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UNIVERSITY OF CALIFORNIA,
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Assessing Referring Provider Identification of Genetic Risk Factors Among Prenatal Genetic Counseling Patients

THESIS

submitted in partial satisfaction of the requirements
for the degree of

MASTER OF SCIENCE

in Genetic Counseling

by

Emily Gao

Thesis Committee:
Professor Manuel Porto, Chair
Adjunct Professor Pamela Flodman
Assistant Professor Linda Freedkin

2026

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DEDICATION

To

My childhood self,

Who could have never imagined taking on this journey,

Thank you for never giving up.

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ABSTRACT OF THE THESIS

Assessing Referring Provider Identification of Genetic Risk Factors Among Prenatal Genetic Counseling Patients

by

Emily Gao

Master of Science in Genetic Counseling

University of California, Irvine, 2026

Professor Manuel Porto, Chair

Genetic counseling plays an increasingly important role as genetic testing becomes more prevalent in healthcare. However, patients who have a personal or family history of genetic risk factors and would benefit from having a genetic evaluation are not always referred. These genetic risk factors that are overlooked can be considered missed indications for genetic counseling. A retrospective chart review was conducted on patients who were seen in the prenatal genetic counseling setting to determine the frequency of missed indications and assess if there are factors associated with a higher likelihood of having missed indications. The results of this study indicated that approximately a third (36%, n=105) of the patients seen had one or more missed indications present. Patients who preferred English and patients who were under the age of 35 were significantly more likely to have a missed indication found compared to patients who preferred a language other than English and patients who were age 35 or older. Of the missed indications found, excluding advanced maternal age (AMA), the most common indication was a family history of cancer followed by a family history of a Mendelian condition, and a family history of intellectual disability/autism. In comparison to previous studies, the frequency of

missed indications remains similar. Ultimately, this study provides further insight on the ongoing need for continuing education for referring providers regarding indications for genetics referrals.

INTRODUCTION

Genetic counseling plays a key role in helping patients understand their genetic health risks and facilitating informed decision making. While there are clear benefits from receiving genetic counseling, not all patients are able to take advantage of it. It has been reported in multiple studies that genetic counseling is an underutilized tool. A recent study determined that within a cohort of 356 prostate cancer patients who met criteria for germline genetic testing, only 122 received a referral to genetic counseling (Greenberg et al., 2026) These missed opportunities to refer to genetic counseling are not limited to oncology. In the prenatal setting, detailed family and personal history that is elicited has been found to reveal risk factors that would have warranted a genetic counseling referral prior to their current visit. In 2017, a study which reviewed 416 prenatal genetic counseling notes determined that 27% of these patients had an additional risk factor identified during their visit (McClatchey et al., 2018). With genetic technology continuing to improve and genetic counseling becoming increasingly important, further investigation of missed referral indications can provide crucial insights to increasing future accessibility of care.

History of genetic counseling

The term genetic counseling was initially created in 1947 by Sheldon Reed, an American geneticist. During this time, other physicians and scientists were practicing what was called "genetic hygiene". Genetic hygiene was defined by Danish scientist Tage Kemp as "the branch of medical science which aims at preventing the transmission of pathogenic genes from one generation to the next and thereby limits their spread in the population" (*Genetic Hygiene and Genetic Counseling, 1953)***.** To move this branch of medicine away from eugenics and towards

social work, Reed created the term “genetic counseling”. While Reed may have wanted genetic counseling and eugenics to be two separate entities, it should be noted that the history of the two are intertwined.

Throughout history, the aims and methods of genetic counseling were constantly evolving and varied based on provider. In the 1940s to the 1960s, paternalism was common in medicine. Most individuals who provided genetic counseling were well-known physician medical geneticists. With the mindset that “doctors knew best,” some physicians took it upon themselves to take on a substantial role within their patients’ decision-making process. As it relates to medical genetics, this meant strongly advising patients on their reproductive decisions when a genetic condition was suspected in pregnancy. Although this stigmatization of genetic conditions was highly prevalent, there were also providers who believed genetic counseling should be non-directive, solely focused on providing the necessary information for patients to make decisions on their own (*Telling Genes: The Story of Genetic Counseling in America - Alexandra Minna Stern*, 2012).

In the 1970s, the demographic of those providing genetic counseling began to evolve rapidly. Following the creation of genetic counseling master’s programs, the profession shifted from being a male-dominated field led by doctors and scientists, to a female-dominated field led by those with specialized degrees which combined both genetics and psychosocial counseling. Even with this shift in profession demographics, the same contrasting opinions on the purpose of genetic counseling and the level of use of non-directiveness continued to exist. In 2005, the National Society of Genetic Counselors (NSGC) approved the following definition of genetic counseling (Resta et al., 2006):

"Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about inheritance, testing, management, prevention, resources and research.
- Counseling to promote informed choices and adaptation to the risk or condition."

This definition was purposefully drafted in a way that defines the overarching practice goals of genetic counseling rather than listing what a genetic counselor does. As the use of genomic medicine expanded into various aspects of healthcare, there was a need to convey what genetic counseling is to other healthcare providers, hospitals, insurances, etc. By providing this guideline, they sought to increase awareness of the role of a genetic counselor while also allowing flexibility for genetic counselors to personalize their work as needed based on a patient's unique circumstances (Resta et al., 2006). Today, genetic counselors practice in multiple specialties to help interpret a patient's family and personal medical history, provide education around genetic conditions and testing, and counsel patients to make choices that are practical and reasonable for themselves.

Genetic counseling settings

Currently, genetic counselors practice in multiple different specialties including prenatal, cancer, pediatrics, adults, cardiology, neurology, and more. As more gene-disease associations are discovered, this list of specialties continues to expand. In practice, genetic counselors are trained to provide care in all specialties. Many genetic counselors subsequently choose to work

within only one domain. According to a professional survey conducted by the NSGC in 2024, 41% of responding genetic counselors worked in adult cancer, 25% worked in prenatal, and 23% worked in pediatrics (Executive Summary. NSGC., 2024). While the overall objective of genetic counseling within these settings remains the same, there are nuances to the work a genetic counselor does across different specialties.

Cancer genetic counselors help inform patients about their risk of hereditary cancer syndromes based on their family and personal history and facilitate genetic testing for those who meet national guidelines for testing. The many tasks of cancer genetic counselors include providing education, comprehensive risk assessments, psychosocial support, facilitation of genetic testing as well as pre- and post-test counseling. (*Genetic Counseling and Clinical Cancer Genetics Services*, 2000). Although there are a multitude of indications that someone may be at risk for a hereditary cancer syndrome, some common ones include: developing cancers at a young age, having a strong family history of cancer, having a personal or family history of rare cancers, having a family history of a hereditary cancer syndrome, and more. Facilitating genetic testing for these individuals not only impacts the patient themselves but can provide important information regarding family members who are at higher risk of developing cancer as well.

When seeing children in a pediatric setting, genetic counselors often work together with a medical geneticist to evaluate children for genetic conditions or provide ongoing medical management for children who have a diagnosed genetic condition. Initial appointments in the pediatric setting can consist of a physical exam by a geneticist, an in-depth review of symptoms within the child and family members, referrals for other specialties, and recommendations for genetic testing (McGlynn & Langfelder-Schwind, 2020). If the testing reveals a genetic cause for the patient's symptoms, the team may suggest occasional visits ranging from annually to every

few years to ensure that their condition is being managed appropriately by their care team. If testing does not reveal a genetic cause, patients and their families are still encouraged to return for follow-up for future reanalysis of their test results as more research emerges. Continued care with the pediatric genetic counselors and geneticists allows patients to remain up to date regarding crucial medical care.

Much like in the pediatric setting, genetic counselors in an adult setting often work together with a medical geneticist. Patients who are seen in the adult clinic can be individuals who have moved on from the pediatric clinic or they can be individuals who have been newly referred. There are a multitude of possible indications that can lead to a referral to the genetics clinic, including conditions affecting the cardiovascular system, neurological system, skeletal system, and more. Due to the variety of symptoms in multiple organ systems which may be part of a patient's presentation, geneticists and genetic counselors provide important information regarding recommended screenings and specialties that should be consulted (Pletcher et al., 2007).

In the prenatal setting, patients are often referred to the genetic counselor by their obstetrician-gynecologist (OBGYN) for the following indications: family history or personal history of a genetic condition, abnormal ultrasound results, abnormal prenatal screening results, positive carrier screening results, advanced maternal age (AMA), consanguinity, exposure to teratogens, etc. (Pletcher et al., 2007). The structure of a prenatal genetic counseling session can differ from one individual to the next. In general, the counselor will obtain the patient's medical history as well as their family history, discuss any genetic conditions that their personal or family history may suggest, discuss any genetic conditions that relate to the referral indication, provide options for further testing, and answer any remaining questions the patient may have. Prenatal

genetic counselors serve an important role in gathering a comprehensive overview of the family history.

Referrals to genetics

A referral for genetic counseling or to a genetics clinic can come from a primary care doctor or a doctor who is following them for their specific health needs such as an oncologist, or an obstetrician gynecologist.

Genetic testing is not limited to being ordered by genetics providers. However, research has shown that non-genetics providers are often not comfortable with ordering and interpreting genetic testing. These studies have also shown that providers lack confidence when it comes to counseling patients on the risks to help them make health management decisions (Conner et al., 2025) (Truong et al., 2021). This demonstrates the importance of having patients seen by professionals trained in this subject area, including genetic counselors and geneticists.

Previous studies

The current study is based upon the work that has been previously conducted by graduate students at the University of California, Irvine. A study conducted by Van Hoang (2010) titled "Genetic Counseling Following a Positive X-AFP Result: Identification of Additional Genetic Risk Factors Before Prenatal Diagnosis" aimed to assess the identification during genetic counseling of other risk factors (beyond the referral indication) that may affect the health of pregnant individuals or their pregnancies. This was specifically examined in the context of prenatal genetic counseling patients who were seen in clinic due to a positive expanded alpha fetoprotein (X-AFP) screening result provided through the state of California. In the X-AFP

screening program, multiple maternal serum proteins and hormones including alpha fetoprotein (AFP), pregnancy-associated plasma protein-A (PAPP-A) human chorionic gonadotropin (hCG), unconjugated estriol (uE3), and inhibin A (inhA) are analyzed to assess for possible neural tube defects (NTDs) and chromosome abnormalities.

The ability of referring providers to identify these risk factors was assessed. The finding of this study determined that 43% of patients who were referred for a positive X-AFP screening result had an unidentified risk factor when they were seen by the genetic counselor that would have warranted a referral for genetic counseling. The study also revealed possible risk factors that may be associated with higher chance of missed indications (Hoang 2010).

Another study conducted in 2017 aimed to assess the number of missed indications found among patients who were seen in the prenatal genetic counseling setting. In this study, 27% of the 416 charts reviewed had a missed indication found that was elicited by the genetic counselor. These patients that were chosen to be reviewed were those who are considered low risk for other possible genetic risk factors. These women were seen in the genetic clinic solely for first trimester screening pre-test counseling. The most common missed indications found were related to a history of birth defects, family history of intellectual disability, or prior positive carrier screening (McClatchey et al., 2018).

Both studies indicated that approximately a third or more of patients who were seen by a genetic counselor did have a risk factor within their chart that would have warranted a genetic counseling visit other than the current one. The amount of time that has passed since these two studies were conducted leaves a gap in information about whether these missed indication frequencies have improved and what areas of knowledge may need to be focused on. This study aims to assess the current state of missed indications and gaps in genetics referrals.

Expansion of indications for genetic counseling

As in all aspects of healthcare, there are guidelines relating to genetic counseling that help providers determine if they are providing appropriate care for their patients. Since the beginning of genetic counseling, there have been ongoing developments to these guidelines which further integrate genetic counseling into an individual's healthcare.

For example, as more information is being discovered regarding hereditary cancer syndromes and the associated genes, more individuals are qualifying for genetic testing for cancer syndromes.

These changes can also be seen in pediatrics. When it comes to genetic counseling in the context of intellectual disabilities in children, the American College of Medical Genetics (ACMG) published guidelines in 2013 that genetic testing should be discussed with all families and patients with autism spectrum disorder (ASD) (Schaefer & Mendelsohn, 2013). In 2021, ACMG strongly recommended the use of exome sequencing or genome sequencing as a first- or second-line test for pediatric patients with congenital anomalies, developmental delays, or intellectual disabilities (Manickam et al., 2021).

California Prenatal Screening Program (CPSP)

Prenatal screening tests are available to identify pregnancies that are at a higher risk of specific chromosomal abnormalities. Chromosomal abnormalities can consist of duplications of chromosomes, deletions, aneuploidy, or translocations. Chromosomes contain the genetic material that is vital for the processes of human development including, cell differentiation, tissue formation, etc. A typical conception will have 23 pairs of chromosomes, 46 total, with each half of the genetic material coming from the egg and sperm of the parents. When there is a

discrepancy in this number of chromosomes, the growth of the embryo may be disrupted, leading to various birth defects and differences in organ function throughout the lifetime. While screening can help to estimate the risk for specific chromosome anomalies, the common category of anomalies that patients may think of are the aneuploidies. Aneuploidies occur when there are either extra or missing entire chromosomes. One of the more common conditions prenatal screening is available for is Down Syndrome, or trisomy 21. Some pregnant individuals are at higher risk of having a pregnancy with chromosomal abnormalities, including individuals who are of AMA, meaning they are 35 years old or older, and individuals who are known to carry a chromosomal rearrangement or who have a family history of chromosome abnormalities or genetic conditions. However, because chromosomal abnormalities can occur in any pregnancy regardless of maternal age or family history, the American College of Obstetrics and Gynecology recommends that all pregnant individuals should be given the option of undergoing prenatal screening and diagnostic testing (*Current ACOG Guidance* | ACOG, 2026).

Over time, the methods for prenatal screening have evolved. Different methods of screening are conducted at different times during a pregnancy due to the various analytes or features that are being assessed. Ultrasounds can be used as a screening test to identify various abnormalities within different organ systems. In the United States, about 1 in every 33 babies will be born with a congenital anomaly (*Data and Statistics on Birth Defects* | Birth Defects | CDC, 2026). While these abnormalities can be isolated, a congenital anomaly can also represent a characteristic of a more complex genetic syndrome. A screening tool that is offered to pregnant individuals in their first trimester of pregnancy is the nuchal translucency ultrasound. Nuchal translucency ultrasounds are used to measure the fluid filled space at the back of a fetus' neck. This space is a normally occurring feature in all pregnancies, however, if the nuchal translucency

is increased in size, this can indicate more severe dysfunction of other organ systems or an underlying genetic condition. An increased nuchal translucency can be indicative of a chromosomal abnormality. Some of these conditions include trisomy 13, 18, 21, Turner syndrome, and Noonan syndrome. This ultrasound is usually done between 11 and 14 weeks of pregnancy (Salman Guraya, 2013).

Maternal serum screening is another option during pregnancy, typically between 15 and 22 weeks. Maternal serum screening assesses the mother's blood for specific biomarkers (a.k.a. analytes) that can be present at different levels when the pregnancy is affected by certain conditions compared to when the pregnancy is unaffected. The biomarkers typically assessed are hCG, PAPP-A, AFP, inhA, and uE3. The conditions that maternal serum screening helps to identify higher risks for include NTDs, abdominal wall defects (AWDs), trisomy 21, and trisomy 18. As with the nuchal translucency measurement, differences in marker levels can also represent natural variation (Ross & Elias, 1997).

The screening method that most recently became available is cell-free DNA (cfDNA) screening that can be done as early as 10 weeks of pregnancy and onward. CfDNA screening helps determine if a pregnancy is at higher risk for chromosomal abnormalities by analyzing fragments of DNA from the placenta within the pregnant individual's blood. The analysis of these fragments may reveal that there is a higher or lower amount of certain chromosomal regions from the placenta than expected. CfDNA is able to assess risks for various aneuploidies, with the main ones being trisomy 18, 21, 13, and the sex chromosome aneuploidies (SCAs) involving chromosome X and Y (Rink et al., 2025). The previously mentioned screening methods are all designed to adjust risks for various conditions, however, they are not a diagnostic test to definitively determine a diagnosis for the pregnancy. For this reason, it is important to

note that diagnostic testing methods should also be offered to patients (*Current ACOG Guidance* | ACOG, 2026).

In the state of California, there is a statewide prenatal screening program offered to all pregnant individuals. As of September 2022, there are two types of screening offered. For pregnant individuals starting in their first trimester of pregnancy, cfDNA screening is available. The recommended timeline for this screening to be completed through the program is from 10 weeks of pregnancy to the first day of 21 weeks of pregnancy. During the second trimester of pregnancy, maternal serum alpha-fetoprotein (MSAFP) screening is available starting from 15 weeks of pregnancy to the first day of 21 weeks of pregnancy. The California Prenatal Screening Program assesses only the second trimester MSAFP analyte for higher risk of NTDs, unlike the previous X-AFP that was offered (*Education Resources for Healthcare Providers*, 2026). As previously mentioned, results from cfDNA screening are able to adjust risks for multiple chromosome conditions including trisomy 21, trisomy 13, trisomy 18, and SCAs. Discussing these possible results with patients is an integral aspect to the care of the patient as well as the pregnancy. When patients undergo screening through the state program and screen positive, there are follow-up services that can be provided at no additional cost. These services include diagnostic testing, ultrasounds, and genetic counseling to discuss the implications of the screening results and further pregnancy and testing options *(California Prenatal Screening Program Provider Handbook Addendum, 2024)*.

Purpose and aims of this study

The purpose of this study is to assess the current genetics and genetic counseling referral practices of providers in a prenatal genetic counseling clinic of a large academic medical center to determine areas of improvement. The aims of the study include the following:

1. Assess the frequency of missed referral indications among patients referred to the UC Irvine prenatal genetic counseling clinic.
2. Characterize the missed referral indications identified among the cohort.
3. Compare the frequency of missed referrals from the current study to previous studies.
4. Assess possible factors that may be associated with missed referrals.

METHODOLOGY

IRB Protocol

IRB approval for this study was granted by the University of California, Irvine under protocol #5493. The study was reviewed as a Category 5 "Expedited" protocol as it poses "no more than minimal risk to subjects". The application for IRB review was initially submitted on December 9th, 2024, and subsequently approved on January 21st, 2025. The protocol was amended on March 12th, 2025, to include the collection of further demographic data. The amended protocol was approved on April 1st, 2025.

Retrospective chart review

The population included in the study was composed of patients seen in the prenatal genetic counseling clinic at UC Irvine Medical Center between the dates of October 1st, 2022, and November 30th, 2024. These patients were referred to the clinic for the following reasons: abnormal ultrasound findings during pregnancy, positive or inconclusive/no-call cfDNA screening results through the CPSP or outside of the CPSP, and positive MSAFP screening results through the CPSP.

Criteria for missed referrals

Missed referral indications were identified with the consideration of the American College of Medical Genetics published guidelines "Indications for genetic referral: a guide for healthcare providers" (Pletcher et al., 2007). Modifications were made to this guide in order to further specify the parameters that would indicate a missed referral within this study.

Patient demographic data and referral information

To gather demographic data, the patient's genetic counseling visit note and pedigree taken during the visit were reviewed. These document the patient's race and ethnicity, age at estimated date of confinement (EDC), referring provider credentials, referring provider specialty, clinic type, patient preferred language, and referral indication. Patient race and ethnicity was categorized based on the CPSP categorizations. Patients who identified with more than one race or ethnicity were categorized as "Multiracial". Referring clinic type was categorized by "internal" or "external". Internal referring clinics were identified as any clinic within the UC Irvine hospital system, and all other referring clinics were external. For referring provider specialty, the nurse practitioners (NPs) who referred patients were categorized under OBGYN due to those specific NPs working exclusively within the UCI Center for Fetal Evaluation.

Independent review

Charts were chosen at random to be reviewed independently by two reviewers. Each reviewer was given 20 charts to review. From the 40 charts that were independently reviewed, there were discrepancies found in four chart reviews, representing 10% of the 40 charts. These discrepancies were corrected, and two charts (5%) were reclassified to "missed indication" from "no missed indication." This level of reclassification was accepted, and the study analyses were conducted without further review of the remaining charts.

Data analysis

The captured data was analyzed using IBM SPSS Statistics Version 31.0.0.0 (117). The software was used to obtain descriptive statistics and conduct chi-squared tests to assess the

significance of association between categorical variables. For the chi-squared tests, a p value of <0.05 was considered significant; nominal p-values are reported with no correction for multiple comparisons.

RESULTS

In total, 289 charts were included in this study. Overall, the charts were analyzed under two different conditions, one including AMA as a missed indication and one excluding AMA as a missed indication. Table 1 shows the count of charts with missed indications and without missed indications based on various patient demographics, referring provider information, and referring clinic information.

With AMA as a missed indication, it was determined that 60% (n=174) of charts had one or more indications identified at the time of their prenatal genetic counseling appointment while 40% (n=115) had no missed indications. The charts that had missed indications identified consisted of 44% (n=126) with one missed indication, 15% (n=44) with two missed indications, and 1% (n=4) with three missed indications (Table 2). When analyzing the presence of missed indications in relation to patient age including AMA as a missed indication, age was categorized into binary variables: patients who were under 35 years old at EDC and patients who were 35 years and older at EDC. The patients who were under 35 years old had missed indications found in 42% (n=77) of charts, while patients who were 35 years and older had missed indications found in 93% (n=97) of charts (Figure 1). This difference was statistically significant (χ^2[1 d.f.] = 74.124, p = <0.001).

When AMA was not included as a missed indication, it was determined that 36% (n=105) of charts had one or more indications identified at the time of their prenatal genetic counseling appointment while 64% (n=184) had no missed indications. The charts that had missed indications identified consisted of 30% (n=88) with one missed indication, 5% (n=14) with two missed indications, and 1% (n=3) with three missed indications (Table 3).

The subsequent analysis of results was focused on the data set excluding AMA as a missed indication.

Table 1: Patient demographics and referral information in relation to presence of missed indications.

	Count of charts (%)	**Missed indication found (including AMA)**		**Missed indication found (excluding AMA)**	
		Yes (%)	No (%)	Yes (%)	No (%)
Patient Age EDC (Years)					
18-34	185 (64%)	77 (42%)	108 (58%)	77 (42%)	108 (58%)
35-46	104 (36%)	97 (93%)	7 (7%)	28 (27%)	76 (73%)
Patient Race/Ethnicity					
White	40 (14%)	30 (75%)	10 (25%)	18 (45%)	22 (55%)
Black or African American	5 (2%)	3 (60%)	2 (40%)	3 (60%)	2 (40%)
Asian	65 (22%)	41 (63%)	24 (37%)	18 (28%)	47 (72%)
Hispanic/Latinx	124 (43%)	61 (49%)	63 (51%)	40 (32%)	84 (68%)
Middle Eastern	15 (5%)	9 (60%)	6 (40%)	6 (40%)	9 (60%)
Multiracial	40 (14%)	30 (75%)	10 (25%)	20 (50%)	20 (50%)
Referral Indication					
Abnormal Ultrasound	138 (48%)	80 (58%)	58 (42%)	54 (39%)	84 (61%)
Positive Screening	100 (35%)	59 (59%)	41 (41%)	27 (27%)	73 (73%)
No call/No results/inconclusive/low fetal fraction screening	51 (18%)	35 (69%)	16 (31%)	24 (47%)	27 (53%)
Referring Provider Specialty					
OBGYN	173 (60%)	107 (62%)	66 (38%)	58 (34%)	115 (66%)
MFM	109 (38%)	61 (56%)	48 (44%)	42 (39%)	67 (61%)
Other[1]	7 (2%)	6 (86%)	1 (14%)	5 (71%)	2 (29%)
Referring Provider Credentials					
MD	265 (92%)	155 (58%)	110 (42%)	94 (35%)	171 (65%)
DO	7 (2%)	6 (86%)	1 (14%)	5 (71%)	2 (29%)
NP	16 (6%)	12 (75%)	4 (25%)	5 (31%)	11 (69%)
PA-C	1 (<1%)	1 (100%)	0 (0%)	1 (100%)	0 (0%)

[1] Other specialties include Family Medicine (n=4), Otolaryngology Head/Neck Surgery (n=1), Pediatrics/Clinical Genetics (n=1), and PA-C No Specialty (n=1)

Clinic Type					
Internal	169 (58%)	97 (57%)	72 (43%)	65 (38%)	104 (62%)
External	120 (42%)	77 (64%)	43 (36%)	40 (33%)	80 (67%)
Preferred Language					
English	251 (87%)	159 (63%)	92 (37%)	97 (39%)	154 (61%)
Chinese	1 (<1%)	0 (0%)	1 (100%)	0 (0%)	1 (100%)
Vietnamese	10 (3%)	5 (50%)	5 (50%)	3 (30%)	7 (70%)
Spanish	26 (9%)	10 (38%)	16 (62%)	5 (19%)	21 (81%)
Arabic	1 (<1%)	0 (0%)	1 (100%)	0 (0%)	1(100%)

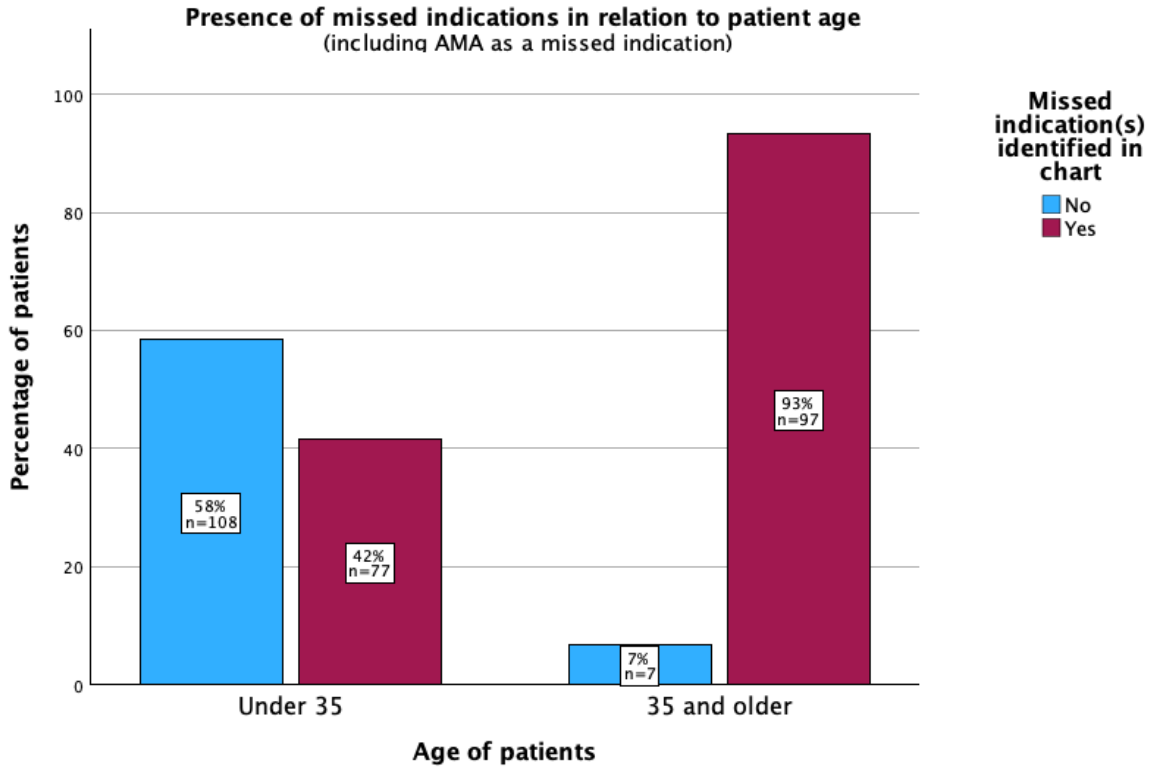


Figure 1: Bar graph depicting the percentage and count of patients with and without missed indication found based on ages under 35 years and ages 35 years and older, including AMA as a missed indication. A statistically significant association is observed (χ^2[1 d.f.] = 74.124, p = <0.001).

Table 2: Count of charts based on number of missed indications (including AMA as a missed indication).

Number of missed indications found	Count of charts (%)
No missed indications	115 (40%)
One missed indication found	126 (44%)
Two missed indications found	44 (15%)
Three missed indications found	4 (1%)
Total Count	289

Table 3: Count of charts based on number of missed indications (excluding AMA as a missed indication).

Number of missed indications found	Count of charts (%)
No missed indications	184 (64%)
One missed indication found	88 (30%)
Two missed indications found	14 (5%)
Three missed indications found	3 (1%)
Total Count	289

Examining the specific missed indications identified

Risk factors that were identified as missed indications were categorized according to the 2007 published guidelines and recommendations from the American College of Medical Genetics (Pletcher et al., 2007). Table 4 shows the count of charts based on the specific missed indications that were found. When including AMA as a missed referral indication, it was noted as a missed indication in 32% (n=93) of charts. Family history of cancer was the second most common missed indication, being identified in 13% (n=39) of charts. Family history of a Mendelian condition, family history of intellectual disability and autism, and teratogen exposure during pregnancy follow close behind, being identified in 7% (n=19), 6% (n=16), 6% (n=18) of charts, respectively. Positive carrier screening results and family history of birth defects were found as missed indications in approximately 4% (n=13, n=11) of charts. Two percent (n=5) of patients had a history of low MCV as a missed indication found. A history of a chromosome abnormality in a previous pregnancy was a missed indication in 1% (n=3) of charts.

Consanguinity was a missed indication in fewer than 1% (n=1) of charts, and early hearing loss was identified as a missed indication in fewer than 1% (n=1).

Table 4: Count of charts based on specific missed indication found.

Specific missed indication found	Count of charts (%)
AMA[2]	93 (32%)
Cancer	39 (13%)
FHx[3] of Mendelian Condition	19 (7%)
FHx[3] of ID or Autism	16 (6%)
Positive Carrier Screening	13 (4%)
Teratogen Exposure	18 (6%)
FHx[3] of Birth Defect	11 (4%)
Personal Hx[4] of Recurrent Pregnancy Loss	4 (1%)
Cardiac	4 (1%)
Personal Hx[4] of Low MCV[5]	5 (2%)
Hx[4] of Chromosome Abnormality in Previous Pregnancy	3 (1%)
Consanguinity	1 (<1%)
Early Hearing Loss	1 (<1%)

[2] Advanced maternal age
[3] Family history
[4] History
[5] Mean corpuscular volume

Assessing the relationship between age at EDC and the presence of missed indications

The age distribution of the patients is shown in Figure 2. The average age of the cohort was 32 years with a standard deviation of 5.8. The minimum age at EDC in this study was 18 years and the maximum was 46 years. For further analysis, age at EDC was split into two categories, those under 35 years and those 35 years and over. Patients who were under 35 had one or more missed indications identified in 42% (n=77) of 185 charts. Patients who were 35 and over had one or more missed indications found in 27% (n= 28) of 104 charts (Figure 3). This difference was statistically significant (χ^2[1 d.f.] = 6.218, p = 0.013).

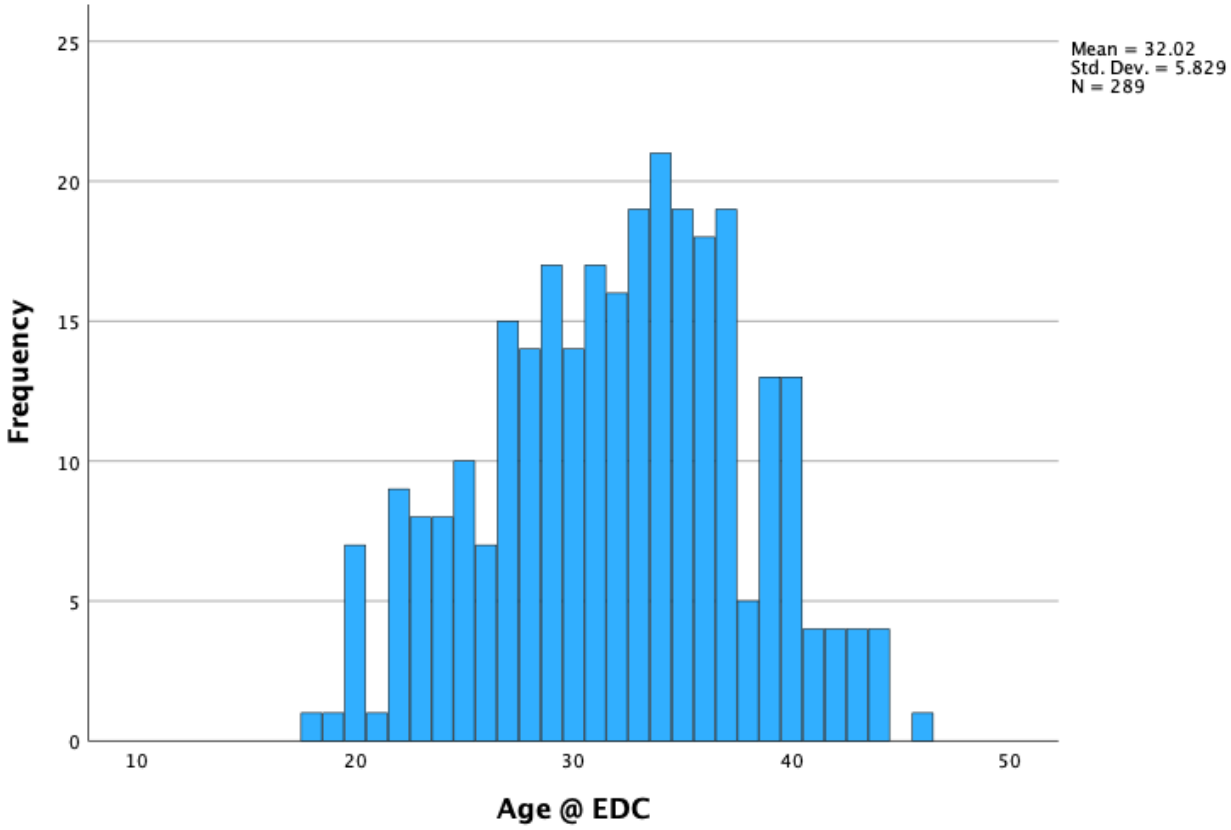


Figure 2: Histogram depicting the distribution of patients' ages within the study. (n=289, Mean=32.02, Standard Deviation=5.829, Minimum=18, Maximum=46).

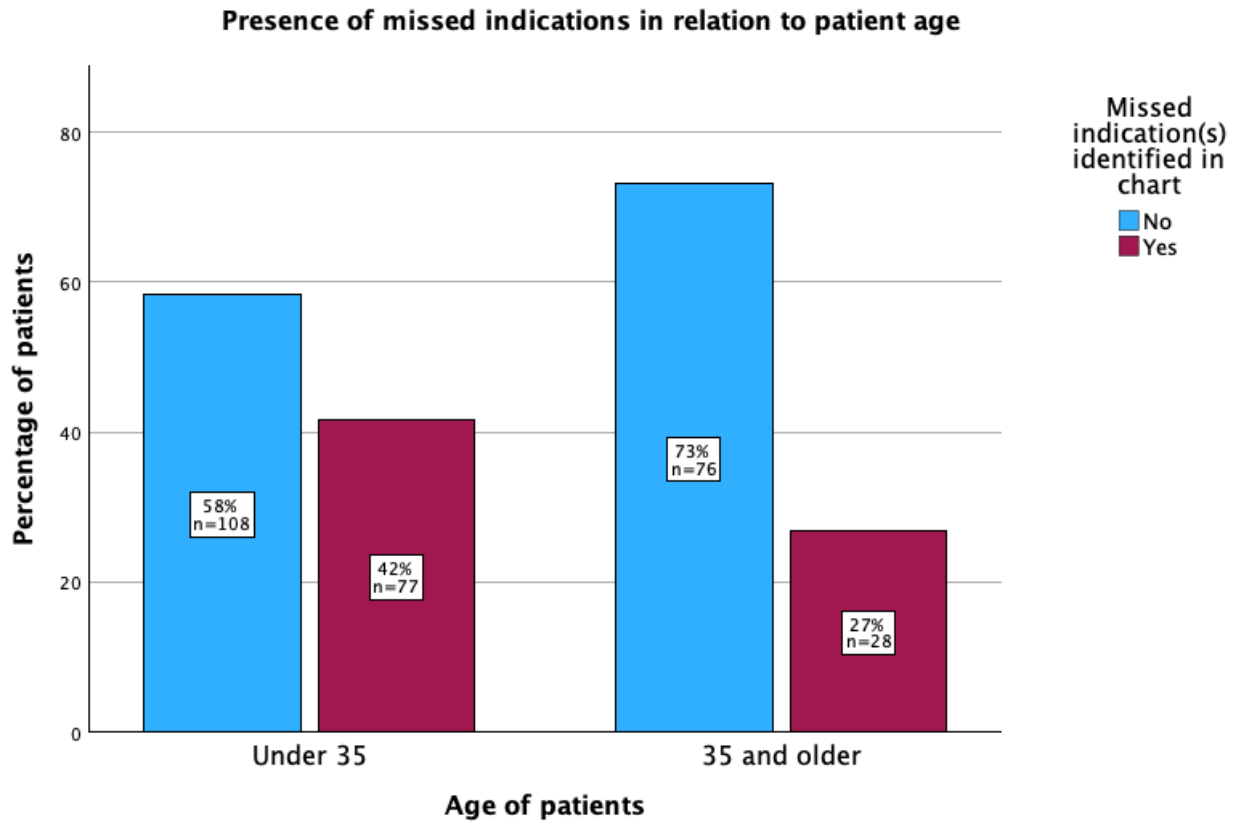


Figure 3: Bar graph depicting the percentage and count of patients with and without missed indication found based on ages under 35 years and ages 35 years and older. A statistically significant association is observed (χ^2[1 d.f.] = 6.218, p = 0.013).

Assessing the relationship between race and ethnicity and presence of missed indications

The racial and ethnic backgrounds of the patients were comprised of 14% (n= 40) White, 2% (n= 5) Black or African American, 22% (n= 65) Asian, 43% (n= 124) Hispanic/Latinx, 5% (n= 15) Middle Eastern, and 14% (n= 40) Multiracial (Table 1). Missed indications were analyzed between "White" and "Non-White" and "Hispanic" and "Non-Hispanic." Missed indications were found in 45% (n= 18) of charts for individuals in the White group and 35% (n= 87) of the individuals in the non-White group (Figure 5); this was not a statistically significant difference (χ^2[1 d.f.] = 1.508, p = 0.219). Missed indications were found in 32% (n= 40) of

individuals in the Hispanic group and 39% (n= 65) of individuals in the non-Hispanic group (Figure 6); this was not a statistically significant difference (χ^2[1 d.f.] = 1.558, p = 0.372).

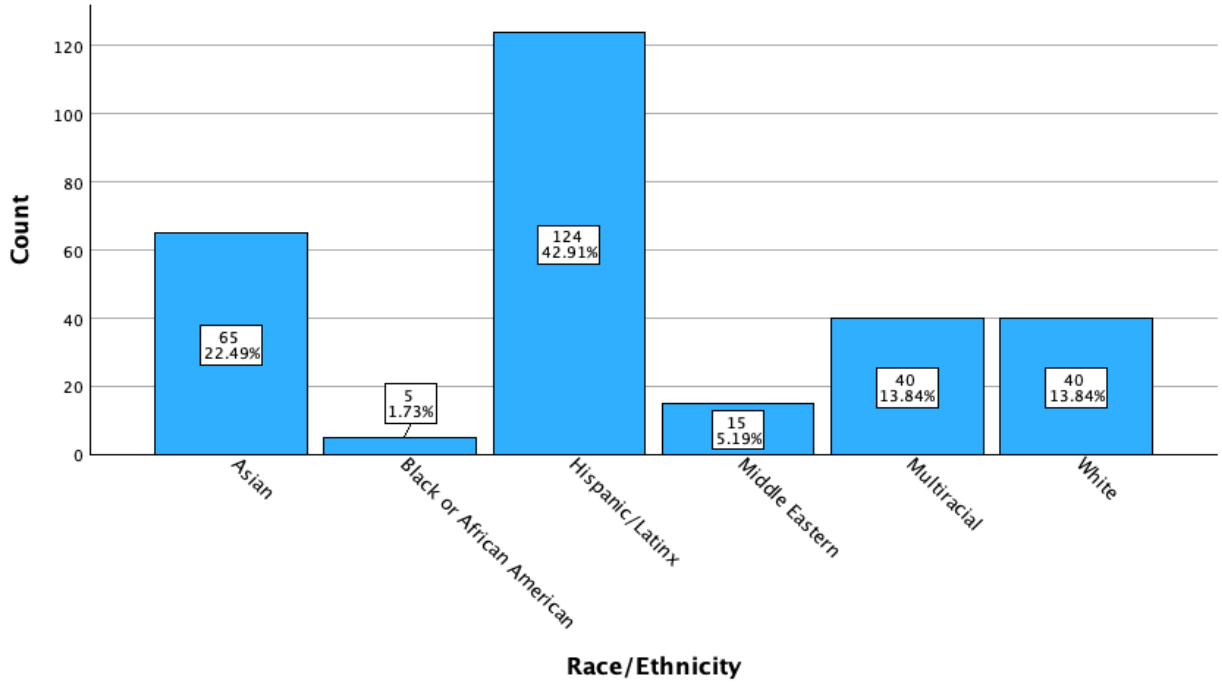


Figure 4: Bar graph depicting the percentage and count of patients based on their racial and ethnic identity.

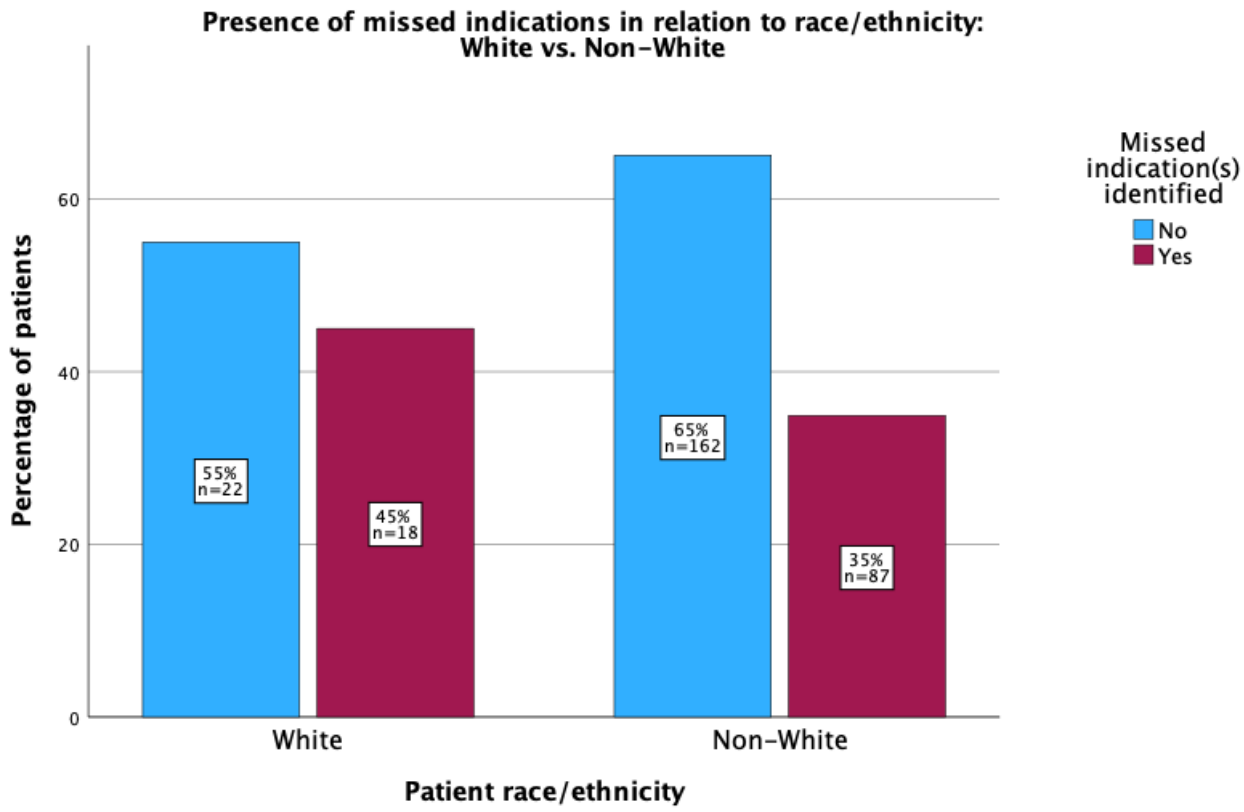


Figure 5: Bar graph depicting the percentage and count of patients with and without missed indications based on their racial and ethnic identity, specifically if they identify as White or Non-White. No statistically significant association was found (χ^2[1 d.f.] = 1.508, p = 0.219).

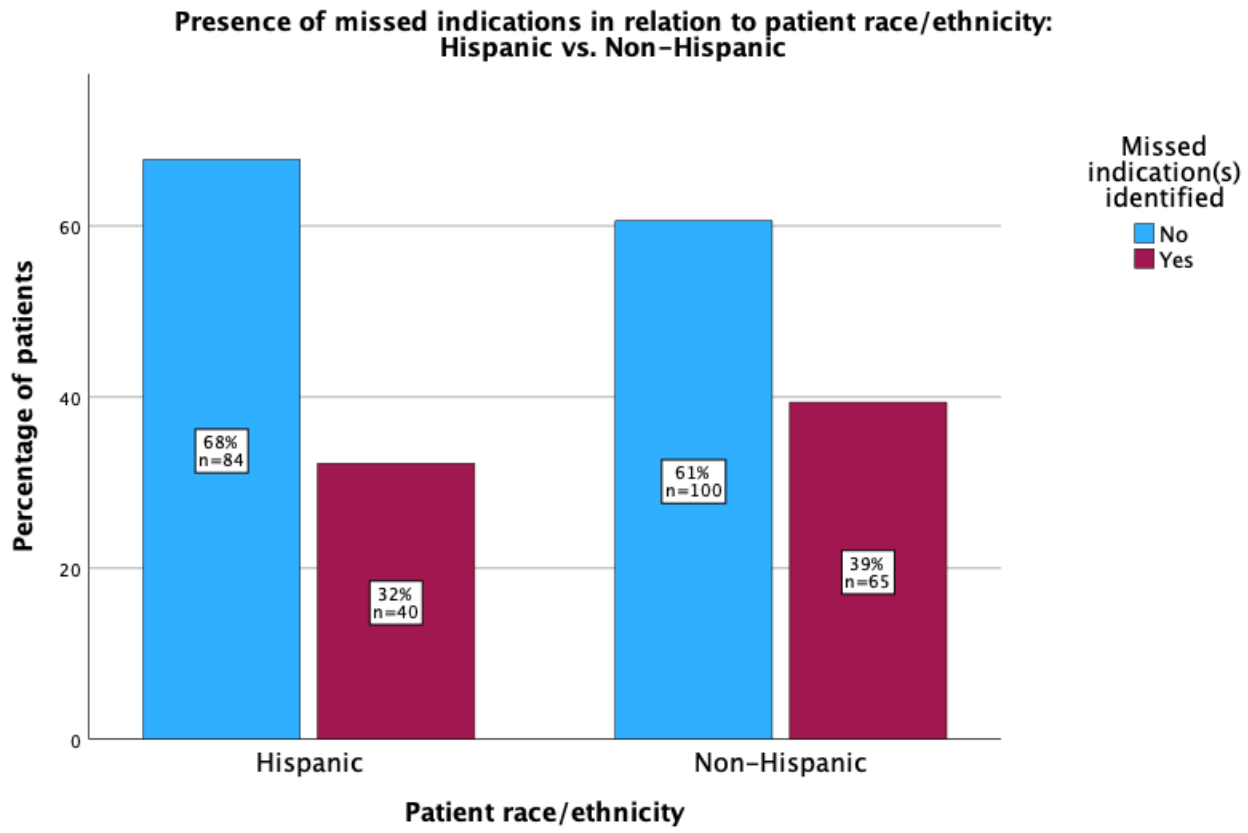


Figure 6: Bar graph depicting the percentage and count of patients with and without missed indications based on their racial and ethnic identity, specifically if they identify as Hispanic or Non-Hispanic. No statistically significant association was found (χ^2[1 d.f.] = 1.558, p = 0.372).

Assessing the relationship between referral indication and presence of missed indications

The referral indication to prenatal genetic counseling varied within the cohort. Forty-eight (n= 138) of patients were referred due to an abnormal ultrasound. Thirty-five percent (n= 100) were referred due to positive screening results from cfDNA screening or MSAFP screening. Eighteen percent (n= 51) of patients were referred due to "No Call", "No Results", "Inconclusive", or "Low fetal fraction" screening results (Table 1). The two categories of screening results were combined to create a binary referral indication variable. Thirty-nine percent (n= 54) of patients who were referred due to abnormal ultrasound had one or more missed indications found in their charts, and 34% (n=51) of patients who were referred due to a

screening result had one or more missed indications found in their charts (Figure 7). Referral indication was not a statistically significant variable in relation to identification of missed indications (χ^2[1 d.f.] = 0.894, p = 0.344).

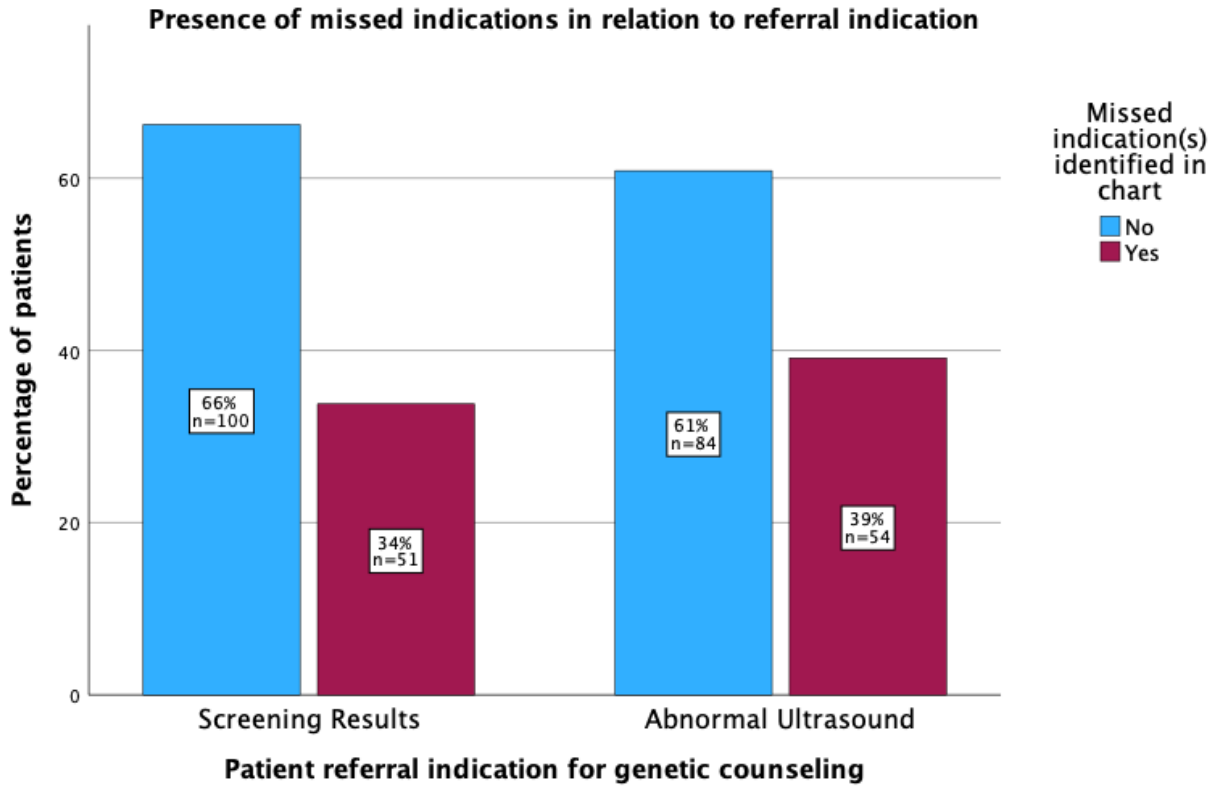


Figure 7: Bar graph depicting the percentage and count of patients with and without missed indications based on their referral indication. No statistically significant association was found (χ^2[1 d.f.] = 0.894, p = 0.344).

Assessing the relationship between referring provider credentials and presence of missed indications

Within the cohort, 92% (n=265) of referring providers were Doctors of Medicine (MDs), 2% (n=7) were Doctors of Osteopathic Medicine (DOs), 6% (n=16) were Nurse Practitioners (NPs), and <1% (n=1) was a Physician Assistant (PA-C) (Table 1). The two credentials, MD and DO, were combined to create a binary variable for analysis. All other credentials were

categorized as “Other”. For patients who had referring providers who were MDs or DOs, 36% (n=99) had one or more missed indications in their charts. For patients who had a provider with credentials other than MD or DO, 35% (n=6) had one or more missed indications in their charts (Figure 8). There was no significant difference in missed indications based on referring provider credentials (χ^2[1 d.f.] = 0.008, p = 0.927).

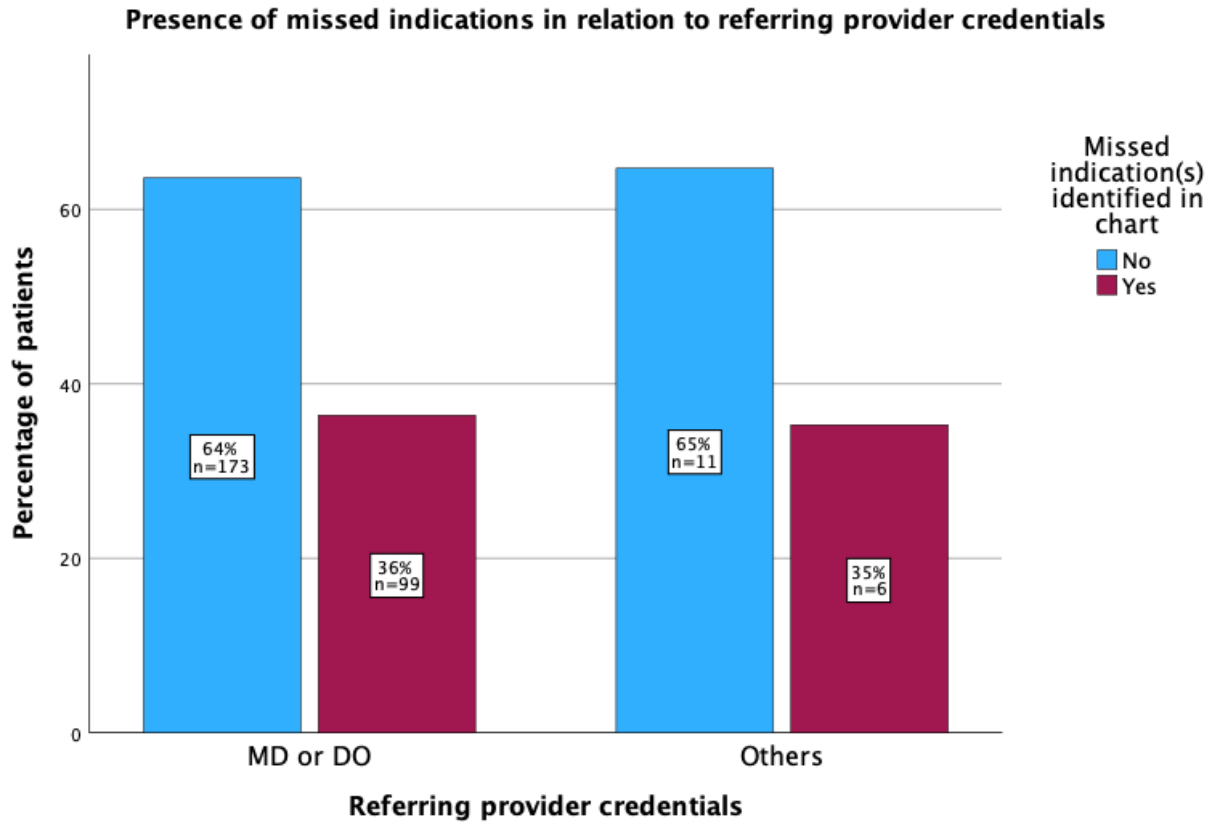


Figure 8: Bar graph depicting the percentage and count of patients with and without missed indications based on their referring provider credentials. No statistically significant association was found (χ^2[1 d.f.] = 0.008, p = 0.927).

Assessing the relationship between referring provider specialty and presence of missed indications

Most referring providers were either specialized as OBGYNs (60%, n=173) or Maternal-Fetal Medicine specialists (MFMs) (35%, n=109). Other referring provider specialties were comprised of 1% Family Medicine (n=4), <1% Otolaryngology Head/Neck Surgery (n=1), <1% Pediatrics/Clinical Genetics (n=1), and <1% No Specialty (PA-C) (n=1) (Table 1). Missed indications were examined in two groups: patients referred by providers who were MFMs, and patients referred by providers other than MFMs. Thirty-nine percent of patients referred by MFMs had the presence of missed indications within their charts while 35% of patients who were referred by providers in other specialties had the presence of missed indications (Figure 9). This difference was not found to be statistically significant (χ^2[1 d.f.] = 0.366, p = 0.545).

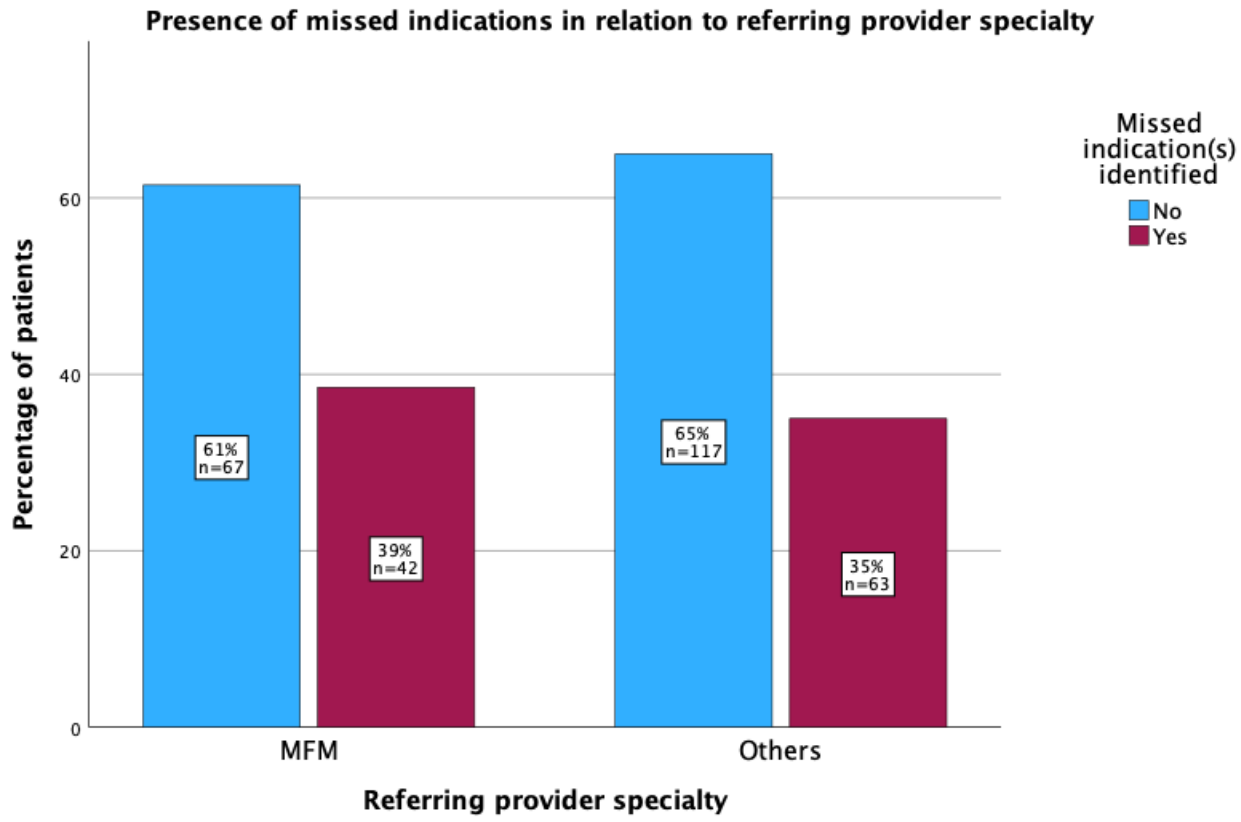


Figure 9: Bar graph depicting the percentage and count of patients with and without missed indications based on their referring provider specialty. No statistically significant association was found (χ^2[1 d.f.] = 0.366, p = 0.545).

Assessing the relationship between referring clinic type and the presence of missed indications

Referrals were categorized based on whether they came from an internal clinic or an external clinic. Fifty-eight percent (n=169) of charts were referred to prenatal genetic counseling from an internal clinic and 42% (n=120) were from an external clinic (Table 1). Missed indications were present in 33% (n=40) of patients who were referred from an external clinic and 39% (n=65) of patients who were referred from an internal clinic (Figure 10). Clinic type was not found to be a statistically significant variable in determining presence of missed indications (χ^2[1 d.f.] = 0.798, p = 0.372).

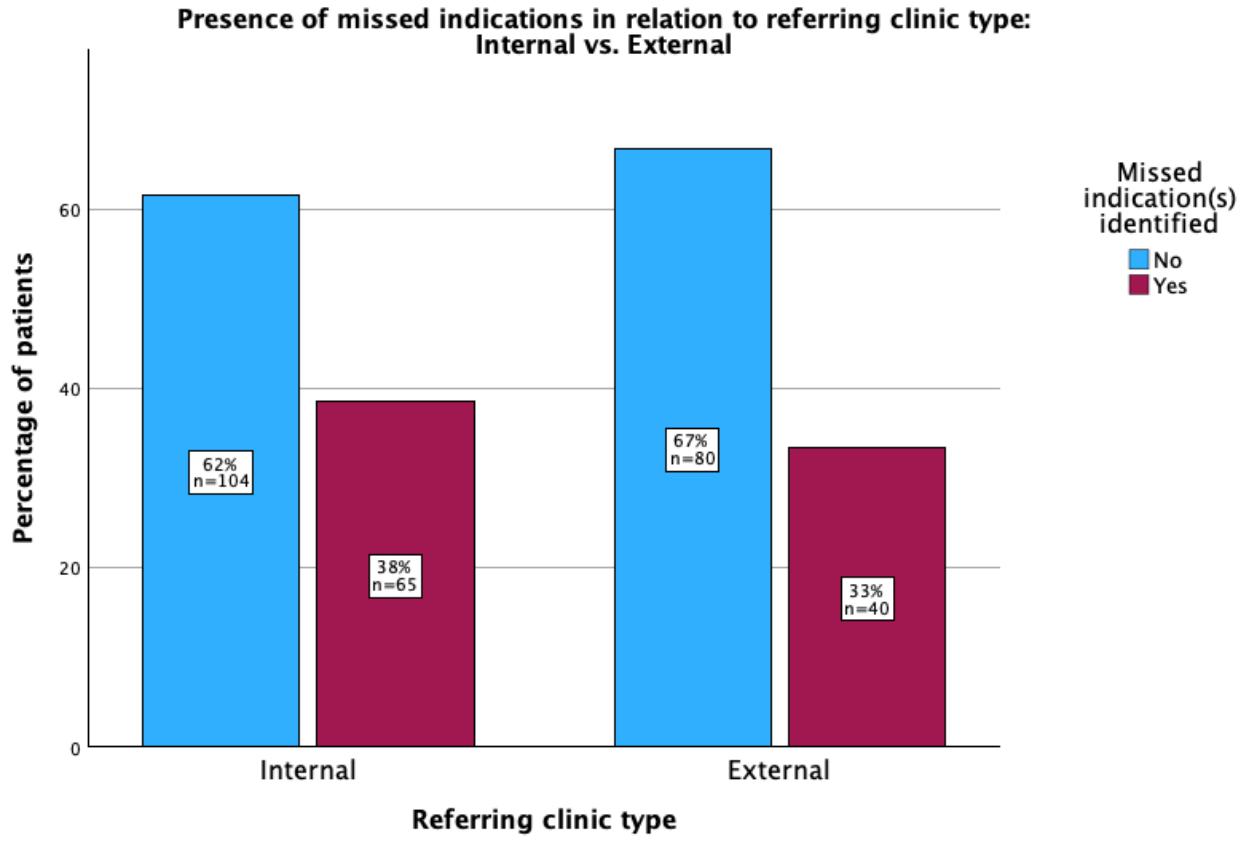


Figure 10: Bar graph depicting the percentage and count of patients with and without missed indications based on their referring clinic type. No statistically significant association was found (χ^2[1 d.f.] = 0.798, p = 0.372).

Assessing the relationship between preferred language and missed indications

English was the preferred language for 87% (n=251) of the patients in this study. For the remaining patients, 9% (n=26) had a preferred language of Spanish, 3% (n=10) had a preferred language of Vietnamese, <1% (n=1) had a preferred language of Chinese, and <1% (n=1) had a preferred language of Arabic (Table 1). Missed indications were analyzed between those who had a preferred language of English and those who had a preferred language other than English. Those who preferred English has missed indications identified in 39% (n=97) of charts. Those who preferred a language other than English had missed indications found in 21% (n=8) of charts (Figure 11). This difference was statistically significant (χ^2[1 d.f.] = 4.416, p = 0.036).

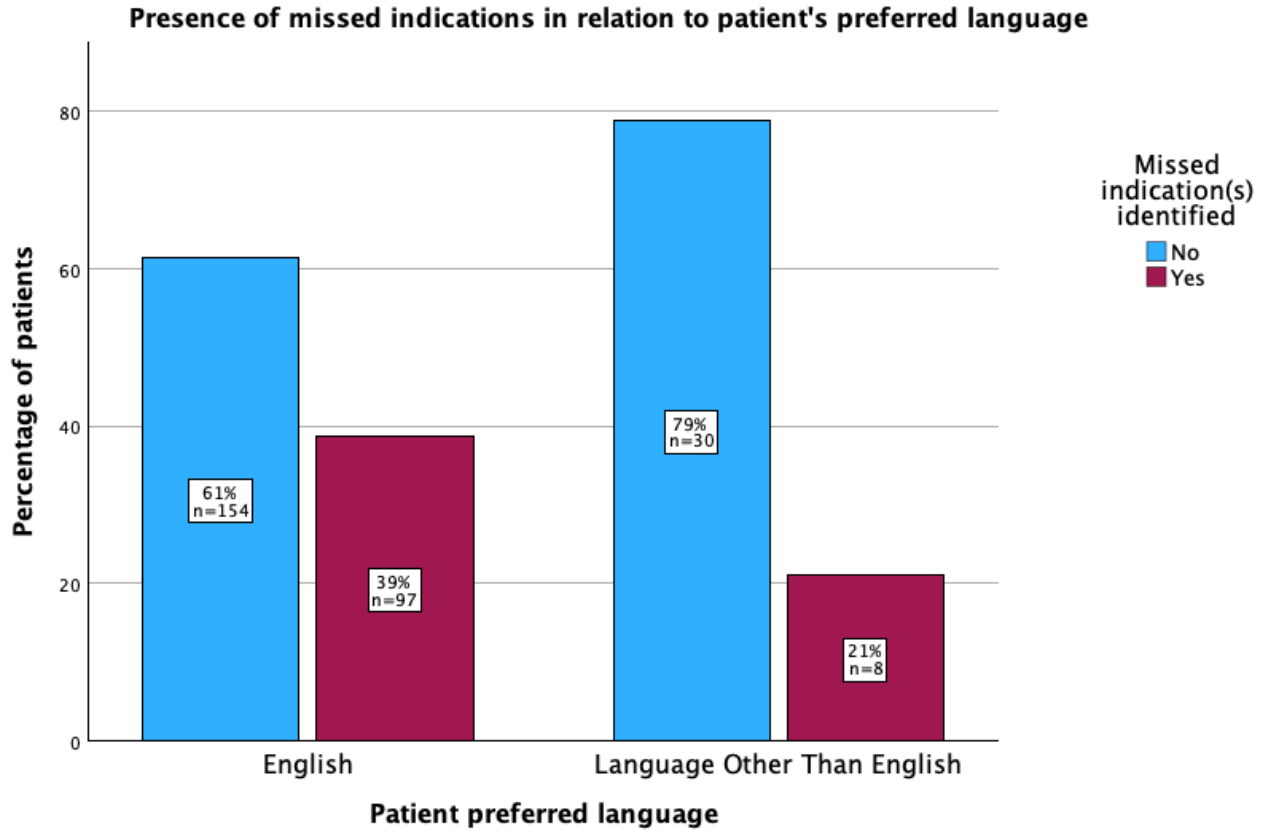


Figure 11: Bar graph depicting the percentage and count of patients with and without missed indications based on their preferred language. A statistically significant association was found (χ^2[1 d.f.] = 4.416, p = 0.036).

DISCUSSION

The primary aim of this study was to determine the frequency of missed indications within patients who were seen in a prenatal genetic counseling clinic. The identification of missed indications in a patient's chart demonstrates that the patient may have benefited from an earlier genetic counseling visit aside from their current one. Risk factors were determined to be considered "missed indications" in this study based on ACMG guidelines (Pletcher et al., 2007). Another aim was to characterize the missed indications identified. This study also sought to examine the possible factors that may be associated with the presence of missed indications to determine what areas of improvement there may be to decrease the number of missed indications for patients moving forward.

The data analysis in this study focused on the data set which did not include AMA as a missed indication. When AMA was included as a missed indication, maternal age (coded as a binary variable defined by whether the patient was at least 35 years old or under 35) was a highly significant predictor of the presence of a missed indication. However, this result does not address the main research aims of this study; the main genetic risk associated with AMA is for chromosomal aneuploidy and there are screening tests for chromosomal aneuploidy available for use in the primary care setting (Gantt et al., 2022). Therefore, it is not surprising that there are women who are in the AMA age group who are not referred for genetic counseling based on that factor alone.

Examining the medical charts of patients who were seen in prenatal genetic counseling (n=289) revealed that approximately a third (36%, n=105) of those patients had a missed indication for genetic counseling that was not identified prior to their current visit. This number is similar to that of the previous study conducted in 2010 by graduate students at the University

of California Irvine. In that study, it was determined that 110 out of 255 or 43% of patients had a missed indication that was identified during their prenatal genetic counseling appointment following a positive X-AFP result (Hoang, 2010). In 2017, a study conducted at Baylor University identifying missed risk factors in prenatal genetic counseling patients found that 27% had a missed indication found (McClatchey et al., 2018). The percentage of patients with missed indications in this study is similar to the percentage found in these previous studies. This suggests that within the last 10 to 15 years, there has not been a substantial improvement in referring patients to genetic counseling prior to their visit during pregnancy. Both studies excluded women who were 35 years and older, thus excluding AMA as a possible missed indication. The current study included women who were 35 years older, which provided the opportunity to assess the frequency of missed indications in this age group. However, as previously mentioned, the data analysis was focused on missed indications excluding AMA.

Referring provider credentials were not seen to have a significant impact on the identification of missed indications. Provider credentials in this study included MD, DO, NP, and PA-C. It was expected that patients who were referred from MDs and DOs would have a lower likelihood of having a missed indication. Given the longer training period for those who earn a degree of MD or a DO, the hypothesis was that they may have a greater ability to identify patients who should be seen by a genetic counselor; however, the findings in this study do not support this hypothesis. This suggests that regardless of credentials, providers may need further processes in place or continuing education to allow for the identification of patients who should be seen by a genetic counselor.

The referring provider specialty also did not have a significant association with the presence of missed indications within the charts. The composition of provider specialties in this

study included MFM, OBGYN, Family Medicine, etc. Comparing MFM with other specialties, the hypothesis was that there would be fewer missed indications found based on their increased training to handle high-risk pregnancies and an increased awareness of risk factors for genetic counseling. The findings of the study do not support this hypothesis and indicate a similar proportion of missed indications regardless of the specialty of the referring provider. Once again, these results may highlight the need for increased education for referring providers and better methods for identifying patients with risk factors for genetic counseling across all specialties.

Patients were referred to the prenatal genetic counseling setting from either an internal clinic or an external clinic. This factor was not found to be associated with the presence of missed indications in a chart. The hypothesis regarding the effect of clinic type was that there would be more missed indications identified from external referrals. Internal referrals require less extensive coordination compared to external referrals. When a patient is referred from an external clinic, there is an exchange of information that must take place including obtaining various documents and test results prior to the visit. If a hospital system's referral process is not streamlined, referrals can be missed and continuity of care can be interrupted (Leila, 2026). External providers who refer patients may end up missing context that may be important as a referral indication for their patient (in addition to the primary referral indication). This could occur, for example, due to the loss of information in the referral of a patient from one clinical setting to the next. This study does not support the hypothesis that external referrals would have a higher likelihood of having a missed indication. Ultimately, this could be suggestive of an organized external referral protocol within the institution, however more research would need to be conducted on the impacts of internal/external referral and referral protocols on healthcare outcomes.

The patient's race and ethnicity was not significantly associated with the presence of missed indications. Prior to analyzing the data, it was hypothesized that minority patients may have a higher chance of having missed indications. Racial disparities in healthcare have been a long-standing issue in the United States. This can cause negative effects in multiple aspects of healthcare including access to care, quality, patient-physician relationship, and health outcomes. Through a social determinants of health (SDOH) lens, racial and ethnic minorities are more likely to experience economic instability, social discrimination, and adverse environmental conditions, which all shape the way they interact with the healthcare system (Monroe et al., 2023; Vrtikapa et al., 2025). For example, economic instability can be a barrier for an individual to obtain adequate health insurance, a lack of transportation can lead to missed appointments, and living in an area without providers for certain specialties can hinder necessary care (Vrtikapa et al., 2025). Implicit biases also play an adverse role in the treatment of and attitudes toward minority patients (Macias-Konstantopoulos et al., 2023). The findings from this study are not consistent with the existing research on healthcare outcomes and race, since no significant difference was detected in the presence of missed indications. It may be that the location of the study affected resources available to patients. California's Medicaid program, known as Medi-Cal, is health insurance that certain low-income Californians can enroll in. As of July 2026, a report from the California Department of Healthcare Services estimates that over 13 million Californians are enrolled in Medi-Cal. Of those enrollees, most are racial and ethnic minorities. Approximately 51% are Hispanic, 10% are Asian/Pacific Islander, and 7% are African American (Management Analytics Division, 2026). Historically, Medi-Cal has covered around 40% of births in California, providing crucial care for pregnant individuals (*California Transforming Maternal Health* | DHCS, 2026) (*Medi-Cal Facts and Figures Almanac — 2024 Edition -*

California Health Care Foundation, 2024). Although state-sponsored health insurances are not without flaws, Medi-Cal provides increased access to care for individuals who may not be able to afford other options.

Patients who were included in the study were referred due to either an abnormal ultrasound, positive/no call/no result/low fetal fraction cfDNA or positive MSAFP screening results. This study demonstrated no significant association between the patient's referral indication and the presence of missed indications. Both groups of patients, those referred due to ultrasound or screening results, are ones who may not have ever had a genetic counseling appointment if not for their current referral. This finding suggests that this frequency of missed indications may reflect the level for all pregnant patients, regardless of whether they were referred to genetic counseling.

The variables that were seen to be significant in identifying missed indications included the patients' age and preferred language. Patients who were under 35 were more likely to have missed indications identified in their chart. Prior studies have shown that ageism is prevalent in healthcare. A roundtable study interviewing young adults with chronic and rare conditions revealed that young adults had shared experiences of dismissal from providers, delays in diagnosis, and misdiagnoses (Rosewater et al., 2026). Based on this information, it is possible that young adult patients in this study had risk factors that were overlooked due to their age. It has also been shown that one in five patients aged 19 to 25 have gaps in healthcare where they are not seen by a healthcare provider for one to three years (Ilango et al., 2025). This gap in care could contribute to fewer opportunities to speak to a healthcare provider about their health history. As patients become older, there also may be an increased responsibility to act as the family historian and gather this information regarding family members' health history. Younger

patients who may not have this same responsibility could simply be unaware of their familial risk factors.

Preferred language was a factor that was associated with whether missed indications were found. Those who had a preferred language of English had a higher chance of having a missed indication than those who preferred another language. This is consistent with a previous study which demonstrated that patients who required the use of an interpreter were less likely to report a family history of cancer diagnoses in the prenatal genetic counseling setting (Palacios et al., 2025). If the patients who preferred a language other than English utilized an interpreter, it is possible that they were unable to share as much information as those who utilized English in communicating with their providers. The use of an interpreter effectively takes twice the amount of time to convey information than without an interpreter. These patients could have had risk factors in their family histories and were simply unable to convey it during their appointment, leading to fewer missed indications found.

Of the missed indications found, the most common was a family history of cancer. In regard to the patient's current pregnancy, this category of missed indications would not necessarily change any immediate medical decisions that may take place. There are, however, other indications that would be pertinent to the current pregnancy. In this study, we discovered missed indications of Mendelian conditions in the family, including autosomal recessive conditions that cause severe congenital anomalies. That information can be crucial in providing the patient with options and resources prior to and during pregnancy.

There are multiple limitations to note within this study. The small sample size in certain demographics makes it difficult to establish statistical significance, even if a true difference exists. For example, there were few referring providers who had credentials other than MD or

DO, causing a lack of statistic power when comparing the two. The review of medical charts in this study also did not examine concurrent referrals made to other genetic counseling clinics, such as cancer genetic counseling, which could have caused a chart to be incorrectly flagged as having a missed indication. Another limitation of this study could be the bias of having one researcher reviewing the charts to identify missed indications. Although there was an independent review conducted, this review only represented a portion of the data. The independent review identified that 5% of charts were misclassified on initial review with respect to the presence or absence of missed indications. These errors were corrected, and the estimated error rate was accepted before proceeding with further analysis. However, it is possible that there may be further insight which could be gained by independent review of all the charts.

Overall, this study has demonstrated that there is still room for improvement in identifying patients who could benefit from genetic counseling. The percentage of patients with a missed indication found during their prenatal genetic counseling appointment is within the same range as previous studies conducted more than a decade ago. Patients who preferred English were significantly more likely to have missed indications in comparison with those who preferred communicating with their providers in a language other than English. The likelihood of having fewer missed indications identified could be due to barriers in relaying their information within the duration of the appointment due to the need for interpreter services. Patients who were 35 years old and over were significantly more likely to have no missed indications identified, in comparison with those who were under age 35. For the other variables examined, there was minimal further insight on whether they are associated with the presence of missed indications. Future directions based on this research could benefit from focusing on tools that can be

implemented within electronic medical record systems to identify individuals who should be referred to genetic counseling.

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APPENDIX A: Chart Review Rules

Indications for a missed genetics referral (focusing on those with risks to patient and/or pregnancy):

Prenatal/Preconception (Family History is defined as second degree relative to the pregnancy)

- MCV <80
- Maternal age
  - 35 years or older for singleton pregnancy
  - 33 years or older for twin pregnancy
- Consanguineous union between patient and partner
- Personal history of stillbirths, previous child with hydrops, recurrent pregnancy losses (more than 2), or a child with sudden infant death syndrome (SIDS)
- Personal or **family history** of chromosome abnormality
- Personal or **family history*** of significant intellectual disability or autism
  - *up to second degree relative to the pregnancy
- Reported carrier of specific genetic condition(s)
- Personal or **family history** of birth defects (up to 2nd degree relative)
  - One key birth defect suggestive of a specific genetic condition
  - Two or more birth defects in multiple organ systems
  - NTD
  - Cleft Lip/Cleft Palate (if in conjunction with another finding)
  - Significant cardiac defects (i.e., HLHS, but not VSD)
- Personal or **family history** of genetic condition
  - No limitation on degree of family member if there is a reported diagnosis of mendelian condition
- Personal history of progressive neurological condition
- Personal or **family history** of significant hearing or vision loss thought to be genetic in cause
  - Early onset
  - Bilateral
  - No external factors

Adults (Family History is defined as first degree relatives of the patient or partner)

- Personal history of recurrent pregnancy losses
- Personal history of abnormal sexual maturation or delayed puberty
- Personal history of statin-induced myopathy
- Personal history of six or more cafe au lait macules
- Personal history of unusually short or tall stature
- Personal history of birth defects
  - One key birth defect suggestive of a specific genetic condition
  - Two or more birth defects in multiple organ systems

- Personal or **family history** of features suggestive of a connective tissue disorder
  - Except for individuals with symptoms of only hypermobile EDS
- Personal or **family history** of progressive neurological/neuromuscular condition
  - Exception for a reported diagnosis of Huntington's in family members further than second degree relatives
- Personal or **family history** of significant vision loss
  - Unexplained sudden vision loss
  - Retinitis pigmentosa
  - Early onset macular degeneration
  - Early onset cataracts (before age 60)
- Personal or **family history** of early onset hearing loss
  - Less than age 50
- Personal or **family history** of cardiovascular complications
  - Early onset less than age 50
  - Multiple individuals with cardiac incidents thought to be genetic in nature.
- Personal or **family history** of excessive bleeding or excessive clotting
  - Exception if there is a reported diagnosis of hemophilia in family members further than second degree relatives.
- Family history of sudden, unexplained death (especially at a young age)

Cancer (Degree of Family History dependent on specifics of disease)

- Personal or family history of one individual with two primary cancers
- Personal or family history of early onset cancer
  - Less than age 50*
    - *Depending on the specific cancer
    - Multiple individuals with unknown cancer at a young age
  - Cancer that is frequently seen in hereditary cancer syndromes
- Bilateral disease (cancer) in paired organs
- Personal and/or family history of two or more cancers suggestive of a hereditary cancer syndrome from the same side of the family
- Personal or family history of uncommon cancers that are associated with hereditary cancer syndromes.

APPENDIX B: Examples of Missing Indications Found

Missing Indication Notes:
CFTR variant history in family, Personal/Family history of BRCA2 variant
2nd degree relative (Patient's grandmother) with ovarian cancer. Patient's partner father diagnosed with kidney cancer at 55
Previous child with "Dandy Walker malformation"
Son with autism
Mother with history of possible ovarian cancer
Prior fetal demise with spina bifida
Alcohol exposure during pregnancy
Brother with autism and intellectual disability
Recent diagnosis of type 2 diabetes likely present before conception
Patient's maternal grandmother with pancreatic cancer in her 60s
Low Mean Corpuscular Volume (possible carrier of hemoglobinopathies)
Maternal aunt died of unknown cancer in 40s
Son with previous partner has autism, father's paternal half-sister has a son with autism
Patient's partner and partner's mother have Factor V Leiden
Smoked 4-5 cigarettes a day during pregnancy
Previous pregnancy with partial loss of chromosome, which was classified as a variant of uncertain significance by the performing laboratory
Half-brother’s son with NAD deficiency
Patient's brother has a history of autism and seizures
Patient's maternal aunt died of an unknown cancer in her 30s

Patient's partner's son with another partner is very short (3'6")
Patient's partner with congenital bicuspid aortic valve, partner's dad with heart issues in 40s (requiring angioplasty)
Patient was vaping up until 20 weeks
Partner's sister born with congenital bilateral deafness
Mother with ovarian cancer at 42
Patient is MCADD carrier from testing at OB office
Patient's paternal aunt diagnosed with breast and uterine cancer in 50s, died in 50s. Paternal grandmother diagnosed with breast cancer before 50, died in 50s
Patient's sister with "high functioning" autism
Previous children with autism. Both have a history of receiving ABA, speech, occupational, and physical therapy services
Previous child with congenital anomalies thought to be VACTERL
Family history of arrythmias and multiple cardiac arrests at young ages
Family history of Galactosemia
Possible hereditary Wolff-Parkinson-White Syndrome (WPW)