fetal surgery. Please indicate which of the following statements apply to EX UTERO INTRAPARTUM THERAPY (EXIT). (Select all that apply).

<table>
<thead>
<tr>
<th>Statement</th>
<th>Answer</th>
</tr>
</thead>
<tbody>
<tr>
<td>The fetus is observed via ultrasound during the procedure.</td>
<td>X</td>
</tr>
<tr>
<td>The fetus is observed via a fiberoptic telescope.</td>
<td></td>
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<tr>
<td>The fetus is exposed.</td>
<td>X</td>
</tr>
<tr>
<td>The pregnancy is allowed to continue to term when the surgery is complete.</td>
<td></td>
</tr>
<tr>
<td>The fetus is required to be delivered by caesarian section.</td>
<td>X</td>
</tr>
<tr>
<td>I do not know what this procedure is.</td>
<td></td>
</tr>
</tbody>
</table>

[end of page]
Section 2. General knowledge about surgical fetal intervention (continued):

***From this point forward please use the following definitions:

Minimally invasive fetal surgery: An ultrasound guided intervention in which access to the fetus is gained via needle-puncture of the maternal abdomen and uterine wall. Minimally invasive fetal surgery is used for procedures such as amnio-reduction, radio frequency ablation, or inserting a fetal shunt to drain a fluid filled structure.

Fetoscopic surgery: An intervention in which a fiberoptic telescope (small camera) is inserted through the mother’s abdominal wall and the uterus to view the fetus. The fetus is treated using small surgical instruments that are either part of the fiberoptic telescope or part of a second thin tube. Fetoscopic surgery is generally done under ultrasound guidance.

Open fetal surgery: A fetal intervention in which, under ultrasound guidance, an incision is made through the mother’s abdominal wall (laparotomy) to expose the uterus and into the uterus (hysterotomy) to expose the fetal surgical area. The fetal surgeon repairs the defect in the fetus, replaces the amniotic fluid and closes the incisions.

Ex utero intrapartum therapy (EXIT): A fetal intervention in which the fetus is partially delivered via caesarian-section while remaining attached to the umbilical chord to receive oxygen from the placenta. This allows for surgical intervention to maximize stabilization and/or more controlled intubation. Once surgery is completed the neonate is fully delivered.
7) Prior to completing this survey, were you aware that fetal intervention surgery was available for the following fetal anomalies?

   a) Yes.  
   b) No.  
   c) I am not familiar with this fetal anomaly.

<table>
<thead>
<tr>
<th>Fetal Anomaly</th>
<th>a)</th>
<th>b)</th>
<th>c)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Myelomeningocele (MMC)/Spina Bifida (SB)</td>
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<td></td>
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<tr>
<td>Cervical Teratoma (CT)</td>
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<td></td>
<td></td>
</tr>
<tr>
<td>Congenital high airway obstruction syndrome (CHAOS)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Congenital cystic adenomatoid malformation (CCAM)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome (HLHS)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Congenital diaphragmatic hernia (CDH)</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Urinary tract obstruction</td>
<td></td>
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<tr>
<td>Sacrococcygeal Teratoma (SCT)</td>
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<tr>
<td>Twin-twin transfusion syndrome (TTTS)</td>
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<td>Twin reversed arterial perfusion syndrome (TRAP)</td>
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<tr>
<td>Amniotic band syndrome (ABS)</td>
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<tr>
<td>Pleural Effusions (PE)</td>
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<td></td>
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</tr>
</tbody>
</table>

8) Do you know of any other fetal anomalies for which fetal intervention surgery is available?

   a) Yes.  
   b) No.  
   If you answered yes, please name the fetal anomalies: ________________
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9) Which surgical fetal intervention is currently available to correct the following fetal anomalies? (Please select all that apply.)

   a) Minimally invasive fetal surgery.
   b) Fetoscopic surgery.
   c) Open fetal surgery.
   d) Ex utero intrapartum therapy (EXIT).
   e) This fetal anomaly cannot be treated by surgical fetal intervention.
   f) I do not know.

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>a</th>
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10) Are you aware of any fetal intervention programs in your area?
   a) Yes.
   b) No.
   If you answered yes, please list the programs: ________________

11) Are there any other fetal intervention centers that you are aware of (in the United States)?
   a) Yes.
   b) No.
   If you answered yes, please list the programs: ________________
Section 3. Experience Counseling for Surgical Fetal Intervention

1) How often do you counsel patients regarding surgical fetal intervention? (Please choose the answer that best describes your practice)

   a) I have never counseled a patient for surgical fetal intervention.
   b) 1 or more patients each year.
   c) 1 or more patients each month.
   d) 1 or more patients each week.
   e) 1 or more patients each day that I counsel.
   f) I only counsel patients who are considering surgical fetal intervention.

2) For which of the following fetal anomalies have you discussed fetal intervention surgery with a patient? (Please check all that you have counseled for)

   __Myelomeningocele (MMC)/Spina Bifida (SB)
   __Cervical Teratoma (CT)
   __Congenital high airway obstruction syndrome (CHAOS)
   __Congenital cystic adenomatoid malformation (CCAM)
   __Congenital diaphragmatic hernia (CDH)
   __Urinary tract obstruction
   __Sacroccyggeal Teratoma (SCT)
   __Twin-twin transfusion syndrome (TTTS)
   __Twin reversed arterial perfusion syndrome
   __Amniotic band syndrome

3) What resources do you currently use when preparing to counsel for surgical fetal intervention? (Please check all that apply)

   a) Written material.
   b) Book(s). (please specify below)
   c) Website(s). (please specify below)
   d) Other genetic counselors.
   e) Other medical professionals (please specify below)
   f) Other. (please specify below)
Surgical Fetal Intervention: Assessing the Current Practices of Genetic Counselors
Principal Investigator: Caitlin Melley

4) How important do you feel it is to discuss the following when you counsel for surgical fetal intervention?

1-Very important
2-Somewhat important
3-Somewhat unimportant
4-Not at all important

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<td>Recurrence risks</td>
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5) Do you offer any resources for your patients to refer to after the session?
   a) Yes.
   b) No.

6) If you answered yes to question 5, what resources do you offer? (please check all that apply)
   a) Referral to a genetic counselor who works directly with fetal intervention.
   b) Referral to a maternal fetal medicine specialist/ pediatric surgeon who performs surgical fetal interventions.
   c) Written pamphlet(s).
   d) Website(s). *(please specify below)*
   e) Book(s). *(please specify below)*
   f) Family referral.
   g) Support group(s).
   h) Educational groups/services.
   i) Other. *(please specify below)*

   [end of page]
Surgical Fetal Intervention: Assessing the Current Practices of Genetic Counselors
Principal Investigator: Caitlin Melley

Section 4. Comfort Level Counseling for Surgical Fetal Intervention:

1) How comfortable are you discussing the following fetal anomalies?

1-Not at all comfortable  
2-Somewhat uncomfortable  
3-Somewhat comfortable  
4-Very comfortable

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2) How comfortable are you discussing the available fetal interventions for the following fetal anomalies?

1-Not at all comfortable  
2-Somewhat uncomfortable  
3-Somewhat comfortable  
4-Very comfortable

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<th>Intervention</th>
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<td>Pleural Effusions (PE)</td>
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</table>
3) How comfortable are you discussing the following surgical fetal interventions?

1-Not at all comfortable
2-Somewhat uncomfortable
3-Somewhat comfortable
4-Very comfortable

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<tr>
<td>Minimally invasive fetal surgery</td>
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<td>Fetoscopic surgery</td>
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<td>Open fetal surgery</td>
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<td>Ex utero intrapartum therapy (EXIT)</td>
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4) Please use the scale below to rate the following statements.

1-Strongly Disagree
2-Disagree
3-Neither Agree Nor Disagree
4-Agree
5-Strongly Agree

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<tr>
<td>All parents of a fetus that has been diagnosed with a fetal anomaly that is potentially correctable by surgical fetal intervention should be referred to a genetic counselor.</td>
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<td>It is important for a prenatal genetic counselor to recognize a fetal anomaly that is potentially correctable by surgical fetal intervention.</td>
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<td>It is important for a prenatal genetic counselor to be knowledgeable about the different surgical fetal interventions that are currently available.</td>
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<tr>
<td>It is important for a prenatal genetic counselor to discuss surgical fetal intervention as an option with all parents of a fetus with an anomaly that is potentially correctable by surgical fetal intervention.</td>
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<td>I would be interested in new educational material on surgical fetal intervention.</td>
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<td>I would be interested in an online seminar on surgical fetal intervention.</td>
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<td>I would be interested in a short educational course on surgical fetal intervention.</td>
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Section 5. Counseling Practices of Genetic Counselors in a Fetal Intervention Setting
(This section is only being presented to those counselors who are directly involved in fetal intervention)

1) Do you have patients referred to you from other genetic counselors?
   a) Yes.
   b) No.

2) Surgical fetal intervention generally has a multi-disciplinary approach. Which of the following professionals do you work with to provide fetal intervention care? (please check all that apply)
   __Other Genetic Counselors
   __General Obstetrician
   __Perinatologist
   __General Surgeon
   __Geneticist
   __Radiologist
   __Cardiologist
   __Nurse(s)
   __Other:_____________
3) In your practice, who typically discusses the following with the patient?

   a) Genetic counselor(s)
   b) General Obstetrician
   c) Perinatologist
   d) General Surgeon
   e) Geneticist
   f) Nurse(s)
   g) Other

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<td>The etiology of the fetal anomaly</td>
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<td>Any genetic syndromes known to be associated with the fetal anomaly</td>
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<td>An overview of the type of fetal surgery that is recommended</td>
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<td>The risk to the fetus</td>
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4) Please summarize what you feel is the current role of genetic counseling in surgical fetal intervention?

_____________________________________

5) Please summarize what you feel should be the role of genetic counseling in surgical fetal intervention? (If you believe the current role of genetic counseling in surgical fetal intervention is consistent with what it should be please write “see question 3”)

_____________________________________

6) Is there anything else that you feel should have been addressed in this survey?

_____________________________________

[end of page]
Section 5. Counseling Practices of Genetic Counselors in a Fetal Intervention Setting (continued):

Thank you for taking the time to complete the survey.

At this point I would like to invite you to participate in a telephone interview to discuss your role as a genetic counselor in a fetal intervention setting in more detail.

The interview will last approximately 30 minutes, and participation is completely confidential and voluntary. Your interview will not be linked in any way to your survey answers. Each participant will receive a $25 gift certificate to Amazon.com upon completion of the interview.

I plan to conduct a total of 6 interviews. If more then 6 genetic counselors express an interest in being interviewed, then I will randomly select 6 individuals to interview.

If you are interested in being interviewed, please email me at camelley@brandeis.edu.

[end of page]
Thank you for your time. Your participation is greatly appreciated.

If you have any questions regarding this study, please do not hesitate to contact me at camelley@brandeis.edu.

Sincerely,
Caitlin Melley

MS Candidate, Genetic Counseling
Brandeis University
Appendix D: Interview Guide

I. Introduction:

Thank you for volunteering to participate in this interview.

Your participation is completely voluntary and anonymous. To protect your privacy I will address you using a study ID number in place of your name. I am going to audiotape this interview so that I can focus on our discussion rather than note taking.

The goal of this interview is to find out about your experiences working with surgical fetal intervention. I will be asking you about your work and your opinions on genetic counseling in a fetal intervention setting. If at any point there is a question that you would rather not answer, please let me know.

II. Interview Questions:

1) Could you briefly describe the setting you work in?

2) What is a typical day at work like for you?

3) As a genetic counselor, are there certain skills or perspectives you have that you feel are unique and distinct from other members of the surgical intervention team?

4) How would you describe your role with parents who are considering surgical fetal intervention?

5) Could you please describe a typical counseling session for surgical intervention?

6) Could you tell me about a particularly difficult case you counseled for?

7) Do you feel that all parents considering a surgical fetal intervention should be required to meet with a genetic counselor? Why/why not?
8) How do you see the role of genetic counseling for surgical fetal intervention changing in the future as this field evolves?

III. Closing:

Thank you so much for sharing your experiences and opinions with me. Is there anything you would like to add?
Appendix E: IRB Study Protocol

STUDY PURPOSE

Over the past 45 years surgical fetal intervention has gone from a promising idea to successful practice. Decades of research and trials have established it as a safe and ideal solution for some fetal lesions. However, despite a profusion of publications on both the surgical techniques and the ethics of surgical fetal intervention, little has been published regarding the decision making process of the parents when presented with the option of fetal surgery. Nor is there data regarding the manner in which medical professionals present this choice to parents. Prenatal genetic counselors are in an ideal position to discuss surgical fetal intervention options with parents for whom it is a possibility; yet surgical fetal intervention has never been studied within the field of genetic counseling. I propose that a logical place to begin research on this topic is to establish what is already known about surgical fetal intervention within the prenatal genetic counseling community and what role the counselors play in the parents’ experiences. A summary of current practices will help to establish how genetic counselors view and address fetal surgery now. It will also potentially provide insight into what might be done to help genetic counselors learn about and counsel for this intervention option in the future and guide the development of educational tools, web seminars, or a conference.

My goal in this study will be to ascertain the current practices of prenatal genetic counselors regarding surgical fetal intervention. In order to gain a complete understanding of what current practices are, recruitment will target 2 subgroups of prenatal genetic counselors– those who are affiliated with fetal intervention centers and those who are not. These two groups are likely to have different perspectives on fetal intervention due to the nature of their positions and their different patient populations. The first group, prenatal genetic counselors who work in or with fetal intervention centers, primarily counsel for fetal intervention options, and therefore should have a greater knowledge base and comfort level on the topic of surgical fetal intervention. The second group, prenatal genetic counselors who are not affiliated with a fetal intervention center, are counselors who might refer their patients to a fetal intervention center once it is clear that the patient may be eligible for fetal surgery. This group would be expected to have a lower level of knowledge about surgical fetal intervention and be less comfortable offering the option to their patients.

STUDY SPONSOR

Brandeis University Genetic Counseling Program.
Surgical Fetal Intervention: Assessing the Current Practices of Genetic Counselors
Principal Investigator: Caitlin Melley

PRINCIPAL INVESTIGATOR’S QUALIFICATIONS TO DO THE RESEARCH

The Principal Investigator, Caitlin Melley, is a student in the Brandeis University Genetic Counseling Master’s Program and is conducting this research project as a requirement for the Master’s Thesis.

RESULTS OF PREVIOUS RELATED RESEARCH

Fetal intervention can be defined as any therapeutic intervention for the purpose of correcting or treating a fetal anomaly or condition. Fetal intervention can be divided into the broad categories of non-invasive and invasive techniques. Non-invasive fetal interventions, those that do not involve direct contact with the fetus or its environment, primarily include chemical interventions such as insulin given to diabetic mothers or tocolytic drugs to prevent premature labor. Invasive techniques, or fetal surgeries, are *in utero* operative treatments of a fetus. They range from inserting a needle into the fetal environment, for procedures such as fetal blood transfusions, to open fetal surgery to correct a fetal lesion, such as a congenital heart defect.

Liley in New Zealand performed the first successful documented invasive fetal intervention in 1963. Liley transfused blood into the peritoneum of a hydropic fetus with severe Rh disease (Redwine, 1993, Kunisaki, 2008). By the 1980s researchers had begun to experiment with techniques for opening and closing a gravid uterus without disrupting the pregnancy or jeopardizing the mother’s health and reproductive potential (Harrison, 1993). Such attempts at open fetal surgery met with limited success until 1989, when an absorbable staple was developed for a surgical stapling device. This allowed surgeons to open the uterus while preventing both excessive bleeding and separation of the fetal membranes from the uterine wall (Bond, 1989). The most significant limitation to fetal surgery since the advent of the absorbable stapling device has been the risk of preterm labor and delivery after the surgery (Kunisaki, 2008).

Today there are three major techniques used for surgical fetal intervention: minimally invasive percutaneous (ultrasound guided) procedures, open fetal surgery, and fetoscopic surgery. The percutaneous approach, developed in the 1980s, is typically used to drain “space-occupying fluid-filled structures” such as the pleural space or bladder of the fetus (Kunisaki, 2008). Under ultrasound guidance, a needle is inserted through the maternal abdomen and into the fluid filled structure. A small wire is then inserted through the lumen of the needle and the needle is removed. A catheter can then be placed over the wire and used to drain the space whenever it is needed. The percutaneous approach is generally an outpatient procedure (Kunisaki, 2008).

Dr. de Lorimier and Dr. Harrison of the University of California, San Francisco, pioneered open fetal surgery in the late 1970s and early 1980s. Open fetal surgery is performed under ultrasound guidance while both the mother and the fetus are carefully monitored. First, an incision is made in the lower abdomen to expose the uterus and the uterus is opened using the absorbable stapling device. Next, the surgical repair of the fetus is completed followed by the closure of the uterus followed by the maternal abdominal wall. After the procedure is complete the mother is awakened from anesthesia. Open fetal surgery requires a 3-7 day hospitalization and cesarean delivery of
the current and all future pregnancies. Preterm labor and delivery is a significant risk with open fetal surgery (Kunisaki, 2008).

Fetoscopy was developed in the 1970s as a diagnostic technique, but lost momentum when ultrasound techniques were refined in the 1980s. However, in the late 1990s, with the invention of smaller lightweight surgical instruments, fetoscopy regained popularity as a less invasive approach for fetal intervention. The technique requires a relatively small (1-2mm) incision in the maternal abdomen, through which a probe fitted with a camera and surgical instruments can gain access to the uterus and be used to repair the fetal anomaly (Kunisaki, 2008). Like open fetal surgery, fetoscopic surgery is always performed under ultrasound guidance, and has replaced open fetal surgery for some, but not all, fetal problems (Kunisaki, 2008).

Over 20 facilities in the United States alone offer fetal intervention services. The most common fetal anomalies to be corrected through fetoscopic surgery include twin-twin transfusion syndrome (TTTS), twin reversed arterial perfusion (TRAP) sequence, and amniotic band syndrome (Senat, 2004; Kunisaki, 2008). Congenital cystic adenomatoid malformation (CCAM) of the lung, fetal sacrococcygeal teratoma (SCT), spina bifida, and some congenital heart defects and urinary tract obstructions are most often treated by open fetal surgery (Eber, 2007; Redwine, 1993; Kunisai, 2008). In some cases, the surgery takes place at the time of delivery via the ex-utero intrapartum (EXIT) procedure, where a cesarean section is performed exposing only the top half of the fetus. Surgery is then performed while the fetus is still connected to the mother and receiving oxygen through the umbilical cord. When surgery is completed, the fetus is fully delivered. EXIT is most frequently used in the case of severe airway obstructions (Marwan, 2006). Additionally, prenatal treatments for congenital diaphragmatic hernia (CDH), myelomeningocele (MMC), and aortic valve stenosis (AVS) are all in the research stage (Kunisaki, 2008).

The diagnosis and assessment of birth defects has greatly improved over the past fifty years because of enhancements in ultrasound techniques as well as the development of fetal MRI, amniocentesis, and fetoscopy techniques (Redwine, 1983; Coakely, 1984). It is important to note however that, when detected prenatally, the majority of birth defects are still best treated with medical therapy during the pregnancy or with surgery immediately after birth (Kunisaki, 2008). Those fetal anomalies that are recommended for surgical fetal intervention are generally anatomic problems that cause ongoing damage to the developing fetus or pose significant risk to the mother, such as a congenital diaphragmatic hernia or a sacrococcygeal teratoma. When a fetal anomaly is first detected, parents are often unprepared for the diagnosis (Baker, 1998). It is important at this point that the parents receive counseling from a medical professional so that they are able to move forward with a complete understanding of their situation and the options available to them (Baker, 1998).

Genetic counseling is a subspecialty of medicine that “deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family” through a process of open communication (Baker, 1998). A genetic counselor is a master’s level professional. The American Board of Genetic Counselors (ABGC) currently recognizes thirty-three accredited graduate programs in the United States. Training in these programs includes four academic semesters or six academic quarters learning genetic, medical, and technical information as well as counseling skills and
ethics. In addition, genetic counseling students are required to complete clinical training during their graduate education to support the development of practice-based skills.

Genetic counselors help individuals and their families to comprehend medical and genetic facts relevant to a genetic disorder, as well as make fully informed decisions regarding how they want to deal with the genetic disorder within their family (Baker, 1998). The philosophy of genetic counseling includes a non-directive approach to patient education with complete disclosure of all available information. Genetic counselors also promote a psychosocial dimension in counseling, helping individuals and families to recognize their priorities, beliefs, and fears in order to make informed decisions (Hansen, 2004; Baker, 1998). Additionally, genetic counselors help affected individuals and their families to adjust as best they can to living with a disorder and to accept the possibility of recurrence in other family members (Baker, 1998).

Prenatal medicine is one of the major areas in which genetic counselors work. In this area, a genetic counselor’s practice is not limited to genetic conditions. They are also called upon to counsel on issues that may have little or no genetic basis, such as teratogenic exposures, history of pregnancy loss, or the diagnosis of birth defects (Baker, 1998). In most clinical situations an individualized approach that considers the indication, time of gestation at genetic counseling referral, and maternal age is needed. When counseling for the prenatal diagnosis of a birth defect or genetic disorder, genetic counselors give information to help parents understand and acknowledge the diagnosis and make choices about the course of action they are going to take (Agnieszka, 2007). A prenatal genetic counseling session is an important interaction on which life-or-death decisions may be based. Generally, parents are presented with three options: terminate the pregnancy, continue the pregnancy while monitoring the fetus, or actively intervene with the pregnancy to treat the fetus (Agnieszka, 2007). If the parents choose to continue the pregnancy, they can choose either to keep the baby or to give the baby up for adoption. A prenatal genetic counselor will often stay in touch with the parents throughout the pregnancy, and for some time after the baby is born (Hansen, 2004).

SUBJECT CHARACTERISTICS

The study participants for the proposed research project will be prenatal genetic counselors who are members of the National Society of Genetic Counselors (NSGC) who currently work in the field of prenatal genetic counseling in a clinical setting where they see patients at least one full day a week, or the equivalent of one full day. Participants must have at least one full year of clinical experience in the prenatal setting.

A subset of the participants will be prenatal genetic counselors that work directly in or with a center that offers fetal intervention. This subset of counselors will be recruited for their specific expertise in fetal intervention.

SUBJECT INCLUSION/EXCLUSION CRITERIA

In order to be eligible for this study, participants must meet the following criteria:

2) Must currently work as a genetic counselor in a prenatal setting
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3) Must have at least 1 full year of experience in a prenatal setting
4) Must see patients at least 1 full day a week, or the equivalent of one full day (8 hours).

A subset of participants must meeting the following additional criteria in order to be eligible for the interview portion of this study:
   1) Must currently work as a prenatal genetic counselor in or directly with a center that offers fetal intervention services.

JUSTIFICATION FOR USE OF ANY SPECIAL/VULNERABLE SUBJECT POPULATIONS

This study does not specifically recruit any subjects belonging to a special or vulnerable population.

STUDY DESIGN

The study design will include a quantitative, five-part, anonymous, online survey of prenatal genetic counselors to gather information on the current practices of prenatal genetic counselors regarding surgical fetal intervention. I will recruit genetic counselors as participants. I will present all of the genetic counselors who are recruited with the first four sections of the quantitative survey. I will only present the final section of the survey to a subset of participants, those genetic counselors who indicate that they work directly in or with a center that offers fetal intervention.

The study will also include a voluntary, semi-structured, qualitative telephone interview. I will interview 6 participants who indicate that they work directly in or with a center that offers fetal intervention in order to explore their role as genetic counselors in a fetal intervention setting in more detail. Each interview will last approximately 30 minutes.

STUDY PROCEDURES

Recruitment

In order to gather information on the current practices of prenatal genetic counselors regarding surgical fetal intervention, I plan to recruit participants through the National Society of Genetic Counselors (NSGC) by posting two recruitment notices on the NSGC’s General and Prenatal Listservs. Please refer to appendices A and B to view the recruitment notices. Appendix A is a notice recruiting all prenatal genetic counselors. Appendix B is a notice specifically recruiting a subset of prenatal genetic counselors that work in or with a center that offers fetal intervention services. Both recruitment notices will have a link that, when selected, will direct participants to an online survey at SurveyMonkey.com. Additionally, I plan to recruit participants who work in with a fetal intervention center via word of mouth.
At the end of the fifth section of the survey, participants who indicated that they work in or with a fetal intervention center will be invited to participate in a 20 to 30-minute interview in addition to the survey. These participants will be asked to email me directly upon completion of the survey if they are interested in being interviewed. I plan to interview a total of six of these counselors. If more than six participants contact me to be interviewed, then I will randomly select six. If fewer than six participants contact me to be interviewed, then I will interview all those participants who are interested.

**Data Collection**

A) Survey: I will administer an anonymous online survey— please refer to Appendix C. The questions will be quantitative, including multiple choice, Likert scale, and open-ended questions. The survey will be hosted by SurveyMonkey.com and will be accessible for a period of approximately two weeks. The survey will consist of five sections:

1) Participant information/demographics.
2) General knowledge about surgical fetal intervention.
3) Experience counseling for surgical fetal intervention.
4) Comfort level counseling for surgical fetal intervention.
5) Counseling practices of genetic counselors in a fetal intervention setting.

The first four sections will be administered to all participants. **Section 1** will ascertain basic information about the participants, including the state in which they practice and how long they have been practicing. It will also establish whether or not they are affiliated with a fetal intervention center. **Section 2** will assess prenatal genetic counselors’ general knowledge, including their understanding of what surgical fetal intervention is and which fetal anomalies are currently treated with fetal surgery. It will also ask how counselors first learned about surgical fetal intervention, and if they were exposed to the topic during their training. **Section 3** will be used to determine how often genetic counselors are called upon to discuss surgical fetal intervention with their patients and how often the patients themselves bring it up. It will address how they prepare for a genetic counseling session in which fetal intervention surgery may need to be discussed, what topics or issues they typically discuss during the session, what resources they use as professionals, and what resources they offer to their patients. **Section 4** will explore whether the counselors are comfortable discussing the option of surgical fetal intervention or if they feel it is not, in fact, their role. It will also ask if they would find new educational material or a seminar in fetal intervention helpful in the future.

**Section 5** will only be presented to those counselors who indicate that they are affiliated with a fetal intervention center in **Section 1**. After the general knowledge, comfort level, and experiences of prenatal genetic counselors are addressed in sections 1-4, those prenatal genetic counselors who are not affiliated with a fetal intervention center will have completed the survey. **Section 5** will assess the role of a genetic counselor affiliated with a fetal intervention center.

B) Interview: At the end of section 5, the participants will be invited to participate in a brief interview regarding their personal experiences working as prenatal genetic counselors in a fetal intervention setting. I will interview no more than six of these genetic counselors. Each telephone interview will last approximately 30 minutes, and I
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will audiotape each telephone interview as it takes place. The interviews will be comprised of qualitative open-ended questions. I will base my questions on my interview guide – please refer to Appendix E. I will use a semi-structured approach that will allow the interviewee to respond freely and without interruption. I will keep my responses and the order of questions flexible and I may adapt questions to account for original thoughts expressed by the interviewee. The overall interview content will remain consistent between all of the interviews.

**Data Analysis**

I will complete statistical analysis of all survey responses using SPSS to determine the knowledge level and most common practices among participants and to compare the responses from the two groups of prenatal genetic counselors – those affiliated with a fetal intervention center and those who are not.

I will transcribe all of the audiotaped interviews and complete descriptive analysis of the interview transcripts using ATLAS software to identify themes revealed during the interviews.

**INFORMED CONSENT**

I am requesting a waiver of informed consent for the survey respondents. The survey will be offered anonymously through Survey Monkey.com. There will be no way for me to obtain personal information about the participants through the survey tool and I will not request any personal information in the content of my survey.

Before proceeding with the interviews, I will obtain informed consent from each participant. I will mail each participant an informed consent form (ICF) – please refer to Appendix D – and schedule a telephone conversation to review the ICF together. Once I have received a signed ICF, I will sign as Principal Investigator and mail a copy to the participant. I will also proceed with scheduling and conducting the interview. I will not interview any participants without informed consent.

**ADVERSE EVENTS**

I do not anticipate any physical or psychological risks to participants.

**COMPENSATION**

Survey respondents will not receive compensation. Each interview participant will receive a $25 gift certificate from amazon.com.
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PRIVACY/CONFIDENTIALITY

The survey will be offered anonymously through Survey Monkey.com. There will be no way for me to obtain personal information about the participants through the survey tool and I will not request any personal information in the content of my survey. If a participant is interested in being interviewed, I will request that they email me. There will be no way to connect an interview volunteer to their survey answers.

To protect the privacy and confidentiality of interview participants I will assign each interview participant a coded ID number. The only link between the participant’s name and ID number will be a password-protected spreadsheet. I will store interview participant names, contact information and demographic information only in this spreadsheet. Only I will know the password to the spreadsheet. During the interviews, I will address participants using their ID number rather than their name. Audiotapes, interview notes and study files will be labeled with the ID number rather than participant names or other identifiers. At the end of the study, I will delete the spreadsheet and study files and destroy the audiotapes and interview notes. In addition, I will delete contact information for and email responses from ineligible individuals and destroy any notes taken during screening interviews after completion of the study.

COSTS

There will be no costs to study participants.

REFERENCES: