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Aminah Sallam

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A QUALITATIVE APPROACH TO UNDERSTANDING THE ATTITUDES OF  
MATERNAL FETAL MEDICINE SPECIALISTS REGARDING PRENATAL  
DIAGNOSIS, DISABILITY AND TERMINATION

A Thesis Submitted to the  
Yale University School of Medicine  
in Partial Fulfillment of the Requirements for the Degree of Doctor of Medicine

By

Aminah Sallam

M.D. Candidate, 2021

## ABSTRACT

**Title:** A Qualitative Approach to Understanding the Attitudes of Maternal Fetal Medicine Specialists Regarding Prenatal Diagnosis, Disability and Termination

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**Background:** Prenatal detection of congenital anomalies has forced Maternal Fetal Medicine specialists (MFMs) to navigate many new ethical and legal issues when counseling expecting parents. And yet, little is known about the attitudes, beliefs and perceptions of MFMs regarding the intersection of prenatal diagnosis, disability and termination. Thus, we aimed to identify the key attitudes and opinions surrounding prenatal genetic testing, disability and termination of MFM specialists who counsel expectant mothers with a prenatal diagnosis of fetal anomaly.

**Methods:** This was a prospective, qualitative study using semi-structured interviews with MFMs across the nation, recruited through the snow-ball method. Interviews were audio-recorded, transcribed, and qualitatively analyzed using modified grounded theory by three reviewers.

**Results:** 2,227 codes from 17 interviews were grouped into six clusters. Codes were then characterized to triangulate MFM perspectives into 18 themes and 39 sub-themes. Difficulties with patient misunderstanding of prenatal genetic screening, diagnostic technology, the implications of certain genetic anomalies, and limited time to explain these

factors were identified as major barriers to effective counseling. All physicians reported viewing themselves primarily as informants in the prenatal diagnostic process, and all offered their patients the option of termination in the case of the diagnosis of a severe or lethal congenital anomaly; however, physician definitions of severe or lethal congenital anomaly differed. Moreover, physicians reported fears of being perceived as coercive by patients, and a high degree of moral and emotional distress during these consultations. Finally, physicians identified legal, financial and structural barriers that prevented patients from accessing prenatal genetic diagnostic services and termination services. It was felt that these limitations ultimately inhibited autonomous decision making by patients.

**Conclusions:** These findings highlight important barriers, perspectives, and conflicts that occur for MFM providers during prenatal consults with pregnant women following prenatal discovery of congenital anomalies. Further exploration of the identified themes in this study would benefit MFM providers in identifying strategies to improve prenatal counseling.

## ACKNOWLEDGEMENTS

**To my mentors:** Matthew Drago, Audrey Merriam and Mark Mercurio. Thank you for listening to my story and helping me transform my experience into action. I have learned so much from each of your perspectives, approach to inquiry, and passion for improving patient care.

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**To my sister:** Mariam Sallam. Thank you for always listening to me and for always cheering me on. The tenacity and fervor with which you approach the things you love and your endless kindness inspire me.

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## INTRODUCTION

Recent and rapidly developing advances in the world of prenatal screening and diagnosis have given future parents access to the genetic makeup of their fetus earlier in gestation than ever before. The scope of each of these tools is increasing, allowing us to more accurately identify anomalous genetic conditions. The choice of which testing modality to use is patient specific, and dependent in large part on both the gestational age of the fetus in question and the level of detail regarding the fetus' genetic makeup that the family is seeking.

For a subset of patients, what starts off as a simple prenatal screening blood test or ultrasound can quickly evolve into an ethical minefield of decisions on further prenatal genetic testing and pregnancy continuation versus termination in the event that a congenital anomaly is identified. And while Maternal Fetal Medicine specialists (MFMs) remain the primary point of contact for families navigating the complexities of this decision-making process, typically after a referral from their primary obstetric provider, little work has been done to understand their viewpoints on issues surrounding prenatal genetic testing, disability in the setting of congenital or genetic anomaly and pregnancy termination following a diagnosis of congenital anomaly. As such, we sought to explore the ethical pitfalls of our current prenatal genetic testing paradigm, the role of MFMs in that paradigm, and MFM perceptions on where this current paradigm both fails and succeeds in ensuring optimal patient care. Ultimately, we hope our findings can be leveraged to improve the delivery of care for patients receiving prenatal genetic testing.

## **Prenatal Genetic Screening and Diagnosis**

In general, the purpose of prenatal genetic testing is to identify chromosomal abnormalities. Indeed, chromosomal abnormalities in pregnancy are relatively common, with approximately 1 in 150 live births involving some type of chromosomal abnormality<sup>1</sup>. While the most common chromosomal abnormality remains aneuploidy, there are a number of chromosomal anomalies to consider. Specifically, aberrations in chromosomal structure, which include deletions, duplications, translocations, and other rearrangements. Of note, these aberrations are not necessarily pathologic. For example, chromosomal translocations can be balanced. This means that the normal genomic content is preserved but rearranged. Additionally, chromosomal anomalies are not necessarily present in all fetal cells. When this occurs, the fetus is said to be mosaic, and it can lead to variable phenotypes in the neonate<sup>2</sup>.

Clearly, prenatal diagnosis can be incredibly complex, and a diagnosis made via prenatal genetic testing is not always sufficient to predict the clinical outcome of the fetus. That being said, prenatal genetic testing remains the primary method through which parents concerned about the presence of aneuploidy in their fetus can learn more about their fetus' genetic makeup. At present, primary testing options offered to expectant mothers are divided into two major categories: screening tests and diagnostic tests. Understanding the difference between these two forms of testing is critical, as screening tests are designed to assess whether a patient is at increased risk of having a fetus affected by a genetic disorder. Diagnostic tests, on the other hand, are intended to determine whether a specific genetic disorder or condition is present in the fetus<sup>2</sup>. Consequently, a clear understanding of the



way these tests work, and how they are incorporated into current practice, are critical to any discussion on their utility.

Current primary screening options include the first trimester screen, the second trimester screen and cell free DNA testing (cfDNA), also referred to as Non-invasive Prenatal Testing (NIPT) in the literature. Prior to the development of NIPT, obstetric providers could offer their patients a first trimester screen, the second trimester screen, or a combination of the two screens through sequential or contingent screening. Factors included in these screening options include nuchal translucency ultrasound, serum levels of beta-hCG, Pregnancy-Associated Plasma Protein A (PAPP-A), Alpha Fetoprotein (AFP), Estriol and Inhibin. These screening modalities integrate various components including the patient's age, race, presence of pregestational diabetes and multiple gestations in order to provide the patient with a risk calculation for the presence of fetal trisomy 21, 18, or 13. Of these, the most common chromosomal disorder is trisomy 21, with an incidence of about 1 in 700 live births<sup>3</sup>. While trisomy 18 and 13 are rarer, with incidences of 1 in 3,315<sup>3</sup> and 1 in 7,409<sup>3</sup> live births respectively, screening for these conditions is desirable to many women given their life-limiting nature<sup>4</sup>.

Use of the first and second trimester screens has decreased in favor of NIPT. While the extent to which NIPT has been adopted among women in the United States is not clear, one study estimates that 25-50% of all pregnant women in the United States receive NIPT<sup>5</sup>. NIPT screens for fetal aneuploidies using the analysis of cfDNA fragments in the maternal circulation. The fetal component of cfDNA originates from placental trophoblasts released

into maternal circulation from cells undergoing programmed cell death. As such, the quantity of cfDNA from the fetus increases throughout gestation. At present, NIPT is the most sensitive and specific screening test for the common fetal aneuploidies. However, it has the potential for false positive and false negative results, and is not equivalent to diagnostic testing<sup>6</sup>. Performance of NIPT in patients who receive an interpretable result is shown in Table 1.

			Age 25 Years	Age 40 Years
	Sensitivity (%)	Specificity (%)	PPV (%)	PPV (%)
Trisomy 21	99.3	99.8	33	87
Trisomy 18	97.4	99.8	13	68
Trisomy 13	91.6	99.9	9	57
Sex Chromosome Aneuploidy	91.0	99.6	20-40%	20-40%

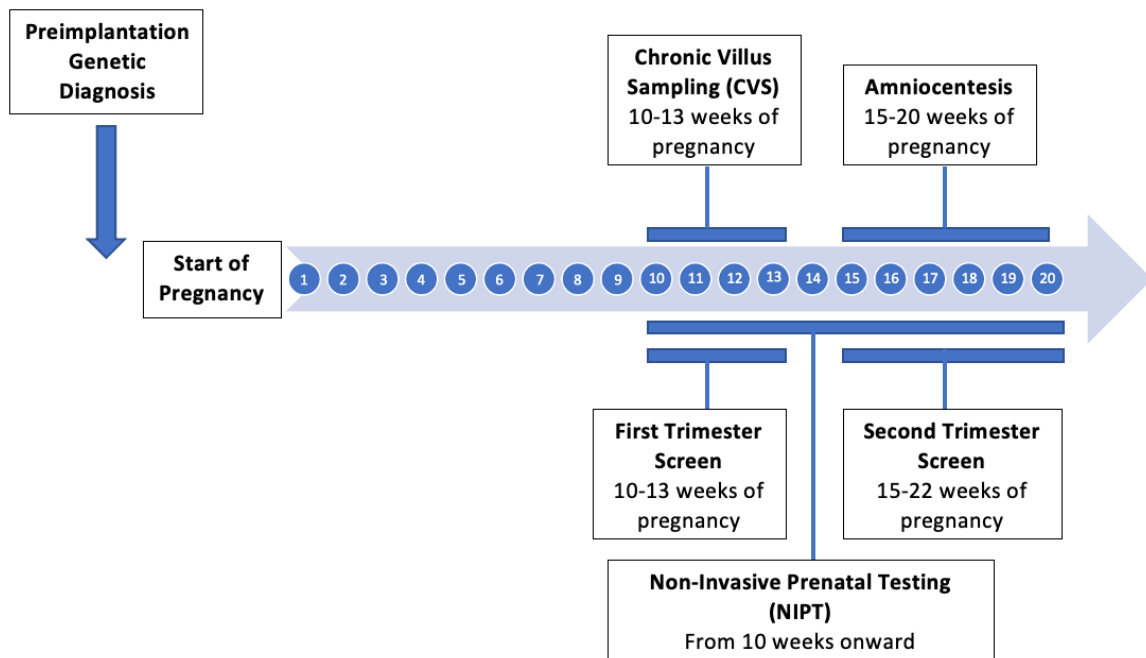
**Table 1.** Performance of NIPT in detection of trisomies 21, 18, 13 and sex chromosome aneuploidies, adapted from ACOG Committee Opinion No. 640<sup>7</sup>. The Positive Predictive Value (PPV) of these tests describes the probability that subjects with a positive screening test truly have the disease and varies based on patient age.

Diagnostic tests are able to diagnose the presence of a genetic disorder in a fetus with certainty. Each of these tests is done at a different stage of gestation, and comes with their own risks and benefits. Diagnostic testing is most commonly performed during pregnancy with fetal cells obtained by amniocentesis or placental cells from chorionic villus sampling (CVS). Patients undergoing in vitro fertilization can also have preimplantation genetic testing performed on cells from their pre-implanted embryo. Preimplantation genetic diagnosis involves the testing of an embryo created via in-vitro fertilization prior to implantation. This mode of diagnosis is generally used to test for a genetic condition in

which a specific mutation has been identified in the family, but can also be used to screen for genetic anomaly in instances where there is no known familial history of mutation. Because this method of diagnosis requires only a few cells from the early embryo, errors in diagnosis are possible, with one study reporting misdiagnosis following preimplantation genetic testing to occur in less than 1 in 200 pregnancies<sup>8</sup>. Thus, confirmation of these results via CVS or amniocentesis later in gestation is recommended<sup>2</sup>.

CVS is performed as early as 10 weeks gestation. Through this method, placental villi are obtained for genetic analysis. Placental villi are acquired through either a transcervical or transabdominal approach, as limited data has shown there is no significant difference in risk between either approach<sup>9</sup>. With ultrasound guidance, the tip of a needle or specialized catheter is placed into the placenta without entering the amniotic sac. Negative pressure with a syringe is then used to aspirate a small amount of placental villi<sup>2</sup>. The main advantage to this method remains how early in gestation it can be performed, with results taking anywhere from 24 hours – 14 days to return. Consequently, expectant mothers might have the results of their CVS test returned within their first trimester. This, in turn, allows for more management options available to women with an abnormal result regarding pregnancy continuation or termination. Complications of CVS include minor risks, such as vaginal spotting, and more significant risks, such as pregnancy loss, limb reduction defects, heavy vaginal bleeding, culture failure, amniotic fluid leakage and infection. However, the actual risk of any of these complications, with the exception of vaginal spotting or bleeding, is less than 1% in patients that have a transabdominal approach<sup>2</sup>. In contrast, up to 32% of patients who undergo CVS via transcervical approach may experience vaginal spotting or

bleeding<sup>10</sup>. Thus, CVS proves to be a safe method of genetic testing and diagnosis early in gestation. Of note, mosaicism is more likely to be found in CVS samples due to cases of confined placental mosaicism. If identified, amniocentesis is typically offered to assess whether mosaicism is present in fetal cells as well. In 90% of cases, the amniocentesis result is normal, giving the diagnosis of confined placental mosaicism<sup>11</sup>.



**Figure 1.** A visual representation of the prenatal genetic testing timeline by gestational age in weeks. Preimplantation genetic diagnosis takes place prior to the start of a pregnancy. The remaining tests can be done at the end of the first trimester through the remainder of the pregnancy.

Amniocentesis is generally performed between 15 weeks and 20 weeks of gestation. Performance of amniocentesis prior to 15 weeks of gestation is not recommended as it results in significantly higher rates of pregnancy loss<sup>2</sup>. Amniocentesis is performed with ultrasound guidance, and requires a 22-gauge spinal needle to obtain a sample of 20-30 mL of amniotic fluid. The most significant risk of this procedure remains pregnancy loss, however, the actual rate of loss as a result of amniocentesis remains under 1%<sup>12</sup>.

Thus, while it is clear patients have access to a variety of screening and diagnostic genetic tests, the method they ultimately choose is influenced by a number of factors. These include, but are not limited to, the time of presentation for prenatal care, personal and family history of chromosomal abnormalities, maternal age, and concerning findings on a screening ultrasound. As such, expectant mothers may receive both a screening and diagnostic genetic test, a screening ultrasound and a diagnostic genetic test, or even just a screening genetic test. The combination of tests an expecting mother chooses to undergo is extremely personal, and is decided in large part through discussions with their OBGYN.

### **The Reproductive Autonomy Rationale as an Ethical Framework for Prenatal Diagnosis**

In general, the purpose of a prenatal genetic screening test lies in its ability to provide an assessment of a patient's risk of carrying a fetus with a chromosomal disorder. Consequently, the current recommendation from the American College of Obstetricians and Gynecologists (ACOG) is that prenatal genetic screening should be discussed with and offered to all pregnant women regardless of maternal age or risk of chromosomal abnormality, as all women should have the right to the knowledge that prenatal testing provides<sup>6</sup>.

This recommendation is in line with the "reproductive autonomy rationale," which is rooted in the idea that women should make informed decisions surrounding their pregnancy. According to this ideology, access to prenatal testing promotes a woman's ability to make informed choices by empowering them to manage their pregnancies in a way that aligns with their preferences and personal value system. This rationale emphasizes

nondirective counseling and consent as the primary means by which women's decisions about testing and subsequent care are informed and free of undue pressure<sup>13</sup>.

Respect for patient autonomy is considered to be the foundation of modern Western biomedical ethics. Beauchamp and Childress, in their "Principles of Biomedical Ethics," define respect for autonomy as a "professional obligation...[and] autonomous choice [as] a right, not a duty, of patients"<sup>14</sup>. Integral to autonomous choice is the "respectful treatment in disclosing information"<sup>14</sup>. Indeed, the primary justification for informed consent in our current ethical model has been to protect autonomous choice. To that end, Beauchamp and Childress have recommended a model for informed choice. As part of this model, they define three major categories: Threshold Elements, or preconditions for informed consent; Information Elements; and Consent Elements. Within each major category, they further define subcategories. In order to meet the preconditions of informed consent, a patient must be considered competent and possess voluntariness to make a decision. In order to successfully complete the Information Elements of informed consent, a physician must: 1) disclose material information; 2) recommend a plan; and 3) ensure the patient has understood the information provided. Finally, in order to complete the Consent Elements of informed consent, a patient must make a decision in favor for or against a given plan, and authorize said plan<sup>14</sup>.

For the purposes of this discussion, I will focus on the Information Elements of this construct, as it is often presented as the sole focus of informed consent. To fulfill this element, patients must be given "facts or descriptions that patients or subjects usually

consider material in deciding whether to refuse or consent to the proposed intervention<sup>14</sup>.”

Additionally, they must “understand at least what a health care professional...believes a patient...needs to understand in order to authorize [said intervention]<sup>14</sup>.”

To fulfill these requirements of informed consent professional bodies have published extensive guidelines that outline the necessary practical steps for clinicians to follow. For example, ACOG recommends that patients should be given access to high quality, accurate information about the technology of the testing modality in question and their results. This includes information surrounding the detection and false positive rates, advantages, disadvantages and limitations of screening tests as well as the risks and benefits of diagnostic procedures<sup>6</sup>. Second, patients should be given information surrounding life with any of the anomalies being screened for, and implications of a positive result. Finally, patients should be given adequate time to discuss prenatal testing with their providers in order to ensure full understanding.

The fact that fulfillment of these standards must be met and documented prior to any medical procedure ensures that women understand that invasive prenatal diagnostic testing is associated with an increased risk of miscarriage. Noninvasive screening tests, on the other hand, have proven to be a challenge as they do not have a procedural timepoint to mark the need for informed consent. Thus, since its inception, an “extensive gap between theory and practice when it comes to informed consent” has existed for NIPT<sup>13</sup>. Indeed, a meta-analysis of thirty empirical qualitative research studies found that women consistently expressed dissatisfaction with the quality and quantity of information provided

to them during counselling on NIPT. Specifically, these women identified three major areas where the counseling they received was lacking. First, they reported that they did not feel their clinician was sufficiently informed about the technology to facilitate informed choice. They felt this gap in knowledge was especially pronounced when discussing the experience of raising a child with conditions such as trisomies 13 and 18 and sex-linked disorders. Second, appointment time constraints contributed to an overall sense of dissatisfaction with counseling received, with women reporting that consultations were too short for adequate counseling to be delivered. Finally, and especially concerning, when asked to describe NIPT, many women described it as easy or just another blood test<sup>15</sup>.

These findings highlight further detractors from informed decision making, including routinization or a pressure to test<sup>15</sup>. Moreover, each of the above identified barriers to informed consent serves as a direct threat to the reproductive autonomy rationale, and warrants further discussion.

#### Gaps in Provider Knowledge Surrounding NIPT Technology

Patients receiving counseling by their healthcare providers on NIPT have raised concerns that health care providers have not been able to provide adequate information to patients in order for them to make a fully informed decision regarding NIPT during their pregnancy<sup>15</sup>. This may be a result of the rapid diffusion and widespread implementation of NIPT. Thus, it is imperative that we understand the difficulties faced by healthcare providers seeking to interpret and explain NIPT to patients.



Pragmatic issues, in particular how the results of NIPT are reported, provide a partial explanation for why providers struggle to counsel women appropriately on the results they can expect from NIPT, and the implications of these results. While NIPT screens for three common aneuploidies and sex chromosome abnormalities, the results of this screen are not reported in a simple, bimodal fashion--in other words, physicians do not receive a print-out detailing the aneuploidies screened for with a “positive” or “negative” result to indicate the presence or absence of aneuploidy in the fetus. Because NIPT is a non-definitive screening test performed by a number of different companies, results can be reported as positive, “high risk,” negative, “low risk,” or some variation of “no call,” without any concrete calculations to qualify these predictions. This stands in stark contrast to the manner in which results from the first and second trimester screens are reported: as an easy to read print out that clearly shows the risk calculations for a given trisomy of a patient’s fetus (Figure 1).

Interpretation of NIPT results are impacted by numerous factors. For example, the positive predictive value (PPV) of NIPT is less than 50% in low risk patients<sup>16</sup>, meaning that the specificity is much lower than the 99% frequently cited by companies and providers alike. This makes counseling on false positive results essential.

Multiple other factors can influence how accurate the results from NIPT are. For example, the likelihood that a woman over 250 pounds receives a result of “no call,” is higher compared to women under 250 pounds<sup>17</sup>. This disparity is due a dilutional effect found in women over 250 pounds and is more common when NIPT is done earlier in the screening

window. Finally, false-positives can occur when there is a confined placental mosaicism, an early demise of an aneuploidy co-twin, a presence of maternal mosaicism, and if there is presence of occult maternal malignancy<sup>17</sup>. Thus, while sensitivity and specificity remain high for NIPT, its predictive value of aneuploidy in any given patient varies.

It is critical that physicians have concrete values for PPV and residual risk value to be able to adequately communicate the variable predictive value of NIPT to their patient. For this reason, ACOG and SMFM recommend that all laboratories include these values in their result reports<sup>18</sup>. However, most laboratories do not include the PPV and residual risk value in their reports. This is because they do not request that false positives, which are needed to calculate PPV, be reported to them. Moreover, there is no central data collection for reports of false positives that OBGYNs can access to estimate the PPV themselves<sup>19</sup>. Without these values, reasonable interpretation of a NIPT result by the providing OBGYN for any given patient is nearly impossible<sup>20</sup>.

To further complicate counseling on NIPT, there has been no established standard of practice across the multiple specialty organizations (SMFM, ACOG, ACMG) by which providers are recommended to determine which patients should be offered NIPT. Despite only showing a high sensitivity and low false positive rate in high-risk pregnancies, defined as pregnancies in women over the age of 35, NIPT is often offered to all women as the primary screening test for aneuploidy by OBGYNs. Moreover, while administration is most common in the second half of the first trimester, as this is the earliest time fragments of fetal DNA can be found in maternal circulation, it is sometimes offered as a secondary



October 12, 2016

LabCorp Test Master  
Test Account  
3060 South Church Street  
Burlington, NC 27215

Test Results of: SAMPLE REPORT, 017500  
DOB: 06/12/1985 Sex: F  
Collected on: 10/11/2016  
Received on: 10/11/2016  
Reported on: 10/12/2016

Branch Number: POE00  
Account Number: 90000999  
Specimen Number: 285-225-9114-0

Patient ID#:

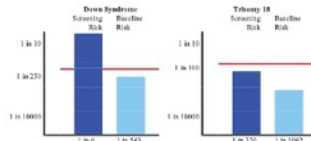
Physician:

First Trimester with NT

**INTERPRETATION:**  
Screen Positive for Down syndrome  
Screen Negative for Trisomy 18

	Screening Risk	Maternal Age Risk	Cutoff
Down Syndrome	1 in 6	1 in 543	1 in 250
Trisomy 18	1 in 320	1 in 1062	1 in 100

	MoM	Value
NT	1.30	1.9 mm
PAPP-A	0.05	30.0 ng/mL
HCG	11.59	1000.0 IU/mL
DIA	45.72	9999.9 pg/mL



CLINICAL INFORMATION USED TO CALCULATE PATIENT RESULTS

Maternal Data	Fetal Data	1st Trimester Sample
Maternal Age at EDD: 31.9	CRL: 56.0 mm	Collection On: 10/11/2016
Race: Caucasian	CRL Date: 10/11/2016	GSA: 12.1 weeks
Fetal#: 1		Weight: 189 lbs
		Sono ID#: 000
		Additional US Markers: None

COMMENTS

Follow up options include genetic counseling, CVS, or amniocentesis.

NOTE

This test does not screen for open neural tube defects (ONTD). Measuring MS-AFP (test number 010801) in the second trimester can provide ONTD screening. The optimal gestational age for ONTD screening is 16-18 weeks.

Geraldine A. McDowell, Ph.D., FACMG  
Principal Genetics Technical Director

References: Available upon request

For further inquiries contact LabCorp Genetic Services at 800-345-GENE.

LabCorp  
1912 Alexander Drive, RTP, NC, 27709 (800) 345-GENE

Arundhati Chatterjee, M.D.  
Medical Director

This document contains private and confidential health information protected by state and federal law.



ABOUT THIS SCREEN: Panorama™ is a screening test, not diagnostic. It evaluates genetic information in the maternal blood, which is a mixture of maternal and placental DNA, to determine the chance for specific chromosome abnormalities. The test does NOT tell with certainty if a fetus is affected, and only tests for the conditions ordered by the healthcare provider. A low risk result does not guarantee an unaffected fetus.

**FINAL RESULTS SUMMARY**

<p>Result <b>LOW RISK</b></p>	<p>Fetal Sex <b>Female</b></p>	<p>Fetal Fraction <b>10.0%</b></p>
-----------------------------------	------------------------------------	--

RESULT DETAILS: ANEUPLOIDIES

Condition tested <sup>1</sup>	Result	Risk Before Test <sup>2</sup>	Risk After Test <sup>3</sup>
Trisomy 21	Low Risk	1/424	<1/10,000
Trisomy 18	Low Risk	1/860	<1/10,000
Trisomy 13	Low Risk	1/2,740	<1/10,000
Monosomy X	Low Risk	1/255	<1/10,000
Triploidy	Low Risk		

1. Excludes cases with evidence of fetal and/or placental mosaicism. 2. Based on maternal age, gestational age, and/or general population, as applicable. References available upon request. 3. Risk after test for aneuploidy incorporates results from the Panorama algorithm and data from a published study of 17,885 women [Dar et al. Am J Obstet Gynecol. 2014. Nov;211(5):527.e1-27.e17] and are reported as PPV (high risk) and NPV (low risk). Maternal age is utilized in this calculation, however the "risk after test" may not reflect the actual PPV for this patient, as additional risk factors, including but not limited to: results of other screening, ultrasound findings, personal/family history, are not included in the risk assessment.

**Figure 2.** A side-by-side comparison of results from a first trimester screen (left) to that of NIPT (right). While first trimester screens provide a clear interpretation, NIPT results only communicate the level of risk of a given aneuploidy.

screening test by OBGYNs to women who do not want diagnostic testing but are seeking clarity on the probability of aneuploidy in their fetus after receiving an abnormal ultrasound finding or first-trimester screen. This practice is problematic as any abnormal NIPT result requires an additional diagnostic test to confirm the finding if there are not sufficient sonographic findings to corroborate the NIPT results. Thus, offering NIPT as a secondary screen serves only to delay definitive diagnostic testing. If too long, this delay could limit a woman's options regarding the continuation of her pregnancy.

Ultimately, without concrete risk calculations and clear guidelines on who to screen using NIPT, obstetric providers are left unprepared to administer and interpret NIPT. A study done in 2013 on 130 OBGYNs found 78% of them reported not being familiar with NIPT and its associated clinical data<sup>21</sup>. Another nationwide survey done in 2014 of OBGYNs found that almost half of those surveyed viewed NIPT as a complete substitution for invasive testing<sup>22</sup>. More recent studies highlight the fact that OBGYN understanding of NIPT remains inconsistent. In 2016 a survey of 103 OBGYNs nationwide found that 15% considered NIPT to be a diagnostic test<sup>23</sup>. Another survey of maternal-fetal medicine fellows found that although they were able to accurately answer questions related to NIPT for trisomy 21 with high accuracy, their answers to questions regarding NIPT in twin pregnancies and monosomy X screenings were found to be far less accurate<sup>24</sup>. This is concerning, as it is estimated that 80% of NIPT requisitions request information regarding the fetal sex, and disorders of sexual development account for 36% of reported discordance between NIPT results and prenatal ultrasound or other clinical information<sup>25</sup>.

Many obstetric providers recognize these gaps in their knowledge and cite the need for more education as new tests continue to roll out<sup>26</sup>. At present, online training is an ideal mode through which to provide educational material to physicians, and there are a significant number of online resources for readers to access in order to learn about NIPT; however, these resources range in reliability as they are subject to various levels of review. Farrel et al attempted to evaluate the quality of a number of online educational resources for NIPT and found that more often than not those resources had no measure of quality review<sup>27</sup>. A significant amount of web-based information is curated by commercial laboratories, and it is often this information that serves as an educational resource for OBGYNs to develop and update their knowledge base about NIPT<sup>28</sup>. This utilization of commercially developed information poses clear ethical risks for practice, as ACOG has previously cited a concern about possible bias that can be introduced from industry playing a role in medical education<sup>29</sup>.

#### Gaps in Provider Knowledge Surrounding Life with Disability

If NIPT shows a high probability of fetal aneuploidy, the expectant mother faces the following options regarding the remainder of her pregnancy: 1) to carry the pregnancy to term without further testing; 2) to pursue diagnostic testing before making the final decision to carry the pregnancy to term or terminate; and 3) to terminate the pregnancy. Decisions to terminate a pregnancy in the setting of a positive screening test for aneuploidy or diagnosis of aneuploidy have been well documented, resulting in an overall decrease in the number of babies born with genetic disease and chromosomal abnormalities as a result of selective termination<sup>30-32</sup>.

The reasons for pregnancy termination in the setting of a suspected or known genetic abnormality are certainly complex, and grounded in a number of factors beyond the scope of this discussion. These include, but are not limited to, the emotional and financial cost of raising a disabled child and the effect on a family's ability to care for their other children. Moreover, "findings that older maternal age, greater socioeconomic status, identification with particular races and religions, decreased strength of religious beliefs, and prior experience with genetic testing correspond to willingness to contemplate abortion [of a fetus with congenital anomaly]<sup>33</sup>."

That being said, the authors wish to focus on one important factor influencing this decision-making process: how providers inform parents on what to expect when raising a child with expected disability from a congenital anomaly. In 2002, a study done by Roberts et al. found that 87% of pregnant women did not feel they got information about the future quality of life for a child with disability. Moreover, 83% of women reported feeling like they did not receive information about both the positive and negative aspects of raising a child with disability<sup>34</sup>. A study done in 2005 by Skotko et al found that mothers who gave birth to a child with Trisomy 21 diagnosed prenatally did not feel that their physicians gave them adequate information regarding Trisomy 21 prior to initiating prenatal testing<sup>35</sup>. In 2011, a study done by Sheets et al. found that genetic counselors were more likely to highlight negative aspects of Down Syndrome during initial discussions surrounding prenatal diagnosis<sup>36</sup>. In 2012, a study done by Brown et al found that prenatal counseling about congenital fetal conditions varied considerably between MFM and pediatric based fetal care practices. Specifically, when asked their attitude about the importance of offering

certain information when patients seek guidance on whether to continue a pregnancy in the setting of Down Syndrome, CDH and spina bifida, MFMs were more likely to respond that for each condition, offering options for pregnancy termination was of high importance<sup>37</sup>. Finally, in a meta-analysis published in 2019, multiple studies showed that women did not feel they received adequate information surrounding the experience of raising a child with conditions such as trisomies 13 and 18 and sex-linked disorders<sup>15</sup>.

These parental reports of not receiving adequate information surrounding life with a disability, coupled with the previously discussed barriers to informed consent for NIPT, leads to the reasonable hypothesis that decisions made surrounding prenatal testing and termination could be rooted in misinformation. Indeed, Press and Browner found that in a very diverse group of women--who all underwent prenatal screening--the primary influencing factor in their decision to undergo prenatal screening was the way in which screening was described to them by their healthcare provider<sup>38</sup>. Moreover, the influence a health-care provider has on a woman's decision-making process following prenatal diagnosis is well documented, with factors such as the provider's approach and specialty (i.e. obstetrician versus general practitioner) playing a critical role<sup>39</sup>. With many women feeling that the negative aspects of raising a child with genetic anomaly are highlighted in the prenatal counseling process, and the reported importance of offering termination to women carrying a fetus with certain genetic anomalies, prenatal genetic testing becomes framed by some as a

“‘search and destroy mission’ with eugenic overtones. [This, in turn]...burdens [women] with the expectation that their individual choices not only align with their own values, but also promote a tolerant and diverse society, one free of stigma,

discrimination and eugenic attitudes. Paradoxically, then, the reproductive autonomy rationale--meant to ensure that women are free to make their own choices--forces them to shoulder the responsibility for the societal consequences of these choices. It also frequently ignores the unequal way these choices affect women with different socioeconomic resources in different political contexts, especially in societies without a strong social welfare safety net<sup>13</sup>.”

### Time Constraints

Health care providers need to relay a significant amount of information to their patients surrounding prenatal screening, diagnosis and disability in order to ensure their patients are making informed decisions. Adequate communication of this information certainly takes time, with a simple overview of current testing options taking several minutes alone. And yet, a recent study published in 2016 found that when health care providers offered genetic screening to their patients, conversations about screening for aneuploidy lasted 1.5 minutes on average, and most providers’ counseling did not adhere to ACOG recommendations<sup>40</sup>.

This finding is reflected in the literature, where women are cited as feeling that their consultations were too short for adequate counseling about NIPT to be possible. As a consequence, many noted that a variety of questions and concerns went unaddressed. Other women described feeling a sense of “information overload,” and were too overwhelmed to process the information given in the time allotted for discussions with their providers<sup>15</sup>.

While full understanding is not required for informed consent, patients should understand the salient aspects of a proposed procedure and the consequences of proceeding with or declining prenatal screening<sup>14</sup>. In this light, the abundance of literature showing women who received prenatal screening did not understand what test they were receiving and why is especially concerning.



### Routinization and the Decision Trap

As it stands, the route to informed consent for prenatal testing is both challenging and time consuming. This, in combination with the simplicity and ease of prenatal screening tests, raises concerns about the potential for routinization of prenatal screening. Indeed, there is a danger that women may just view it as simply “another blood test”<sup>41-43</sup> and opt-in to testing without fully understanding the importance or implications of receiving NIPT<sup>41,43</sup>. This is described as the “decision trap” by Silja Samerski in his book by the same name. There, he discusses how the choice to engage with genetic technologies can become a ‘trap’ that people entered both willingly and eagerly because they didn’t understand the implications of their initial choice. Indeed, patients start by undergoing a screening test they considered to be routine care and are forced to face ultimate questions about the meaning and value of human life<sup>44</sup>.

The implications of this decision trap are far reaching. Most notable are the threats routinization of prenatal screening pose towards attitudes surrounding disability. As Samerski puts it, “Prenatal testing creates patients who cannot be helped. They cannot be healed, only aborted.<sup>44</sup>” This is especially true in a paradigm that does not prioritize education of patients about disability but encourages testing and termination. Moreover, it raises concerns about a

“possible slippery slide toward a society that...penalizes those who resist [prenatal testing] by stigmatization and even punishing them, for instance, by ‘refusing to allocate public funds to pay for the medical and other costs for children born with a disorder that could have been diagnosed but whose mothers refused testing.<sup>13</sup>”

This same paradigm could apply to women who chose to continue their pregnancy in spite of a diagnosed disorder. Consequently,

“while the principle of autonomy easily justifies offering prenatal testing, a narrow focus on autonomy overlooks the tremendous pressures women face to accept the offer of such testing and to terminate pregnancies when a serious disability or condition is diagnosed. The burden of decision-making about whether to test and what to do following a diagnosis falls on the shoulders of individual women.<sup>13</sup>”

### **The Role of the Maternal Fetal Medicine Specialist**

Maternal-Fetal Medicine is a branch of obstetrics that focuses mainly on the medical and surgical management of high-risk pregnancies. They also have special competence in the diagnosis and treatment of women with complications of pregnancy. Moreover, MFMs have the specific training and experience required to perform complex diagnostic and therapeutic procedures involving the mother and or fetus during pregnancy. Finally, MFMs provide peer and patient education concerning the most recent approaches and treatments for obstetric problems in order to promote risk-appropriate care for complicated pregnancies<sup>45</sup>.

Of particular importance is the role MFMs play in prenatal diagnosis of congenital anomalies. In general, patients with positive, high risk or “no call” prenatal genetic screening tests are often referred to MFMs for further counseling or testing. This is because MFMs are able to perform complex procedures for advanced prenatal diagnosis, including CVS and amniocentesis<sup>45</sup>. As such, MFMs are often the first to interact with parents of a patient with congenital anomaly and often are the first to make the diagnosis.

Given the role of MFMs as front line in the prenatal diagnosis of congenital anomalies, they are uniquely positioned to uphold the Reproductive Autonomy Rationale for patients facing the possibility of undergoing prenatal genetic testing. And yet, while there is

abundant literature surrounding patient experience and perception of the counseling they received regarding NIPT, little is known about the experience of MFMs as specialized providers at the forefront of counseling on prenatal screening diagnosis and pregnancy management.

### **Statement of Purpose**

As such, we sought to understand how MFMs perceive the role their counsel plays in the complex decision-making process surrounding prenatal genetic screening, prenatal genetic testing, prenatal diagnosis and termination. We additionally sought to shed light on the important attitudes, beliefs, perceptions about social norms and perceived behavioral control, as described in the theory of planned behavior<sup>46,47</sup>, to fully understand the choices MFMs make in counseling in their patients. We chose a qualitative approach as it will best allow us to capture this complex social phenomenon from the perspective of the participants<sup>48</sup>.

### **SPECIFIC AIM**

To identify the key attitudes and opinions surrounding prenatal genetic testing, disability and termination of MFM specialists who counsel expectant mothers with a prenatal diagnosis of fetal anomaly.

### **METHODS**

#### **Study Design**

This is a prospective qualitative study of semi-structured interviews with MFMs. IRB approval was obtained prior to the onset of the study, and all subjects provided informed

consent prior to participating. We enrolled participants via purposeful snowball method. Through this method, existing study subjects recruited future study subjects from their personal network<sup>49</sup>. This process was initiated by the MFM on our study team, AM, who provided the medical student, AS, with referrals to MFMs in the field at different institutions. AS reached out to each of these contacts for study enrollment. Following each interview, AS requested interviewees to refer them to MFMs in their personal network for study enrollment. Participant recruitment continued until conceptual saturation was reached. We defined this point of conceptual saturation as one where we achieved both depth and breadth in our discussions of MFM experience. This target is based on the central tenets of grounded theory analysis, as described by Corbin and Strauss, which emphasizes identifying and exploring a variation of concepts with sufficient detail to ensure depth of conceptual understanding, rather than how representative our participants were of the overall population of MFMs<sup>50</sup>.

Interviews with all study participants were conducted in English by a single interviewer, AS. Because the literature supports the use of both face-to-face interviews and telephone interviews within the same study without compromising the trustworthiness of findings<sup>51</sup>, five interviews were conducted in person and 12 were conducted over the telephone. All recruited participants were given an optional demographics survey to complete at the time of the interview.

To facilitate open discussions with study participants, we created a semi-structured interview guide with questions based on literature review of prior publications on the study

topic. We then piloted the interview guide on a group of three MFM fellows for face validity, and to determine if length and flow of questioning were appropriate prior to study commencement. These interviews were not included in our final analysis, but allowed us to refine our interview guide such that all questions asked to our research cohort were clearly stated, exploratory in nature, and encouraged differences between physicians' perceptions and experiences to emerge during the interview. The final semi-structured interview guide can be found in Table 2. Of note, questions were added to each interview at the discretion of AS to encourage discussion or provide for clarification of concepts brought up by the study participant.

Tell me a little about yourself:

Tell me about your training

How would you describe what you do to someone who does not know much about MFM?

Why did you choose this specialty (as opposed to other OBGYN specialties)?

How do you think other physicians view your job/role in patient care?

What do you see your role as in managing referral patients with prenatal/genetic diagnosis?

When you talk to expectant mothers/parents about a prenatal genetic diagnosis, what topics do you like to cover?

How do you organize this type of consult?

Prompt: What do you do in the case of an unclear genetic diagnosis?

Once you've talked about diagnosis, how do you proceed? What are the options?

Do you give specific advice to parents about-termination when you diagnose a congenital anomaly?

Tell me more about your feelings regarding termination in this instance.

How do you handle your own biases in counseling these patients?

Is there any emotional conflict in your work with prenatal diagnosis?

Prompt: Do **you** have any emotional conflict ...

Tell me about your hardest or most difficult patient

Tell me what's difficult/common difficulties

What was difficult about this case?

What did you struggle with the most?

Could you imagine raising a child with a disability?

Prompt: What if it is a severe disability?

What if the child requires long term intensive care?

What do you imagine life is like for kids with a disability as a result of a genetic disorder?

How does that impact the advice you give to parents about raising a child with disabilities?

Tell me about your patient population

Are there any characteristics about your patient that might change your approach or interaction?

Is there anything else you feel impacts your approach? (Prompt: culture/religion/financial status)

Prompt: How does that affect the way you feel about what a patient should do?

Tell me about the reactions, interaction and conversations with parents after providing the option of termination

What is the most surprising thing you heard a patient say in response or after their consult with you?

What do you think a patient leaves the room understanding after your discussion about termination?

Who do you think influences a patient's decision?

How do you incorporate family into your discussion?

Outside resources

What support services do your patients have access to?

Do you refer your patients to other providers (genetic counselors, social workers, care coordinators) to help them understand their child's condition better?

Do you have a pediatric or neonatal palliative care specialist?

Do you refer your patients to pediatric specialists to prepare them for complications they might face once their child is born?

Do you personally perform terminations, or do you refer patients if they desire abortion?

Does your institution have a family planning division?

**Table 2.** Questions from our interview guide. The questions above were used to jumpstart conversations with MFMs.

### **Data Analysis**

Each interview was digitally recorded and transcribed verbatim. Transcripts were then imported to NVivo (Version 12.6, QSR International, Burlington, Massachusetts, USA), a

qualitative software program that helps to organize and retrieve data for qualitative analysis. Analysis of this data was conducted by a medical student (AS), a MFM (AM) and a neonatologist (MD). As described above, transcript analysis was conducted using a grounded theory approach, wherein the researchers conducting this study independently analyzed a subset of transcripts, and attached descriptive codes to segments of text in each transcript. Following this independent first-pass analysis, researchers met to discuss their impressions and reconcile variations in their developing codes. Once discrepancies in codes were reconciled, codes were grouped into broad topic-oriented clusters. Within each cluster, codes were assigned to a specific “theme.” These themes allowed us to construct a cohesive idea or theory about an investigated phenomenon, and served as the basis for our coding framework, which was then used to code subsequent interviews. As our understanding of these themes deepened, we further classified codes according to “sub-theme.” Simultaneous data collection and analysis allowed for an iterative process wherein investigators would independently analyze interview transcripts, identify and agree upon emerging themes to subsequently pursue in the following interviews<sup>50</sup>. Analysis concluded when theoretical saturation was reached, as described above.

## RESULTS

In total, we interviewed 17 participants. Of these, 15 returned a completed demographics survey, and their responses are shown in Table 3. Of the MFM’s who returned a demographics survey, nine were female and six were male. The majority of respondents were under the age of 45 (n = 9). Additionally, the majority of respondents (n = 11) reported practicing at a major academic center. Finally, while the greatest number of respondents



were from the Northeastern United States, we were able to interview MFMs practicing in all major regions within the continental United States: North East, South, Midwest, and West.

Coding analysis of the interview transcripts yielded a total of 2,227 codes grouped into six clusters. Codes were then characterized to triangulate MFM perspectives into 18 themes and 39 sub-themes, shown in Table 4. These findings express the varying perspectives on the MFM providers' role in counseling expectant parents on prenatal genetic testing, disability and termination.

### **Interview Clusters and Themes**

#### **1. Physician Methodology**

Our first cluster, Physician Methodology, includes statements made by MFMs describing the approach they take when counseling patients on all issues related to prenatal screening, testing and diagnosis. We identified four major themes to characterize the overall structure MFMs follow when counseling their patients.

#### **Physician Goals**

First, MFMs described their approach to prenatal counseling as being rooted in goals identified prior to each counseling session. While some MFMs described universal goals to ground each discussion with patients, others felt it was important to tailor each session to the patient's goals.

*[I] try to identify their goals for the visit or for the testing. What their background or fund of knowledge is, and how we can meet their goals. (MFM 17)*

Universal goals were rooted in the notion that all patients are entitled to as much information as possible:

*I think it just comes back to my whole philosophy of practice [which] is that it's not my choice to make anything. It's my place to inform them, and help them to understand and make them aware of their options and whatever their options--whatever they choose--help them access that option. (MFM 15)*

### Establishing Diagnosis

Second, an MFM's approach to establishing a prenatal diagnosis varied based on the level of information surrounding the diagnosis that their patient came to the MFM with. In general, MFMs saw themselves as on the frontline for both establishing the diagnosis and explaining the implications of said diagnosis to the patient.

*I feel like pretty much the first line of communication with the patient and family who have some new issue or complication in their pregnancy and really going over with them what that means, what their options are and what the next steps are. (MFM 14)*

This sentiment was felt to be especially true with referral patients.

*You have to keep in mind I am not usually the first person that someone sees because of my job description, so they usually know that there is something wrong and that I am there to explain to them what is wrong, basically. Because they understand [that] there is some sort of a complication and that they need a specialist and that's why I am there. (MFM 10)*

Whether or not the MFM was the first to walk the patient through their prenatal testing options to reach a diagnosis or if a patient was referred to the MFM with concerning findings in hand, MFMs described their approach to establishing a diagnosis as multifaceted. Steps to take included gathering appropriate imaging, establishing a wide differential diagnosis, reviewing potential genetic associations to the identified anomaly, and counseling patients on how their pregnancy management might be impacted based on the findings and potential diagnosis.

### Best Practice

Interestingly, while discussing their overall approach to patient counseling, MFMs repeatedly referenced parts of their methodology they considered to be paramount to appropriate counseling, what we called ‘best practice.’ This included how MFMs tailored their consultation to meet the goals they defined for the consultation. Although almost all MFMs referenced how they thought it was most appropriate to deliver bad news to patients, we noted two separate approaches. While some MFMs felt it was necessary to cushion or qualify the delivery of bad news to make it easier for patients to digest, others felt they had an obligation to patients to be as up-front and direct about the bad news as possible.

*I try not to finalize anything immediately...They say I'm having them come back because I think, you know, that I'm wrong or I'm trying to soften the blow [but] I said...I just don't like to make these diagnoses that are going to affect decisions of Life off one ultrasound, so I'll give the baby one more week and take a look at it again. (MFM 1)*

*When I was a fellow I actually helped with a support group for women who terminate wanted pregnancies because of a genetic or ultrasound diagnosis...they [also] said they often could tell there was a problem before someone actually said there was a problem, and they don't like that. So that's the first thing I do when I walk in the room. I say: 'Hi, I'm Dr. MFM 4, nice to meet you, I've looked at your images and I want to let you know I think there's a problem' because I think the prolongation of even just saying those words builds more anxiety in the patient. (MFM 4)*

Repeatedly, when discussing what they thought to be best practice when counseling patients, MFMs emphasized the value of consistency in counseling. This was generally in reference to whether or not MFMs tailored their approach based on certain demographic characteristics of their patient.

*Their responses vary, I'm sure of that. My approach doesn't. (MFM 3)*

*I'm going to discuss pretty much everything the same with the patient regardless of her, you know, insurance status, socio-demographics, or anything...regardless of their religion, I'm still going to say the same thing. (MFM 5)*

Tasked with having to communicate complex and difficult information during a single counseling session was noted by many MFMs to be extremely difficult. This is both because the MFMs felt patients often find information difficult to digest in one sitting, and because MFMs felt that when tasked with making decisions about termination, patients should be given time to process before they make such a permanent decision. To that end, MFMs describe repetition as a valuable tool in order to help patients understand what a diagnosis means for them and the future of their pregnancy.

*You know, sometimes I think people are not ready to understand everything and it may take more than one visit for the information to sink in...[so] we will go through it again and sometimes another time. (MFM 12)*

All MFMs interviewed felt they had a professional obligation to discuss termination as an option in the event that a patient was given a prenatal diagnosis of genetic anomaly. Many noted that discussions surrounding the option of termination are extremely loaded, and have the potential to become points of contention during a counseling session. MFMs described ways they diffused this tension through neutral language to normalize the conversation.

*I've sort of learned to always couch it as "hey look, we're going to talk about something that's really uncomfortable for people, but me as your doctor you know, I think it's really important for me to at least bring up. I'm not your friend, I'm not your family member, I'm not your priest, I'm not your pastor, like here's the doctor, we're going to talk about this. And if you never want to hear this word again, if you never want to speak about it again, please let me know. But I always sort of say it's my obligation as your healthcare person to at least bring it up. (MFM 6)*

One MFM discussed framing the topic using neutral, nondirective language.

*I think our job as MFMs is not meant to be prescriptive. I think especially in a situation like this where there are things way above and beyond what we do that are involved in this decision....we have to try and not insert our personal beliefs onto this situation and let them make this decision themselves...unless I see an*

*obvious reason to [guide patients in one direction or another] I try to be as neutral as possible...we respect your decision one way or another we will support you with either decision...but we just want to make sure that you know you have the information to make the decision that is right for your family. (MFM 13)*

Beyond the use of neutral language, many MFMs felt it was easier to approach the option of termination by framing it as an option many patients with the same diagnosis have either considered or taken. By saying termination is something other people with the same diagnosis have pursued, many MFMs felt they were making the option both easier to digest for the patient, and easier to accept for patients leaning in that direction.

*So the phrase I usually use is like, 'There are some people who, when seeing this on ultrasound, would choose to end a pregnancy or would choose to terminate a pregnancy or would choose to not continue' and see, depending on who I'm speaking to, one of those phrases, so that they hear that this is an option for them that's...I don't want to say reasonable, but that people do this. And in my mind somehow using that phrase allows them to accept that a little bit more. (MFM 7)*

Finally, to achieve best practice MFM providers described the importance of consulting colleagues especially when they felt they were not the best person to answer specific patient questions. In those instances, many relied on the expertise of their colleagues either in the field of maternal fetal medicine, genetic counseling or pediatrics in order to help patients get all the information they needed to make a decision about their pregnancy.

*So, for example, if...a woman's fetus is diagnosed with Down Syndrome...I can talk to her about it generically, but I don't take care of children with Down Syndrome. So, it would be difficult for me to tell her, 'Oh this is what it means--your child will need this and your child will need that and this is what to expect,' because it's not my specialty. So, I can speak in very broad strokes, but ultimately...[I would try] to help her get information either, I mean usually from professionals, whether they're pediatricians who take care of children with Down Syndrome or geneticists or even the internet or what not. But generally a consultation with someone who takes care of these kids to give the woman a better sense of what this means. (MFM 16)*

<u>Gender</u>		<u>Years in Practice</u>		<u>Religion</u>	
Female	9	0-4	5	Agnostic	2
Male	6	5-10	1	Atheist	3
<u>Age Group</u>		11-15	3	Catholic	3
24-34	1	16-20	3	Jewish	2
35-44	8	26-30	1	Orthodox Church	1
45-54	4	31-35	1	Prefer not to answer	1
55-64	2	<u>Political Identification</u>		Protestant	3
<u>Practice Location</u>		Conservative	1	<u>Type of Practice</u>	
Midwest	4	Independent	3	Academic	11
Northeast	6	Liberal	6	Hospital based clinic	1
South	4	Prefer not to answer	2	Employee of HMO	1
West	1	Progressive	3	Private practice	2

**Table 3.** MFM Demographics. Participating MFMs had varying levels of practice and came from diverse backgrounds. We spoke with MFMs from all over the U.S., including those from the Midwest (ND, SD, NE, KS, MN, IA, MO, IL, WI, IN, MI, OH), Northeast (ME, NH, VT, MA, RI, CT, NJ, NY, PA), South (TX, OK, LA, AR, MS, AL, GA, FL, TN, SC, NC, KY, VA, WV, DC, MD, DE) and West (WA, OR, CA, AK, NV, AZ, NM, HI, CO, UT, WY, ID, MT, WY).

<i>Clusters</i>	<i>Themes</i>	<i>Sub-Themes</i>	<i>Example</i>
<i>Physician Methodology</i>	Physician Goals		“It’s just making sure that they have as informed consent...as they possibly [can] have and whatever I can do to give them that I think is my goal.”
	Establishing Diagnosis		“What I like to do is...to try to get as much information as possible prior to the patient coming, from her referring doctor, what their concerns were based on the ultrasound...And then sit down with the patient, her family, and anybody else, to really have the opportunity to discuss everything in the context of our findings... and explain what we see and how it impacts on that diagnosis.”
	Best Practice	Delivery of Bad News	“I would say [I am] very direct in counseling. I try not to sugar coat anything because it doesn’t help anybody I feel like while they might be angry with me in the beginning about it, they have time...they would be angry regardless of who delivered that news. It’s not me, it’s the news.”

		Consistency	“Understanding what their cultural and religious beliefs can sometimes be helpful in counseling them...but I still provide them all with the exact same counseling.”
		Repetitive Counseling	“It may take more than one visit for the information to sink in...and we’ll sit down and we will go through it again and sometimes another time.”
		Neutral Language/ Normalizing Termination	“I feel like it’s particularly important to discuss it [termination] in an unbiased way for prenatal diagnosis patients.”
		Colleague Opinion	“...sometimes it’s better to ask for a second opinion from a colleague or somebody who’s kind of looking with fresh eyes at the case and who the patient hasn’t already associated with receiving bad news and a negative experience.”
	Barriers to Best Practice	Inconsistencies in Practice	“I don’t like when people order the test and then the patient really didn’t want the test. I think that makes it challenging that we have some providers



			who will do certain tests and then the patient doesn't want to know the information..."
		<p>Technical Limitations</p> <p>Time Restraints</p> <p>Scientific Limitations</p>	<p>"A patient is scheduled for a number of minutes, so if there's an abnormality, clearly you need to spend more time...but in the back of your mind there's always just like the [thought that] others are waiting for you to see [them] as well."</p> <p>"But like, with down syndrome we can't predict how severe the disability is going to be. So, we're very careful not to, you know, promote one way or another. That if you've got this diagnosis of down syndrome that you'll have mild intellectual verses severe intellectual disability, I guess, it's often the</p>

		Unclear Significance	<p>moderate to severe. We always tell them that genetics, in amniocentesis results, can't predict that."</p> <p>"Well then we have to tell them we don't know...this is I call it the dark side of prenatal diagnosis. Stuff that is uncertain, and depending on the location and the size is why we have genetic counselors, and they help us to counsel if there's any potential bad implication but the reason it's unknown is that it's so rare, problems with is are so rare..."</p>
		Lack of Understanding about Disability	"I have really zero concept of what it might be like to live with a child that has disability."
<i>Physician Perception of Disability</i>	Objective Viewpoint	Define Disability	"I think it depends on the disorder. Some kids live their life in bliss, some kids live their life without any recognition of it because they don't have the capacity to recognize it. And for some kids, it's very difficult based on their cognition of it and their experience compared to other kids. So I think it

			ranges, sort of between a bad and difficult life to a very happy life and everything in between...”
		Define Severity	“[I define a severe anomaly as one that] would need for a pediatrician to attend to the patient at the time of delivery or soon after....something that requires intensive care.”
		Define Lethality	“...lethal is a moving target. Like twenty years ago...lethal was completely different than lethal is today. And there was that editorial in JAMA pediatrics...about Rick Santorum and his daughter and just the shifting place of the word lethal.”
	Subjective Viewpoint	Chasing Perfection	“I think that there’s this search for perfection in the world. And I understand it, I’m a parent, like I get it, I get it very very strongly. But I think that we’ve come to expect things in life that aren’t possible, this idea of a perfect life.”

		Common Uncommonality	“Don’t forget, 3% of all pregnancies have significant congenital abnormality however it’s....quite a bit of patients are normal and everything...but when you start getting specialized, pretty much everybody has something.”
		Stigma of Disability	“Whatever you’re given [if your child has any disability] ...there’s a community for you...So, it just felt ‘normal’ as a term is so heavy and so judgmental. That I just can’t say it anymore, it just doesn’t come out anymore.”
<i>Physician’s Experience</i>	Defining Moral Role	Moral Distress	“Yeah, I think that I have a difficult time personally, um, with aborting a pregnancy that isn’t again, a lethal condition or a situation where the mother’s health is at significant risk.”
		No Judgment	“It’s not for me to tell the family whether that’s a pregnancy “worth terminating.” So, if someone says to me, I’m going to have a termination, I say ok, I help them with that. And if they say we’re going to keep the baby,

			I say great, you should probably meet with someone who takes care of these kids so you have a sense of what to do. And I uh, go that way.”
		Not My Role	“It’s not my place to tell her what to do, it’s just not in the conversation at all.”
		Pro-Choice	“I support it. I mean, I do them. Um I have always been very pro-choice, I donate to planned parenthood, I used to work at planned parenthood as a resident.”
		Pro-Genetic Testing	“So you know while I think the old school thinking is only get these [genetic] tests if you’re thinking you might consider termination, I would say that’s silly, I mean if knowledge is power right?”
	Emotional Response	Difficulty Conveying Bad News	“...people get very sad. And um, especially for things that are ambiguous – I’m thinking like posterior fossa abnormalities, like Dandy-Walkers, I don’t freaking know what to tell people. I find those very, very, very hard.”

		Empathy	“It’s easy to sit from here, to make comments to the patient’s choices and everything but unless you are in their shoes, unless you are facing that question at a personal level, it is not a question that can be answered.”
		Fear of Being Perceived as Coercive	“I think we’re very careful so that it doesn’t sound like we’re recommending or suggesting abortion. So, I don’t think that people get mad at us for that. But I have seen instances where people get mad. ‘Why would you ever tell me to kill my baby?’ Um, and uh, but I don’t, that doesn’t typically happen to me, fortunately.”
		Self-Doubt	“[...] in situations where I feel like I must not have explained or counseled well enough for the patient to have a truly good and logical understanding of the situation...And so then I feel that I myself am to blame for not providing enough education and counseling for them to have a true understanding.”

<i>Physician-Patient Relationship</i>	Effective Communication	Avoid Jargon	“So there are a lot of people I’m explaining what a chromosome is—they don’t understand that you get a set from your mom a set from your dad. So I use colloquialisms or other terms like for example for like the karyotype or the chromosomes I use the word ‘blueprint’ or ‘these are the instructions that tell the cell to do what it needs to do.’”
		Eliciting Patient Understanding	“I start with asking the patient what they have heard already and what their understanding is of the diagnosis is and try to tailor my counseling based on that kind of what their level of understanding already is.”
	Responding to Parental Needs	Handling Family Dynamics	“In some instances, we notice that the [family] dynamic might not be a healthy dynamic so we cannot let that go so we try sometimes [to make] excuses...we try to catch the patient while she is not with the other person.”
		Patient Dictating Care	“It depends on the woman of course, what her—what she considers as an option or not an option for her.”

		Reactive Values Elicitation	“I really just say from the get-go that I am required and obligated to offer you all the full range of options, but some families tell me that they would never consider pregnancy termination, and if that’s how you feel then I won’t talk about it anymore.”
	Shared Decision Making	Guiding Family	“And that’s what I think the role of the doctor is, not to tell them what to do but to put things into context for their decision.”
		Informational Role	“I try to give some examples, I give them a little bit of time to digest, ask questions, and often I ask them to tell me what they are understanding so that I feel whether they were able to receive the diagnosis I try to deliver to them.”
		Maternal versus Fetal Health	“If it’s something lethal...I might talk more about termination and also related to risks of continuing pregnancy for mom. Because just being pregnant is a higher risk condition than not being pregnant.”



<i>Patient Experience</i>	Cultural Influence	Faith	“I often try to get a sense, even near the beginning of counseling, of whether or not the family has given any thought to pregnancy termination. And most families are pretty open with me in terms of if they feel like based on their religious view or personal values that it’s something they would never consider...”
	Familial Influence		“I think family’s like the number one influence...most patients really rely on their families and their spouse.”
	Barriers to Care	Socioeconomic Barriers	“I’m upfront with them with the options, telling them that this one here could be very expensive, you know we can see what we can do to help decrease the price but I also think you can’t go through the counseling and tell them all the options and then they choose the one that’s going to cost them \$20,000...”

		System Barriers	“We’re limited by practice in a federal hospital where, by law, we’re not allowed to perform a termination unless it’s rape, incest, or life of the mother [is at risk]....”
	Emotions	Patient Hope	“I quickly realized that she was a reasonable person in an unreasonable situation, and this was a pregnancy that was so desired after years of IVF and want not...she was sort of grasping for anything.”
		Patient Regret	“...there are parents who say now that my child is 14, 15, wheelchair bound, you know all these things I would not have chosen what I did.”
	Inability to Understand Child’s Condition	Language Barriers	“So with a language barrier all these difficulties and these grey areas become so much more complicated to communicate...”

<i>Termination</i>	Abortion Culture		“In general, the population is a fairly politically conservative and there’s a pretty low rate of pregnancy termination even after a prenatal diagnosis or genetic abnormality.”
	Abortion Tourism		“I think it’s going to be interesting to see what happens in the next year or so because I think we could start having a lot of abortion tourism here. So that could be very interesting to see from our perspective. Because we will be doing ultrasounds and there might be a lot more anomalies and stuff coming now to access our abortion services.”
	Legal Considerations		“It is a very strange law [where] you can’t terminate in Pennsylvania or Ohio anymore based on Down Syndrome, so...basically, a patient can terminate at 20 weeks if she doesn't want the pregnancy, but [not] if the baby has Down syndrome.”

**Table 4.** Thematic clusters from interview analysis. We subdivided clusters into themes and sub-themes for more nuanced and detailed analysis.

### Barriers to Best Practice

In addition to emphasizing how to best approach a patient undergoing prenatal counseling, MFMs discussed barriers they faced in attempting to meet the benchmarks outlined in the “Best Practice” theme. Of these, we identified three principle sub-themes: Inconsistencies in Practice, Technical Limitations and Lack of Understanding about Disability.

MFMs expressed frustration surrounding inconsistencies in practice between themselves and their colleagues both in maternal fetal medicine and in pediatrics. MFMs felt that in these instances, counseling of patients was compromised as they received different messages from different providers.

*What does not work is when one of your partners has seen the patient and has already promised them the world. You know, we'll do a C-section, we'll do anything you want. And then I'm seeing them or I'm on labor and delivery when they come in and I have to deal with all of these things that I think are wrong, but you know my partners have already planted the seed because let's face it if there's x# of us, there's a 1 out of x# chance you're going to be the one delivering the baby, so it's probably not going to be you, so you can tell them anything you want to get them out of the office. (MFM 1)*

*Unfortunately, because our NICU is so pro-life it puts the MFM's in a very hard spot because how can [they] plan to do a resuscitation on a kid and us not do anything from an antenatal standpoint to prepare that child for life because we know earlier that baby [would need] magnesium and steroids...how do you [as an MFM] withhold that stuff if [the NICU] going to intubate the kid...[our relationship with the NICU is] a little bit dysfunctional. (MFM 2)*

MFMs also expressed frustrations surrounding the inconsistencies or lack of prenatal counseling their patients received prior to consultation. Multiple MFMs highlighted instances in which their patients had received poor prenatal counseling by their referral providers.

*It's almost like what happens is now that we have cfDNA [it's] even worse because patients aren't counseled appropriately. They're just getting it, low risk patients [...] they're not being counseled appropriately before being sent to us. (MFM 1).*

*I find that that's a very confusing topic for lay people, for parents, for med students, for residents. I think that the average patient, um pregnant woman going into the doctor's office, is not counseled appropriately in any way shape or form because I don't think general OBGYN's have the time or frankly the knowledge to go through all that, and they all have their own biases as well. (MFM 8)*

Ultimately, MFMs felt that inadequate counseling by referral providers saddled them with the task of having to deliver bad news to unsuspecting patients.

*The OB really, I don't think had not prepped the family for how bad it was. So maybe they walked into our office thinking maybe the head was a little big. And I think I walloped them with just something horrible. (MFM 6)*

MFMs also felt that various technical barriers served as barriers to their best practice. The first of these barriers were time constraints. These constraints were noted to exist both within the context of the appointment itself, and in the broader sense when considering how long a woman has to make a decision regarding her pregnancy. Because the majority of states in the United States do not offer late term abortion, women are forced to make decisions regarding termination sometimes only a few days after receiving a diagnosis of anomaly in their fetus.

*Just time...you know there's...other logistic problems as well. You know a patient is scheduled for a number of minutes, so if there's an abnormality, clearly you need to spend more time. [But] in the back of your mind there's always...others waiting for you to see as well [...] [Ultimately] it is an arbitrary decision, isn't it? Just like how much time you think you should spend with patients or you can afford to spend with patients. (MFM 15)*

*There are sometimes emergency situations where you have a quick decision [to terminate]...[sometimes] a few days if not a few weeks to make a decision to terminate. (MFM 12)*

Complicating matters is the second technical limitation identified by MFMs: scientific limitations. Indeed, inherent to prenatal diagnosis is a level of uncertainty regarding the outcome of a diagnosis as we are rarely able to predict the exact manifestations of an aneuploidy. As one MFM described:

*The diagnosis is always assumptions. It's never 100% certain, so trying to explain how to make sense of this uncertainty and trying to pinpoint....I cannot provide that answer to them. It makes them even more frustrated, so that's I think, the biggest thing about my job. (MFM 15)*

Certainly, anomaly exists on a spectrum that we have some insight to but cannot predict. MFMs also touched on how the limitations in the science and technology available for prenatal diagnosis make prenatal counseling challenging.

*I think a lot of what we do....raises anxiety for no reason. I think a lot of it is basically us trying to prove how smart we are when the reality is we don't know anything until the baby comes out. Most of the time [it leads to] weeks of anxiety. (MFM 1)*

Indeed, in many instances of prenatal screening and testing, patients receive information they were not expecting. In response, MFMs described a sense that the current diagnostic technology we have has outpaced our knowledge on the implications of certain genetic findings. This was felt to be especially true in reference to the use of microarrays for analysis of genetic material, as this method often turns up “Variants of Unknown Significance,” or genetic anomalies that may or may not be pathogenic. This sense of unclear significance, our final technical limitation, was referred to by one MFM as “the dark side or prenatal diagnosis” (MFM 3), as it leads to the communication of a “diagnosis” without actual evidence to show whether the diagnosis was pathologic or simply a benign variant.

The final barrier to best practice highlighted by our MFMs was the fact that many did not actually feel qualified or prepared to speak on life with a disability. For some, this lack of understanding stemmed from a lack of familiarity with existing literature, or minimal interactions with disabled persons. For others, they felt they could not understand and therefore could not counsel on life with a disability because they did not experience disability themselves. Either way, this lack of understanding surrounding life with disability served as a major limitation for MFMs when trying to counsel their patients on the meaning of a diagnosis of a congenital anomaly resulting in a disability.

## **2. Physician Perception of Disability**

The concept of disability and how it was perceived by our interviewees emerged as our second major thematic cluster. This cluster emerged from statements that captured the personal definitions MFMs used to describe disability and their varying perspectives on topics surrounding disability. We grouped these statements into two main themes, objective viewpoints and subjective viewpoints, and seven total sub-themes.

### Objective Viewpoints

The need to describe to patients what disabilities could be associated with prenatal findings that arose during consultations was a common finding in interviews. From this discussion emerged the concept of needing to set objective parameters for dialogue. Providers shared their perceptions of what disability meant. Consistent among MFM definitions of disability was the use of the term “spectrum” to describe disability. The way MFMs defined each end of this spectrum were based on a number of factors. Some used the terms ‘major’ and ‘minor’

*Trisomy 13--major. Multiple, multiple, severe anomalies, like a big abdominal wall defect with abnormal limbs and a big-you know-an enlarged brain and you know a big heart defect, that's not good. Something in between would be a major heart defect. Something minor would be clubfoot or ventriculomegaly. You know in my mind everybody has their own scale. (MFM 4)*

Others defined their spectrum of disability based on their perception of a disabled individual's quality of life.

*I think it depends on the disorder. You know, I mean I think nowadays, for many of these, many infants that are born with Down Syndrome who get an early intervention program and all that, I think for many of them for those children, they can have a meaningful existence, very pleasant, happy lives. And some of them can be very functional, or pretty functional, depending on what their genetic background gives them in terms of parental intelligence. But, you know, for many of them, they will have significant lifelong challenges. I think for some of the other more significant genetic disorders, I think their life is much much more challenging. (MFM 5)*

*I think quality of life is something that we're all very poor at measuring and we all have different markers for what defines someone's quality of life. And I think some of that is personal, so I think it's more describing to the family what this child's life is like. You know, I've had families who are happy because their child is able to smile. Other families, they're happy because their child is able to eat. And you know, other families who would be very disturbed by those things if they weren't able to feed themselves, if they weren't able to go to a regular school. (MFM 9)*

Multiple MFMs defined this spectrum based on their perception of a disabled individual's level of awareness surrounding their disability. Meaning, there exist disabled individuals who are unaware of their disability or difference, and are able to live happier lives than those disabled individuals made to be aware of their disability. Some MFMs referenced the ability of disabled individuals to experience happiness as a qualifying factor of their spectrum of disability. Others defined disability as a nonmainstream condition. Finally, the degree of visibility (i.e. intellectual versus physical versus both), and the degree of



additional care a disabled individual would require were evoked by providers to describe the spectrum of disability.

The concept of a “severe disability” came up multiple times throughout our interviews. Usually, the degree of disability severity served as the basis for the MFM to discuss termination options with their patient. As such, we asked MFMs to describe their personal definitions of disability severity in order to get a better understanding of what they would consider reasonable grounds for termination assuming the condition could be diagnosed prenatally. MFMs also identified conditions they did not consider to be severe and thus did not or would not warrant termination.

*People with obviously lethal or severe abnormalities, lethal in the neonatal period or the early childhood period, yeah, I certainly have that discussion with them [regarding termination]. [...] For people with minor abnormalities, I don't typically have the discussion with them, especially if they have declined genetic testing about pregnancy termination. (MFM 5)*

*I don't bring it [termination] up with cleft lip palate, for instance. Um, I don't bring it up with something like gastroschisis, which is something very surgically resect-able. All of this is nuance. With anomalies that are fixable and don't really impact the quality of life, I normally don't bring it up. (MFM 6)*

*The severe one. Uh, something like Fragile X or cerebral palsy that was severe or probably Downs Syndrome probably in that more severe category. (MFM 14)*

We also found variability in what MFMs conceived as lethal conditions. Some MFMs were hesitant to use the term lethal to describe a condition if a fetus could survive with their condition for a few minutes following birth. Other MFMs were hesitant to describe a condition as lethal due to developments in medicine that allow for more conditions to be survivable.

*They don't call it "incompatible with life" we call it "life-limiting" here in Ohio because we're not allowed to say the other one [...] Yeah so when I say "life-limiting" I mean like "incompatible with life." It's been ingrained in me here, they don't like the other one [...] here they will do a C-section on a t13 so that if the mom wants to spend some time with the baby before it dies. [...] Yeah and we're not unique to it you know there's all kinds when you discuss viability what it is its definition it is very murky and cross in. (MFM 2)*

*I don't even want to use the word lethal, because lethal is a moving target. Like twenty years ago, and I keep telling this to our fellows too, twenty years ago lethal was completely different than lethal is today. And there was that editorial in JAMA pediatrics, did you read that one, about Rick Santorum and his daughter and just the shifting place of the word lethal. (MFM 7)*

*But we always bring termination up as an option whenever there is a severe birth defect that will affect the fetus after birth. Um, particularly if it's lethal or if it's life limiting, is another term that we'll use. People don't really like the term "lethal" as much anymore. Or if it's something where the fetus has a high mortality, the neonate I should say, or the fetus, because a lot of the diagnoses will have a high fetal demise rate as well. (MFM 8)*

### Subjective Viewpoint

In contrast to providers' attempts at objectively describing disability, we identified statements discussing the subjective viewpoints MFMs had surrounding disability. Throughout our discussions on disability, many MFMs referenced the notion of 'perfection' that they feel patients have come to expect in their pregnancies perhaps as a consequence of the connotation a "positive screen" or "positive diagnosis" confers onto a fetus. MFMs referenced this sentiment among patients who view their fetuses as less desirable or less perfect because they carry a particular diagnosis even if, chances are, the fetus would not have been perfect diagnosis or not. As one MFM put it:

*I think first people initially have to grieve that their perfect child isn't perfect. We all have imagined perfection in our children, and it's never true [laughs]. (MFM 3)*

We found that while congenital anomalies and their associated disability are considered to be rare in the general population, MFMs noted that for them they are relatively common in

light of their overall practice population. This is because of the specialized nature of the MFM's practice.

To varying degrees, MFMs referenced what we perceived as general stigma surrounding the concept of disability and what it means to be disabled. One MFM referenced this stigma in the context of patient reactions to diagnosis of an anomaly. In reference to the community she served:

*It is well known in [this] community that if [you] have a child with Down Syndrome they actually get shipped off to like –I don't even know what its called- it's like facilities where these children are just take care of because it's seen as such a bad mark on the family if you have a child with disability. Because like so much of the importance of the female in their culture is for propagation. You know? So if you are seen as having any blight on your record that's a really big deal. And you can be shunned from your community or your husband could turn you out basically. (MFM 4)*

Another discussed this stigma in the context of how pregnancies that do not carry a prenatal diagnosis are considered to be 'normal,' while those that do are not.

*I don't know that I made that uh that very like a cog – “today I'm going to start saying typical.” But I have changed the way I approach things. [...] It just felt normal as a term is so heavy and so judgmental. That I just can't say it anymore, it just doesn't come out anymore. (MFM 7)*

### **3. Physicians Experience**

MFMs detailed their experiences counseling patients as being incredibly emotionally involved. We grouped MFM statements in this cluster into two overarching themes and nine sub-themes that fit under the cluster of physician's experience.

### Defining Moral Role

MFMs frequently brought up their moral role in consultations. In general, we use the term role morality, or the theme, “defining moral role,” to describe statements made by MFMs where they indicate that they see themselves as playing a certain supportive role with the burden of ethical decision making falling on the patient. Thus, MFMs are able to subvert their own ethical reasoning so long as the patient’s ultimate decision is ethically permissible. In their capacity as an MFM, providers viewed themselves as needing to be non-judgmental and unbiased. Consequently, MFMs described themselves as supporting their patients in whatever decision they ultimately made, even if they personally did not agree with it.

When a patient’s choice regarding termination of a pregnancy was not in line with the choice the MFM would have made, many expressed a sense of moral distress. In particular, MFMs mentioned instances where patients terminated their pregnancy for conditions the MFM considered to be not worthy of termination. These included Down Syndrome, cleft lip, ambiguous genitalia, heart defects and clubbed foot.

Despite their own moral distress, MFMs repeatedly discussed their efforts to not judge patients based on the decisions they made regarding their pregnancies following a prenatal diagnosis of anomaly. This was especially true in the instances where they did not feel they would make the same decision as the patient.

*I can't provide the option [of termination] but I can provide the avenue and try not to be judgmental about whether they would consider it or not for whatever indication, if it arises...I would try to be supportive of whichever decision they make. I try not to say [that] I probably would not perform a termination if my child*

*had Down Syndrome, but I have had lots of women that do. So I realize that's my more conservative [opinion]...But I try not to be judgmental or I recognize that's my own belief and it isn't something that's universally held. (MFM 17)*

MFMs also felt that they could not judge a patient's decision because they are not the ones who have to live with the consequences.

Underlying this sense of duty to not be judgmental was the sense that determining what they would do if the situation was reversed was not their role. For instance, a frequent occurrence referenced by the MFMs we interviewed was a patient asking them, during some point in the counseling process, what they (the MFM) would do when faced with a prenatal diagnosis of anomaly. This question usually arose in the context of a patient considering termination. In response, MFMs felt that it was never their place to answer this question.

Our final two sub-themes were pro-choice and pro-genetic testing. We used the theme "Pro-choice" to group statements made by MFMs that expressed pro-choice sentiment. This was generally in relation to a patient's choice to terminate a pregnancy, but MFMs discussed being Pro-choice in the context of management decisions with regards to the patient's pregnancy. Some MFMs expressed strong sentiments towards the value of genetic testing. Specifically, they felt that genetic testing was on balance a positive choice, as it equips patients with valuable information to better manage their pregnancy. As one MFM put it, "knowledge is power, right?" (MFM 4)

### Physician's Emotional Response

All MFMs detailed the emotions they experienced while counseling patients. More often than not, MFMs are put in the position of conveying bad news to a patient. The difficulties they describe in the process relate to their own emotional response when delivering this news. While MFMs emphasized a professional obligation to deliver and explain bad news to their patient, they noted their own emotional response as they coped with the difficulties of such a task.

Despite the emotional difficulties MFMs experienced when conveying bad news to their patients, many expressed a sense of empathy for their patients receiving bad news. While many could not relate to the choices the patients were facing, they did describe a sense of sharing patients' emotional response. Some MFMs described learning from their patients' reactions to bad news, while others discussed connecting with patients on a personal level.

*That was really very educational for me as far as uh motherhood doesn't have to be a lifelong experience. You know like you'll always be a mother but for some people it's going to be a short period and learning how to acknowledge that they are a mom even though their baby may have died or whatever- they have a very, very, premature delivery. And you know it's just, I think people if you've had a loss people don't think of that, you are a mother, you were a mother. Recognizing the life that was is very important. That's something I learned ... That people still remember, they want to remember and they want to celebrate the life that was there even though it ended not the way they intended. (MFM 4)*

*I think the most difficult [thing] emotionally is when you're a doctor and you have these bonds with a family and you feel their struggles in some ways and you feel really bad for them. It just highlights how unfair [things are]. So, like I had a patient today who came to the hospital with severe, severe growth restriction, a baby whose prognosis is abysmal. And she tried 5 IVF cycles and they all failed, you know Like you think to yourself ugh, how is this—ugh. I think that's, that's the worst. (MFM 6)*

That being said, one sentiment expressed repeatedly by MFMs is the fear they have about being perceived as coercive by their patients in the context of termination. Many worried that patients might perceive the professional obligation MFMs felt to offer termination as an option in instances of severe anomaly as a directive to terminate because of said anomaly.

*You know, the doctor told me I should terminate and that's not ever my intention, but that is sometimes all someone can hear when it's brought up for the first time. Um, but my hope is that they feel like they have more information than before they started to help figure out what the best choice is for them and their family. (MFM 14)*

In addition to expressing fears around being perceived as coercive, some MFMs expressed feelings of self-doubt with regards to their own capabilities and how well they were counseling their patients.

*[In some situations] I feel like I must not have explained or counseled well enough for the patient to have a truly good and logical understanding of the situation. Because it seems like they're making decisions that don't, um, from a perspective that doesn't have a good understanding of the consequences. And so then I feel that I myself am to blame for not providing enough education and counseling for them to have a true understanding. (MFM 14)*

*[In reference to a specific case] I think there was also a lot of guilt on my part. Like did I miss something. So that's number one, like did I do this family wrong somehow. You always second guess yourself, that's what we do as doctors, did I somehow fail this family? (MFM 6)*

#### **4. Physician-Patient Relationship**

MFMs discussed how they established positive relationships with their patients in order to improve the delivery of care. In general, most MFMs described their relationship with their patient through their communication patterns and the extent to which decision making was shared between physician and patient. Within this cluster, we identified three themes and

eight sub-themes to describe the physician-patient relationship in the context of prenatal diagnosis and decision making.

### Effective Communication

MFMs discussed the different communication techniques they employed in order to maximize the amount of information patients understood during their appointment in order to further foster the physician-patient relationship. The MFMs we spoke with discussed the fact that the information they were tasked with explaining to patients was often of a very high degree of complexity. In general, many felt that the average patient was not prepared to understand the intricacies of prenatal diagnosis, including the technical aspects of testing, the meaning of results and the implications of given findings. Consequently, they felt it was important to try to convert as much information as possible into layman's terms. Some MFMs described employing simplifying language, while others relied on numbers, and some used drawings and images to explain concepts to patients.

In order to ensure they were employing effective communication methods, some MFMs discussed eliciting patient understanding throughout the counseling session.

*I initially have them parrot some of it back to me just so I can make sure that they have a good understanding. I will ask 'do you guys have a good understanding about the outcomes?' And so throughout the conversation I gauge them on their level of understanding and if not we might have to go back and revisit it. But when I am talking it is not like 10 minutes at a time it's usually like snippets here and there and then gauging their response. I usually ask them if they have any questions in between. In my mind I still organize it kind of the same way you would with a dissertation or a speech with an introduction, basic pathophysiology, stuff like that. [And I] gauge understanding kind of throughout. (MFM 13)*



### Responding to Parental Needs

MFMs further described the process of counseling patients as iterative--changing in response to the needs of the patient in order to ensure they were receiving appropriate counsel. This was especially the case when patients came to their appointment with a spouse or other family member. The MFMs we spoke to felt that many women do not anticipate bad news when bringing family members to their appointments. As such, they described an obligation to ensure the patient was comfortable with family being around to hear bad news before conveying it.

*The patient is really our patient right so I feel that firstly she has to be the one that allows everybody else to be in the room and participate in the counselling and so if that is not something that she is okay with, then we will politely ask for everyone to leave. (MFM 12)*

Moreover, MFMs felt it was important that the patient feel they were dictating the type of care they received as opposed to feeling like the MFM was forcing something (either a test or an intervention) on them. To that end, MFMs felt it was important to only offer patients further testing if patients felt they would act upon that information. If a patient did not feel that a screen or test would change their management, MFMs did not feel it was appropriate to offer it to them.

*Ultimately the way I counsel patients regarding this is...the right answer is whatever you want to do. You need to want the information because if you don't want the information and it's not going to change anything then there really isn't a good reason to do the test. We can do a [screening] ultrasound for you, but we're doing this screening test and we're screening for Down Syndrome--this test could come back positive for Down Syndrome or it might not. I always say there are three different types of people: there's somebody who would say you know what I absolutely need to know because it would completely change things that I do. I say if that is you that's the right answer for you and we will proceed with things. There are people who say you know it wouldn't change anything that I would do but I would just like to be informed to know so that I can plan accordingly. Also the right answer if that's your choice. And then somebody else might say "I don't really need*

*to know I don't want to know if not going to change anything," then you probably should not have this test done. (MFM 2)*

As has already been touched upon, many MFMs described the process of offering termination to patients as being a charged topic. That being said, all MFMs we spoke to felt that they needed to bring up the option of termination at least once during their discussions with their patients. In order to do this, many MFMs utilized a technique we dubbed as "Reactive Values Elicitation," wherein they broach the topic of termination with their patients and, pending their patient's reaction, either continue to discuss termination as an option or abandon discussions of termination all together.

*You tell them [termination is an option], and if they get upset, you know their values. (MFM 15)*

*So, it's obviously very gentle, because you have to sort of get a sense for who the patient is. And usually the patient is the one, it'll be pretty obvious, usually pretty clear if termination is something she's considering or if it's off the table. (MFM 16)*

### Shared Decision-Making

While MFMs avoided prescriptive counseling, many felt that patients needed their help in determining what decision was best for them. In order to assist patients in reaching their ultimate decision, MFMs described themselves as guides.

*I just try to do what I can for the patient. Sometimes it's asking slightly guided questions like, would you want to hold the baby, would you want to see with your own eyes what it is that I'm seeing on ultrasound, would you want pictures? (MFM 4)*

*Most people don't really know what to do and they're looking for some guidance as well. It's a really tight balance...And so, I would say it's hard to give hard and fast rules because a lot of it depends on the conversation at the time and the woman and her partner and what's going on in the room so you get a sense. (MFM 16)*

When discussing their role in the shared-decision making paradigm, all MFMs viewed themselves as informants. In other words, MFMs felt their main role was to ensure patients had all the information necessary in order to make an educated decision regarding the course of their pregnancy.

One consideration some MFMs felt was important to bring to patients is the maternal risk associated with pregnancy. They felt this was an important consideration especially in the instances where women are carrying fetuses with lethal anomalies, as some MFMs felt that if there was no chance of meaningful survival of a fetus, the maternal risk to carrying the pregnancy to term might outweigh the patient's desire to carry their fetus to term.

## **5. Patient's Experience**

MFMs defined their perceptions of the patient's experience while undergoing prenatal counseling according to factors they believed to influence patient decisions and the patient's emotional response to news received throughout the diagnostic process. These factors framed how patients understood the information given to them by their MFMs, and their decision making process. Within this cluster, we identified five themes and six sub-themes.

### Cultural Influence

MFMs identified varying aspects of a given patient's culture as impactful in their decision making process. These ranged from their heritage, their faith and the language they spoke. MFMs identified these factors as intersecting to different degrees in order to influence a patient's ultimate decision regarding termination of their pregnancy.

In general, MFMs felt that religious patients were less likely to consider termination. They spoke of individuals from all major monotheistic religions--Christianity, Islam and Judaism--when illustrating this point. MFMs felt that these patients leveraged their religious beliefs and leaders when deciding how to approach a diagnosis of fetal anomaly. While most felt faith had a positive effect on patient coping and decision making, one MFM spoke of a time where a patient began to question their faith in response to a diagnosis of lethal anomaly.

*I can think of a couple, but probably the one that sticks out most in my mind is I had a patient [...] I don't remember exactly what the diagnosis was, but it was a patient who had a fetus that had some significant structural anomalies and I think had hydrops as well, and she in the mid-trimester was diagnosed with hydrops, her husband was a preacher. A Baptist minister. And you know, they really didn't want to accept what was going on. And they came once and they said, "well we think everything's going to be ok." And we said, "so come back in a few weeks and we'll reassess." They came back in a few weeks, and we told them "no, nothing was any better, there were still significant problems there." And he refused to accept it, he became belligerent, started screaming at the staff, punching the walls, and things like that [...] I think it's because for him, his faith was challenged. And you know, it made him question what he had believed in. So, it was much easier to lash out at the care team than it was for him to sort of accept the reality. (MFM 5)*

### Familial Influence

As previously touched upon, women often come into a counseling session with family in tow. MFMs discussed the influence they thought families had on a patient's ultimate decision regarding termination of a pregnancy. Thoughts were divided into positive and negative categories. On the one hand, MFMs identified the support a family can provide emotionally as being incredibly helpful for women considering termination of a pregnancy. Moreover, they identified familial support as being instrumental for those women deciding to keep a pregnancy with diagnosed anomaly. However, some MFMs felt that friends and

families can be a source of stress for patients who are ashamed about a diagnosed anomaly or the choice to terminate.

*I would also throw in for those that are leaning towards termination, I tell them listen, a lot of people probably know you're pregnant, and if you decide to end the pregnancy, what you should tell them is I lost the baby. It's a true statement and it's vague enough that it covers a lot of potential, and it's also...um...a respect my privacy type statement. I tell them you can tell more to people who you want to tell, but it will shut up most people if you just say I lost the baby. Say 'I don't want to talk about it.' And I say the reason it's true is because you've lost the baby you were dreaming about. This baby is not what you were dreaming about, and that pregnancy is not - it's lost. (MFM 3)*

*So I'd still say probably his or her family more than anyone else. Or what I have seen especially with people with strong social support, they rely very heavily on their family background. And have pretty in-depth discussions to the point where family members sometimes take over those discussions. I think we- our job is to provide them with the tools to make this decision. Then it is the family and the patient that ultimately has the long-term outcomes so hopefully they make it as a group. (MFM 13)*

### Barriers to Care

Throughout our discussions, many MFMs lamented the extent to which financial considerations impacted their patient's ultimate decision. Whether patients were interested in genetic testing, termination, or continuing a pregnancy, MFMs noted that they were limited by the costs of the testing they could receive, whether or not they could afford an abortion, and their ability to support a medically involved child. One MFM commented on the cruelty of a medical system that forces patients to pay out of pocket for termination and saddles them with the costs of labor and delivery in the event that they continue a pregnancy for a fetus with a lethal condition.

*We have [the ability to offer patients terminations] but it's all out of pocket pay [...] Like we [also] have Planned Parenthood here who will do terminations after 20 weeks but once again that's not free of cost.*

*[Later, when discussing a fetus with anencephaly] I didn't bill a professional fee for delivery because I think that's rude, but they'll get the same bill [from the hospital] that they would have if they had a baby they got to go home with. (MFM 2)*

Moreover, MFMs identified two major areas where insurance coverage for their patients is lacking. One is insurance coverage for genetic testing and other advanced methods of prenatal diagnosis. The other was for termination.

*I mean I offer all the patients amniocentesis for you know, obviously the early enough CVS, karyotype, and microarray. If there are certain things that I feel like I should narrow the testing down to. Would offer specific genetic, you know, mutation analysis for a specific disorder. The fact of the matter is, being down here and what I mean here really overall in the south. Insurance coverage for some of the more comprehensive testing like whole exome um and even some of the panel testing is not well covered. (MFM 5)*

*They frequently want to have the termination in the hospital, paid for by their insurance. And when I inform them that they can't have it done in the hospital, and their insurance may or may not pay for it, depending on their insurance, they get really angry. Because in their mind it's not an abortion, and it's not something they'd ever do, it's extenuating circumstances. (MFM 8)*

In addition to financial barriers, many MFMs lamented the restrictions placed on them by their institutions when providing prenatal counseling to a woman with anomaly. This was especially discussed in the context of termination. MFMs discussed limitations their institution placed on them either regarding the extent to which they discuss termination with their patients, or their ability to offer patients termination in house.

*We're limited by practice in a federal hospital where by law we're not allowed to perform a termination unless it's rape, incest, or life of the mother, so regardless of a genetic diagnosis, it's something that I would have to refer them to a civilian provider, but I do bring that up with them. (MFM 17)*

*So, we don't have a full spectrum of options available to carry out with the patient. We're a Catholic non-profit facility. But we do talk to patients about everything from treatment if its amenable to treatment all the way to abortions. [...]But we actually have to...[they don't allow us to] hand out resources around that, they*

*have to actually just take down the phone number we give them [...] We had a handout that we used to give our patients, on available various, variable places to terminate and resources around that. But then Catholic health said that they wouldn't allow that. They said we'd be ok if we had them write down the number or the name. Kind of a silly thing but they don't want it to be publicized that we're necessarily promoting that, so we have to kind of navigate within the system to give the patient all their options. We still do, but it's kind of weird, I guess. (MFM 11)*

### Emotional Response

When discussing patient reactions to a prenatal diagnosis of anomaly, many MFMs highlighted the sense of hope patients maintain surrounding the diagnosis. Specifically, MFMs noted that many patients maintained a hope that their baby's condition was not as bad as predicted based on the diagnosis given by the MFM.

*I think it could be good and I think there's always that chance that you're going to have the miracle baby. Going to have a reality show and everything is going to be happy. and I think that's what these patients are looking out for it I think the chance of that happening is less than 1% but it's not zero. and everybody hopes for that. (MFM 1)*

*[Patient's think] "that won't happen to my kid" (MFM 7)*

*I don't want to take away hope you know if there is reason for hope I will never take that away and often times patients will say "well you know I pray to God that everything will be fine" and that is good. that is fine. I encourage that but it does not take away what I see, and I can share that with them. (MFM 12)*

While prior to the birth of their child, many patients hope for a different outcome, MFMs discussed instances where patients expressed regret at continuing a pregnancy after realizing the extent of complications their child faced.

*I don't know what it is like from the child's perspective. I definitely see it from the parent perspective and it once again a variety of outcomes. I have heard families tell me before if they had to do it again they would not have done it again 6 years-10 years out [...] For some people [continuing a pregnancy] is the right decision and for other ppl they feel external pressures that make them decide [to continue the pregnancy] and [later the way they feel about it] may manifest as regret. (MFM 13)*

### Inability to Understand Child's Condition

MFMs identified many instances where they felt that their patient was unable to understand a given diagnosis of anomaly in their fetus. Some MFMs felt that their patients lacked the background knowledge to understand the basis of genetic testing in general, much less the diagnosis of an anomaly. This discrepancy in patient education level made it extremely difficult for patients to make informed decisions surrounding their pregnancy because MFMs felt they were not prepared to understand their child's condition at baseline.

*That's probably the hardest part in some ways because the vast majority of Americans actually have pretty poor literacy and medical understanding. So there are a lot of people I'm explaining what a chromosome is – they don't understand that you get a set from your mom a set from your dad. So I use colloquialisms or other terms like for example for like the karyotype or the chromosomes I use the word like blueprint or- "this is the, you know, instructions that tells the cell to do what it needs to do" – and that's something that comes with years of doing it. And like observing senior faculty or your senior resident, your senior fellow counsel patients and you pick up on these little terminologies that you can use to convert complex medical concepts to layman's terms. So yeah I think that's one of the hard parts. (MFM 4)*

Some MFMs expressed concerns that patients who spoke another language had difficulties understanding their fetus' diagnosis even in the setting of interpreters. As one MFM described:

*With a language barrier all these difficulties and these grey areas become so much more complicated to communicate. (MFM 12)*

## **6. Termination**

A large part of our discussions with MFMs revolved around the termination of pregnancies in the context of an unfavorable prenatal diagnosis. As has been mentioned repeatedly throughout our results thus far, all MFMs offered the option of termination to all of their patients, and offered additional counseling on the matter if patients requested. That being



said, we identified three unique themes during our discussions on termination that we felt warranted additional exploration.

### Abortion Culture

Many MFMs referenced the overall “abortion culture” of their patient population. We took this to reference the likelihood that patients in their population would strongly consider the option of termination or proceed with a termination in the event of a prenatal diagnosis of congenital anomaly. Per our MFMs, abortion culture varied regionally.

*Most of the time with an abnormal diagnosis, because I practice in NYC where people terminate pregnancies, it's not that unusual. (MFM 16)*

*I think that because termination, pregnancy termination rates are so low in the south for genetic anomalies... (MFM 5)*

*[In the mid-west] they just don't accept it, like, here politically there's tons of barriers to termination in this state, it's very challenging to do that here, so I think there's definitely a political trickle-down for this that that's not an acceptable option in pregnancy, so I think it's a combination of the both of them but I almost think it is more political than religious. (MFM 2)*

### Abortion Tourism

An interesting topic mentioned by many MFMs was that of ‘Abortion Tourism.’ As has already been discussed, many MFMs are limited in their ability to offer patients terminations due to institutional or regional laws restricting abortion access. Consequently, many MFMs counsel their patients that they have the option to travel out of state to get the care that they need. On the flip side, MFMs who practice in areas with fewer restrictions surrounding abortion discussed the abortion tourism from the perspective of receiving an influx of patients seeking terminations from out of state.

### Legal Considerations

Finally, many MFMs brought up the fact that they had to counsel their patients on the legal considerations surrounding pregnancy termination. This was usually in reference to the gestational age limitations for abortion, which varied from state to state.

*I just tell them up to state law, termination is up to 23 weeks and 6 days that's the law if you're considering that let us know we'll get you hooked up with the resources and they kind of leave it at that. If someone is coming to us a little bit later, they've missed the cut off. Then I try and hook them up with the correct resources that would still be viable but those are really few and far between.*  
(MFM 4)

Some MFMs discussed recent implementations of laws in their state surrounding the termination of a fetus with a known diagnosis of Trisomy 21. In these instances, patients could get a termination if they were at a gestational age before the designated cut off in their state for any reason, unless it was discovered that the patient's fetus carried a diagnosis of Trisomy 21.

### **DISCUSSION**

This study is an exploratory qualitative analysis meant to shed light on the experiences of MFMs who sit at the forefront of prenatal genetic screening and diagnosis. Our goal was to gain a better understanding of key attitudes and opinions of MFMs surrounding prenatal genetic testing, disability and termination. The perspectives and experiences of these MFMs provide insight on the pitfalls surrounding our current prenatal screening, diagnostic testing and counseling paradigm, and help us identify areas of needed improvement in order to ensure patients are informed enough to make autonomous decisions regarding the course of their pregnancy, and thus achieve the aim of prenatal consultation as set forth by ACOG. This is of great significance as, while MFMs are not the sole responsible actors in

ensuring these criteria are met for patients, they do hold an important obligation given that they are often either the first provider to discuss prenatal genetic screening and diagnostic options with a patient, or the first to explain the results of a screening test to a patient<sup>28</sup>.

To that end, we identified six thematic clusters that characterize how MFM providers in the US conceptualize prenatal consultations of women with a finding of a fetal anomaly. Each cluster has important impacts on the three major requirements of informed consent set forth by Beauchamp and Childress in their “Principles of Biomedical Ethics.” They are as follows: 1) Patients should be given access to high quality, accurate information about the technology of the testing modality in question and their results; 2) Patients should be given information surrounding life with any of the anomalies being screened for, and implications of a positive result; and 3) Patients should be given both adequate access to prenatal testing and adequate time to discuss prenatal testing with their providers in order to ensure full understanding. Here, we discuss the potential extent to which our findings reflect fulfillment and barriers to fulfillment of these requirements.

### **Methodological Barriers to Care**

As referral providers, MFMs were often not the first to discuss or administer prenatal genetic screening tests to a patient. However, they were often the first to explain the results to a patient referred to them for findings of likely anomaly. In these instances, MFMs found that inconsistencies in practice between themselves and referral providers served as a major barrier to educating patients on the tests they received and the implications of their results. As a result, MFMs felt that patients had not made informed decisions regarding prenatal

screening, thereby highlighting concerns of routinization of prenatal screening identified in other studies<sup>41-43</sup>.

Beyond inconsistencies in practice between MFMs and referral providers, some MFMs identified inconsistencies in practice between themselves and the other MFMs they worked with. These inconsistencies in practice led to patients receiving mixed messages about their options for prenatal diagnosis and the implications of their results. It was unclear if these inconsistencies were a consequence of personal bias or a lack of an established standardized approach to counseling from professional bodies. Addressing the methodological pitfalls raised by MFMs in our study could help improve patient and provider experiences with prenatal consultations through improvement in referral systems and establishing practice guidelines within physician groups.

### **Providing Sufficient Patient Education on Testing Modalities and Results: Straining the Physician-Patient Relationship**

Another major challenge in discussing testing modalities with patients came when trying to explain unclear results. In these instances, MFMs expressed difficulties communicating to patients that there is no way of knowing whether the genetic finding in question would lead to some sort of pathology in their fetus or not. These findings of unclear significance force patients and providers to grapple with uncertainty when trying to make decisions regarding pregnancy continuation.

This uncertainty was further reflected in the diagnosis of known genetically anomalous conditions, like Trisomy 21, where there exists a significant spectrum of outcomes. Indeed,

within Trisomy 21, some children may present as higher functioning than others. The same is true for many other diagnosable genetic anomalies. And yet, patients often seek a clear picture of what exactly to expect when raising a child with an identified genetic anomaly, which MFMs felt unable to provide. Consequently, many MFMs described a sense that the technology available to patients for screening and diagnosing congenital anomaly has far outpaced our understanding of anomaly. This gap in knowledge was identified as a barrier to ensuring patients were making fully informed decisions regarding the course of their pregnancy following a diagnosis of genetic anomaly.

To minimize the ambiguity of genetic findings, MFMs described a number of approaches to ensure patients were appropriately informed about the tests they could receive and the implications of a positive or negative test result. They described creating a set of standard, internal goals for each counseling session in order to ensure they were administering as much information to patients as possible. Moreover, when establishing a diagnosis of anomaly in a fetus, MFMs did not solely rely on genetic results, but discussed using a multifaceted approach that combined imaging with genetic testing to provide patients with more information regarding their pregnancy.

That being said, many MFMs identified a lack of insurance coverage for certain genetic tests as being a barrier to providing patients with as much information about their pregnancy as possible before making decisions regarding management. Indeed, financing of genetic screening and diagnostic tests is complex and variable. And while our current practice model encourages the provision of as much information to patients as possible in

order to ensure informed decision making, not all patients have equal access to the testing modalities needed to make fully informed decisions. Thus, inadequate insurance coverage of prenatal genetic testing may make it difficult for patients to fully understand the implications of a given screening result.

Thus, while many MFMs discussed personal strategies to overcome barriers to educating patients on prenatal genetic testing, our findings highlight the fact that our current testing paradigm cannot provide patients with absolute answers regarding the implications of a positive genetic finding. This is because limitations in our current technology preclude MFMs from giving patients definite answers on the implications of their results. Inconsistencies in referring obstetric provider understanding of genetic testing and screening modalities compound this ambiguity, and make it more difficult for patients to be informed on issues of prenatal screening, diagnosis, and management. Moreover, variable insurance coverage of prenatal genetic testing means that not all patients receive the same amount of information before having to make decisions about their pregnancy course. Ensuring baseline patient and provider education through a universal approach, which can be published by ACOG or SMFM, and ensuring universal coverage of genetic screening and testing for all patients could be ways to act upon our findings.

### **Implications of a Positive Screen: Lack of Meaningful and Consistent Discussions on Life with Disability**

Because the genetic conditions most commonly screened for by current prenatal screening modalities result in some level of disability in the neonate, MFMs are often tasked with

explaining the implications of said disability on the neonate's quality of life. Of course, the term 'disability' encompasses a broad number of conditions despite often being referred to as a monolith. Consequently, it was unsurprising that each MFM we spoke with conceived of disability in a different way. While almost all MFMs described disability along a spectrum, the endpoints of each spectrum varied, with some MFMs thinking of disability in terms of major and minor conditions, others in terms of perceived quality of life, and so on. There was no standardized criteria MFMs used when classifying a condition as a major or minor disability. Most relied on gestalt.

This lack of standardization was of particular importance when discussing what MFMs considered to be "major disabilities," "severe disabilities," or "lethal conditions," as MFMs felt obligated to initiate discussions about termination in the setting of both severe and lethal congenital anomalies. We found this lack of standardization regarding definitions of disability, disability severity and lethality incredibly interesting in the setting of the fear of being perceived by patients as encouraging termination that many MFMs described. Indeed, it is possible that the varying thresholds among MFMs for discussing termination with their patients might contribute to the documented patient perception of providers encouraging termination in the setting of a prenatal diagnosis of anomaly<sup>35</sup>.

Beyond having varying definitions of disability severity and lethality, many MFMs described a complete lack of understanding of what it meant to be disabled, and to live with and care for a child with a disability. Consequently, many struggled to provide this information to patients when prompted. However, the majority of MFMs we spoke to

described good relationships with Pediatricians who they could refer their patients to in order to provide them with information on life with a disability.

To our knowledge, our findings of a lack of a standardized approach for discussions of disability severity and termination are the first of their kind. Moreover, they highlight the need for professional bodies to establish guidelines for MFMs to follow when initiating conversations about termination in the setting of congenital anomaly. Without the establishment of a standardized approach to these discussions, it is impossible to evaluate whether or not patients are making truly informed decisions regarding termination in the setting of a prenatal genetic diagnosis.

### **Focus on the Patient Experience: Ensuring Patient Understanding of Screening Results**

The final criteria of informed consent is ensuring patients “understand at least what a health care professional...believes a patient...needs to understand in order to authorize intervention”<sup>14</sup>. In order to ensure patient understanding of prenatal screening and testing modalities, as well as the implications of a given result, MFMs discussed a myriad of tactics they employed in their practice. These included adopting a standardized/consistent approach to patient counseling and engaging in repetitive counseling as much as possible. For many, this meant scheduling multiple follow up appointments with patients in order to give them time to think about the information received, and to come up with additional questions to discuss at subsequent visits.



That being said, MFMs described the lack of time they are afforded to devote adequate attention to counseling patients on their options and results as a major barrier to ensuring patient understanding. This barrier has been previously documented in the literature, highlighting the fact that MFMs do not feel that they are given sufficient time to thoroughly educate patients on issues related to prenatal genetic testing<sup>26</sup>. These findings highlight the need to identify ways in which time allotted to prenatal counseling sessions can be expanded. Possibilities include the expansion of insurance coverage for longer counseling sessions or through more widespread involvement of genetic counselors who can offload the burden of counseling placed on MFMs.

### **Restrictions on Termination also Restrict Discussing Termination**

Within the context of this study, we chose to focus on informed consent as the basis for the Reproductive Autonomy Rationale. And while inadequate understanding serves as a direct barrier to meaningful choice, the meaning of personal autonomy extends beyond this to include the ability to make choices free from controlling interference<sup>52</sup>. And while the MFMs we spoke with did everything in their power to ensure patients were able to make autonomous decisions by providing them with as much information as possible, many described institutional restrictions surrounding discussions of pregnancy termination and legal barriers to access termination services. Consequently, some MFMs reported the inability to engage in open conversations surrounding pregnancy termination because the institution they practiced under prevented them from doing so.

With regards to legal barriers to accessing termination services, many MFMs described instances where patients received diagnosis of lethal congenital anomaly at a gestational

age after a given state's cut off for termination. Consequently, those patients could not choose to terminate even if pregnancy termination was felt to be in the patient's best interest. Moreover, because many insurance providers do not cover termination costs, if patients could not afford to pay for a pregnancy termination out of pocket, the choice to terminate a pregnancy was lost to them. This highlights a concerning disparity in terms of access underscores the need for universal access with coverage for all necessary medical procedures, including pregnancy termination.

### **Understanding the MFM Experience: Informationally engaged, morally detached**

Throughout our discussions, we asked MFMs questions regarding their feelings on the role they play throughout the process of prenatal counseling. MFMs predominantly described themselves as guides for patients navigating the process of prenatal diagnosis, helping to provide patients with the information and resources they needed to make decisions regarding the course of their pregnancy. Interestingly, in order to fill this role as a non-judgmental and unbiased informant, MFMs expressed a sense of role morality. This meant that MFMs felt that their role as an MFM excused them from abiding by their own personal moral reasoning to support the moral decision making of their patients. As a result, in an attempt to remain as unbiased and non-judgmental as possible, many MFMs expressed feelings of moral distress when discussing times where patients made decisions that the MFM did not agree with regarding the course of their pregnancy. Perhaps to alleviate this discomfort, many MFMs attempted to distance themselves from a patient's ultimate decision by refusing to answer patients when asked "What would you [the MFM] do if you were in my shoes?" These responses highlight the efforts made by MFMs to provide

unbiased care to their patients by separating their personal opinions from their professional ones.

### **Strengths and Limitations**

This qualitative study utilized a grounded theory approach to thematic analysis of open-ended semi-structured interviews with MFM providers across the United States. This approach allowed us to identify robust and novel themes to characterize how MFM perceptions of prenatal genetic testing, disability, and termination might influence patient reproductive autonomy. However, there are several strengths and limitations to this study that present opportunities for future investigation.

In using an open-ended and semi-structured approach to interviewing MFMs, we allowed for providers to identify topics they felt were most important when discussing issues of prenatal genetic diagnosis, disability and termination. However, in recruiting participants using the snowball method, we may have limited the variety of perspectives explored in this study by only speaking to providers from a similar social network. Moreover, because the majority of providers we spoke to practiced in major academic centers, the perspectives of private practitioners and practitioners in rural settings is missing from our work. Additionally, this sample is ultimately composed of MFMs who were willing to participate and discuss their experience, which might result in responder bias. Finally, the mix of professional backgrounds within the research team promoted a consideration of a variety of interpretations after thorough scrutiny of data. However, we did not include a general OBGYN or Primary Care Physician (PCP) in our team. As providers who often refer

patients to MFMs, the perspective of an OBGYN or PCP may have been useful in the analysis of MFM discussions on their role as referral providers.

### **Conclusions and Future Directions**

As the technology behind prenatal genetic screening and diagnosis continues to expand, it is imperative that we understand how the application of these technologies impacts a patient's ability to make autonomous choices regarding the course of their pregnancy. While in many instances information gleaned from prenatal genetic testing can positively influence patient care, the clinical trends and ethical concerns identified in this study indicate there is room for significant improvement regarding counseling, informed decision-making and access to care.

While providers supported the use of prenatal genetic screening and diagnostic tests, significant issues emerged at the frontline of integration of these tests into clinical practice. These include: 1) Minimal patient education on screening tests by referring providers; 2) Inconsistencies in practice among administering providers; 3) Limitations in our current screening and diagnostic technology that prevents us from helping patients to understand the implications of a diagnosis; and 4) Variable insurance coverage for more comprehensive genetic testing.

Moreover, here we discussed the novel finding of a lack of standardization of provider understanding of disability, disability severity, and lethality. Because these definitions often serve to frame discussions of termination between patient and provider, it is

imperative that professional bodies establish standardized definitions of these concepts in order to ensure uniformity of patient care and informed consent.

That being said, providers highlighted serious structural barriers to ensuring informed consent and autonomous decision making among patients. These include inadequate time to explain screening and testing options, and limited access to abortion services. Unless both are addressed, it is difficult to imagine a way in which all patients can make truly autonomous decisions regarding the course of their pregnancy following a prenatal diagnosis of genetic anomaly. It is imperative that women carrying fetuses with similar diagnosis have access to equal quality of care.

As the technology surrounding prenatal screening and diagnosis continues to evolve, so must the informed consent process and the resources available to patients making decisions. Addressing the ethical issues discussed above can help MFMs maximize the advantages of prenatal genetic testing for patients and minimize barriers to reproductive autonomy. We hope this study serves as the foundation for future studies aimed at establishing more comprehensive guidelines for MFMs to rely on when discussing issues related to disability and termination with patients, with the ultimate goal of ensuring more equitable healthcare outcomes for women undergoing prenatal consultation for fetal anomalies.

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