

**SPECIAL ISSUE**

# The COVID-19 pandemic and reproductive genetic counseling: Changes in access and service delivery at an academic medical center in the United States

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**Abstract**

The COVID-19 pandemic widely disrupted the delivery of healthcare services, including genetic counseling. To ensure continuity of care, the reproductive genetic counselors at a large academic medical center in the United States rapidly transitioned their practice from 90% in-person patient consultations to a predominantly telehealth model. The present study describes this transition in regard to patient access to genetic counseling and genetic screening. A chart review of patients seen by the reproductive genetic counselors from January 2020 to August 2020 was completed. The time frame included the three months prior to the COVID-19 pandemic and the first five months during COVID-19. Patient demographics and clinical and appointment data were compared between the pre-COVID-19 and during-COVID-19 timeframes. Overall, 88.6% of patients were seen via telehealth during COVID-19 and there was no significant difference based upon patient age ( $p = .20$ ), indication for appointment ( $p = .06$ ), or gestational age ( $p = .06$ ). However, non-English speaking patients were more often seen in-person than by telehealth ( $p < .001$ ), and more patients residing farther from the clinic were seen via telehealth ( $p = .004$ ). During COVID-19 results for prenatal cell-free DNA screening and expanded carrier screening were delayed ( $p < .001$ ). Additionally, after consenting to screening, patients seen during COVID-19 were more likely to not complete a sample collection for their intended screening when compared to those seen pre-COVID-19 (OR = 6.15, 95% CI = 1.43–26.70,  $p = .015$ ). Overall, this study supports that access to genetic counseling services and genetic screening can be maintained during a global pandemic like COVID-19. Genetic counselors are well-equipped to pivot swiftly during challenging times; however, they must continue to work to address other barriers to accessing genetic services, especially for non-English speaking populations. Future studies are needed to pose solutions to the obstacles confronted in this service delivery model during a global pandemic.

**KEYWORDS**

COVID-19, genetic counseling, genetic counselors, genetic services, prenatal genetic counseling, service delivery models, telemedicine

## 1 | INTRODUCTION

In the pre-COVID-19 era, telehealth was not widely adopted as a means of clinical patient care (Greenberg et al., 2020; NSGC Professional Status Survey: Executive Summary, 2020; Wootton, 2012). Data suggest that telehealth increases patient access to providers and has high patient satisfaction (Abrams & Geier, 2006; Greenberg et al., 2020), yet there are known barriers to implementation, such as technology appropriation, billing and reimbursement, and patient and provider resistance (Cohen et al., 2016; Khoong et al., 2021; Scott Kruse et al., 2018; Shivkumar et al., 2021; Wang et al., 2021). Genetic counselors were early adopters of telehealth, as their services are easily adapted for telecommunication (NSGC Professional Status Survey: Executive Summary, 2020; Stoll et al., 2018; Zierhut et al., 2018).

The reproductive genetic counseling team at one large academic medical center in the southeast United States consists of six (4.15 FTE for clinical care) genetic counselors and supports ten Maternal-Fetal Medicine specialists. Before the COVID-19 pandemic, this team provided in-person reproductive genetic counseling consultations for approximately 1,500 patients a year located in six women's health clinics. Referral indications ranged in acuity and included pre-test counseling for prenatal cell-free DNA screening and expanded carrier screening (ECS), infertility, recurrent pregnancy loss, family history concerns, and ultrasound anomalies, as well as post-test counseling for abnormal genetic screening results. The reproductive genetic counseling team began providing telehealth with live audio and visual communication (USA Department of Health & Human Services, 2020) to three rural areas in 2015 via a clinic to clinic (C2C) model in which patients are located in a satellite clinic for their visit and use the institution's technology. While the institution had technology embedded into the electronic medical record (EMR) for robust telehealth services, logistical challenges such as state and institutional billing practices limited expansion of telehealth services beyond C2C despite the positive satisfaction data from patients. Direct-to-Patient (D2P) visits, in which patients are located in their home (or other location) and use a personal device to complete the visit, were less common due to billing restrictions at the state level prior to the COVID-19 pandemic, despite the fact that D2P visits allow for greater flexibility.

A public health emergency due to COVID-19 was declared in Davidson County, Tennessee on March 15, 2020, and the 'Safer at Home' order was declared on March 23, 2020. Tennessee's 'Safer at Home' order encouraged employers to have nonessential employees work from home whenever possible. Given the genetic counseling team's previous experience with the telehealth model of care and the institution's readiness for D2P telehealth encounter types, telehealth for the majority of reproductive genetic counselor appointments began on March 23, 2020. The institution requires that telehealth be completed with both audio and visual media; therefore, phone consultations were not conducted. Scheduling for a telehealth appointment required the

### What is known about this topic

Telehealth for genetic counseling services increases patient access and maintains high patient satisfaction. The COVID-19 pandemic rapidly shifted the model of service delivery across many areas of medicine, including genetic counseling.

### What this paper adds to the topic

This retrospective chart review of a reproductive genetic counseling service during five months of the COVID-19 pandemic illustrates one institution's successful shift to telehealth genetic counselor services. There are limited data on how the sudden expansion of telehealth services along with a concurrent decrease in in-person appointments has affected patient access to genetic counseling services and genetic screening.

following: participation in the institution's online medical portal to allow secure access to the virtual appointment, language preference of English, Spanish, or Arabic (other language interpretation was not available through telehealth), and location in the state of Tennessee at the time of the visit due to licensing requirements. Patients who did not meet these requirements were offered in-person consultations. One day per week was reserved for in-person consultations for patients undergoing prenatal diagnostic procedures, patients unable to successfully utilize telehealth technology due to access or language barriers, or patients who requested an in-person appointment.

The COVID-19 pandemic and the shift to telehealth also dictated changes to sample collection for genetic screening. Prior to the COVID-19 pandemic, samples were collected by clinic staff and/or phlebotomy technicians immediately following the genetic counseling appointment. With the implementation of D2P telehealth visits due to COVID-19 patients had the option to go to an independent phlebotomy clinic or elect mobile phlebotomy services through the genetic testing laboratory. When possible, patients were mailed saliva kits for self-collection. The genetic counselors worked closely with all parties to swiftly put these options in place and revised protocols as needed.

This study comprises a retrospective chart review to describe the changes in access to reproductive genetic counseling services and reproductive genetic screening at Vanderbilt University Medical Center after the expansion of telehealth services due to the COVID-19 pandemic. The purpose of this study is to increase knowledge and understanding of telehealth for reproductive genetic counseling services by comparing the following metrics between the pre-COVID-19 and during-COVID-19 service delivery models: characteristics of the patient population, indications for referral, access to genetic counseling services, and timelines for sample collection and result reporting.

## 2 | METHODS

This descriptive study was approved by the Vanderbilt University Medical Center (VUMC) Institutional Review Board (IRB#200993). For the purposes of this study, 'pre-COVID-19' appointments are considered prior to March 23, 2020, and 'during-COVID-19' appointments are after March 23, 2020. Telehealth for the majority of reproductive genetic counselor appointments began on March 23, 2020, and partial return to in-person appointments began on September 1, 2020. Telehealth consults were conducted with synchronous audio and visual technology.

### 2.1 | Study population

Patients seen at the Vanderbilt Center for Women's Health by a reproductive genetic counselor between January 1, 2020, and August 31, 2020, were included. At the beginning of the 2020 calendar year, there were 13 Vanderbilt Center for Women's Health locations in middle Tennessee with plans for expansion throughout the year, and the genetic counselors provided services to patients at five of the locations with referrals from all clinics.

### 2.2 | Instrumentation

Study data were collected from the electronic medical record (EMR) and managed using REDCap electronic data capture tools hosted at Vanderbilt University. REDCap (Research Electronic Data Capture) is a secure, web-based application designed to support data capture for research studies. De-identified data from the chart review was downloaded from REDCap. Quantitative data were analyzed using R (R Core Team, 2020).

### 2.3 | Procedures

A retrospective chart review was conducted to gather descriptive data for patients who had an appointment with a reproductive genetic counselor in the Vanderbilt Center for Women's Health between January 1, 2020, and August 31, 2020. Demographic data (e.g. age, gender, zip code) was recorded along with appointment referral information. Patient language was recorded as the patient's documented preferred language from their medical chart. Clinical data about the genetic counseling appointment (e.g. referral indication) and the patient (e.g. gestational age at appointment, gravidity) was collected along with clinical testing information. Multiple data points regarding the timeline were recorded, such as time from referral to appointment and time from appointment to sample collection and results in disclosure.

Each patient chart was independently reviewed by two of six study authors (C.M., B.G., A.G., J.S., R.N., or M.D.) to ensure the accuracy of the data recorded. A third review was performed by a

different author if a discrepancy in the data was identified between the first and second reviews. As the data were objective, inter-rater reliability was not calculated.

### 2.4 | Data analysis

Patients' demographics, clinical data, appointment, and reproductive genetic screening characteristics were summarized with mean and standard deviation (*SD*, continuous variables) or frequency and percentage (categorical variables) by the timing of seeking genetic counselor (pre-COVID period versus during-COVID period). Differences between groups were assessed using Wilcoxon's Rank-Sum or Pearson's chi-squared tests. The primary study endpoint was the patient's disclosure of screening results, mediated by completion of a sample collection for their intended screening. To evaluate the associations between the period and the primary endpoint, logistic regression models were used. Estimated odds ratios (ORs), and associated 95% confidence intervals (CIs) were reported as effect measurements. Due to the limited number of events, a priori defined list of factors (maternal age, distance to zip code 37,204, indication for screening) were adjusted one at a time in separate models, and no further multivariable-adjusted modeling was attempted. All missing covariate values were imputed 10 times using the MICE (multiple imputation using chained equations) implemented by *aregImpute* function in *rms* R package. Statistical significance was considered for all two-sided *p* values <5%. All analyses were conducted using R version 4.0.2 (R Core Team, 2020).

## 3 | RESULTS

Medical records for 923 patients who completed a reproductive genetic counseling visit at Vanderbilt University Medical Center between January 1, 2020, and August 31, 2020, were abstracted. Three hundred and forty-five patients (37.4%) were seen in the pre-COVID-19 time frame and 578 patients were seen during COVID-19 (62.6%). Ninety-eight percent (911/923) of patients were female with a median age of 34 years. The majority of patients were white (590/923, 64.0%) and indicated in the EMR that their preferred language is English (821/923, 88.9%) (Table 1). Of the 911 female patients, 51 (5.6%) presented as nulligravida and for preconception consultation, while the other 860 patients presented as gravid with at least one pregnancy (Table 1). Appointment indications varied with advanced maternal age (387/923, 41.9%) and family history concern (260/923, 28.2%) being the most common.

For the purposes of quantitative data analysis only reported female patients were included, giving a final cohort of  $n = 911$ .

**TABLE 1** Demographics

Characteristic	Pre-COVID (N = 340) <sup>a</sup>	During COVID (N = 571) <sup>a</sup>	Combined (N = 911) <sup>a</sup>
Age	33.2 ± 5.9	32.7 ± 5.9	32.9 ± 5.9
≤35	54% (184)	60% (343)	58% (527)
>35	46% (156)	40% (228)	42% (384)
Race/Ethnicity			
White	61.5% (209)	64.4% (368)	63.3% (577)
Black/African American	12.9% (44)	10.5% (60)	11.4% (104)
Hispanic/Latinx	7.0% (24)	8.4% (48)	7.9% (72)
Middle Eastern	6.5% (22)	7.0% (40)	6.8% (62)
Asian	5.0% (17)	5.3% (30)	5.2% (47)
American Indian/Alaska Native	0.3% (1)	0.2% (1)	0.2% (2)
Native Hawaiian or Other Pacific Islander	0.3% (1)	0.0% (0)	0.1% (1)
Mixed Race	2.1% (7)	3.0% (17)	2.6% (24)
Other	1.5% (5)	0.7% (4)	1.0% (9)
Unknown	2.9% (10)	0.5% (3)	1.4% (13)
Language			
English	87.9% (299)	89.5% (511)	88.9% (810)
Spanish	4.1% (14)	3.9% (22)	4.0% (36)
Arabic	5.0% (17)	3.7% (21)	4.2% (38)
Kurdish (Badini)	0.3% (1)	0.3% (2)	0.3% (3)
Burmese	0.3% (1)	0.3% (2)	0.3% (3)
Other	2.4% (8)	2.3% (13)	2.3% (21)
Gravidity			
Nulliparous (G0)	5.9% (20)	5.4% (31)	5.6% (51)
Primigravid (G1)	25.3% (86)	29.8% (170)	28.1% (256)
Multigravid (G2+)	68.8% (234)	64.8% (370)	66.3% (604)
Gestational Age (weeks)	14.0 ± 4.7	14.4 ± 4.8	14.2 ± 4.7

<sup>a</sup>For the purposes of quantitative data analysis, only reported female patients were included, giving a final cohort of  $n = 911$ .

### 3.1 | Access to genetic counseling and service delivery

Overall, 58% of female patients (530/911) were seen via telehealth with the remaining (381/911, 41.8%) being seen for in-person consult. The majority of telehealth consults (506/530, 95.5%) were conducted during the COVID-19 period. Method of service delivery did not significantly differ based upon patient age ( $p = .20$ ), indication for appointment ( $p = .06$ ), or gestational age ( $p = .06$ ). However, non-English speaking patients were more often seen in-person than by telehealth ( $p < .001$ ), and more patients residing farther from Vanderbilt Center for Women's Health (zip code 37204) were seen via telehealth ( $p = .004$ ). The majority of patients (842/923, 91.22%) resided within three clusters of zip codes in middle Tennessee reflecting proximity to the Center for Women's Health.

The COVID-19 pandemic did not delay appointment scheduling for genetic counselors ( $p = .90$ ) (Table 2). The amount of patient consults seen per week did not significantly differ with an average of 28

visits per week pre-COVID-19 and an average of 24 visits per week during COVID-19. There was a brief decrease in the number of consults during the transition to telehealth as seen in Figure 1, and there was a delay in scheduling patients for telehealth appointments when compared to in-person appointments ( $p = .014$ ). The number of visits appeared to trend down leading up to March 23, 2020, and trended back upwards after the conversion to telehealth (Figure 1).

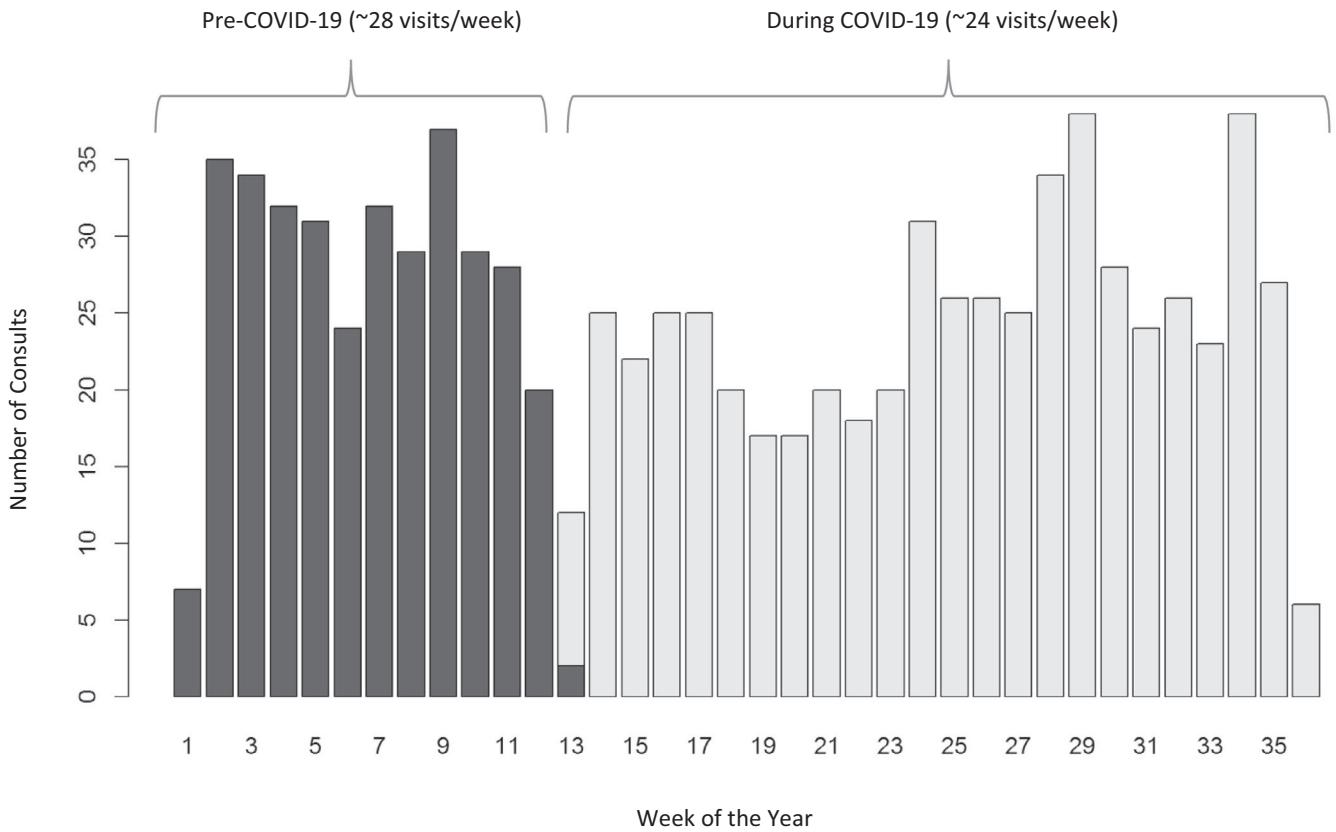
### 3.2 | Changes in reproductive genetic screening

During the COVID-19 pandemic results for prenatal cell-free DNA screening and expanded carrier screening were delayed ( $p < .001$ ). Significantly more patients had a delay from the time of genetic counselor consult to sample collection ( $p < .001$ ). Additionally, there was a noted delay in the time from sample collection to results disclosure for both prenatal cell-free DNA screening ( $p < .001$ ) and expanded carrier screening ( $p = .015$ ) (Table 3). There were no

**TABLE 2** Access to genetic counseling

	N	Pre-COVID (N = 340)	During COVID-19 (N = 571)	Combined (N = 911)	P-Value
Zip Code Distance (from 37,204, miles)	911	21.6 ± 32.8	21.9 ± 26.7	21.8 ± 29.1	0.63
Interpreter used when indicated					0.32
No	101	14.6% (6/41)	8.3% (5/60)	10.9% (11/101)	
Yes		85.4% (35/41)	91.7% (55/60)	89.1% (90/101)	
Indication for referral <sup>a</sup>					0.63
AMA	911	33.2% (113)	28.6% (163)	30.3% (276)	
Family History		26.8% (91)	27.5% (157)	27.2% (248)	
Discuss Genetic Screening		16.2% (55)	18.6% (106)	17.7% (161)	
Fetal Abnormality		15.6% (53)	16.1% (92)	15.9% (145)	
Preconception		8.2% (28)	9.3% (53)	8.9% (81)	
Service delivery model					
In-Person	911	92.9% (316)	11.4% (65)	41.8% (381)	--
Telemedicine		7.1% (24)	88.6% (506)	58.2% (530)	
Days from Referral to Consult	911	20 ± 14	21 ± 17	21 ± 16	0.9

<sup>a</sup>Indications for referral were grouped into five categories: advanced maternal age, family history, discuss genetic screening (low-risk patients, wanted more information, etc.), fetal abnormality (serum screening, fetal anomaly, fetus with genetic condition, etc.), and preconception (preconception, recurrent pregnancy loss, and infertility).



**FIGURE 1** Genetic counseling consults per week of the 2020 year

significant differences in re-draw rate or sample failure rate (prenatal cell-free DNA screening,  $p = .52$ ; expanded carrier screening,  $p = .12$ ); however, observationally, the sample failure rate for

expanded carrier screening increased from 0.0% pre-COVID-19 to 7.3% during COVID-19.

**TABLE 3** Changes in genetic screening

	N	Pre-COVID (N = 340)	During COVID (N = 571)	Combined (N = 911)	P-Value
Days from Consult to cDNA <sup>a</sup> Draw	588	0.15 ± 1.31	4.98 ± 8.95	3.12 ± 7.44	<0.001
Days from cfDNA Draw to Results	587	6.8 ± 2.6	8.8 ± 2.9	8.0 ± 3.0	<0.001
Days from Consult to cfDNA Results	587	6.9 ± 3.0	13.8 ± 9.3	11.1 ± 8.2	<0.001
Days from Consult to ECS <sup>b</sup> Draw	164	0.57 ± 3.39	4.82 ± 5.87	3.32 ± 5.51	<0.001
Days from ECS Draw to Results	163	12.5 ± 6.5	13.7 ± 5.6	13.3 ± 6.0	0.015
Days from Consult to ECS Results	163	13.1 ± 7.2	18.6 ± 7.6	16.6 ± 7.9	<0.001
ECS re-draw required					
No	113	100% (31/31)	92.7% (76/82)	94.7% (107/113)	0.12
Yes		0% (0/31)	7.3% (6/82)	5.3% (6/113)	
cfDNA re-draw required					
No	588	94.7% (215/227)	95.8% (346/361)	95.4% (561/588)	0.52
Yes		5.3% (12/227)	4.2% (15/361)	4.6% (27/588)	
cfDNA ever drawn					
No	600	0.4% (1/228)	3.0% (11/372)	2.0% (12/600)	0.032
Yes		99.6% (227/228)	97.0% (361/372)	98.0% (588/600)	
ECS Ever collected					
No	181	0.3% (2/60)	12.4% (15/121)	9.4% (17/181)	0.049
Yes		96.7% (58/60)	87.6% (106/121)	90.6% (164/181)	
Sample obtained					
Ever	688	99.2%(261/263)	95.1% (404/425)	96.7%(665/688)	0.003
Never		0.8% (2/263)	4.9% (21/425)	3.3% (23/688)	

<sup>a</sup>cfDNA=prenatal cell-free DNA screening.

<sup>b</sup>ECS=expanded carrier screening.

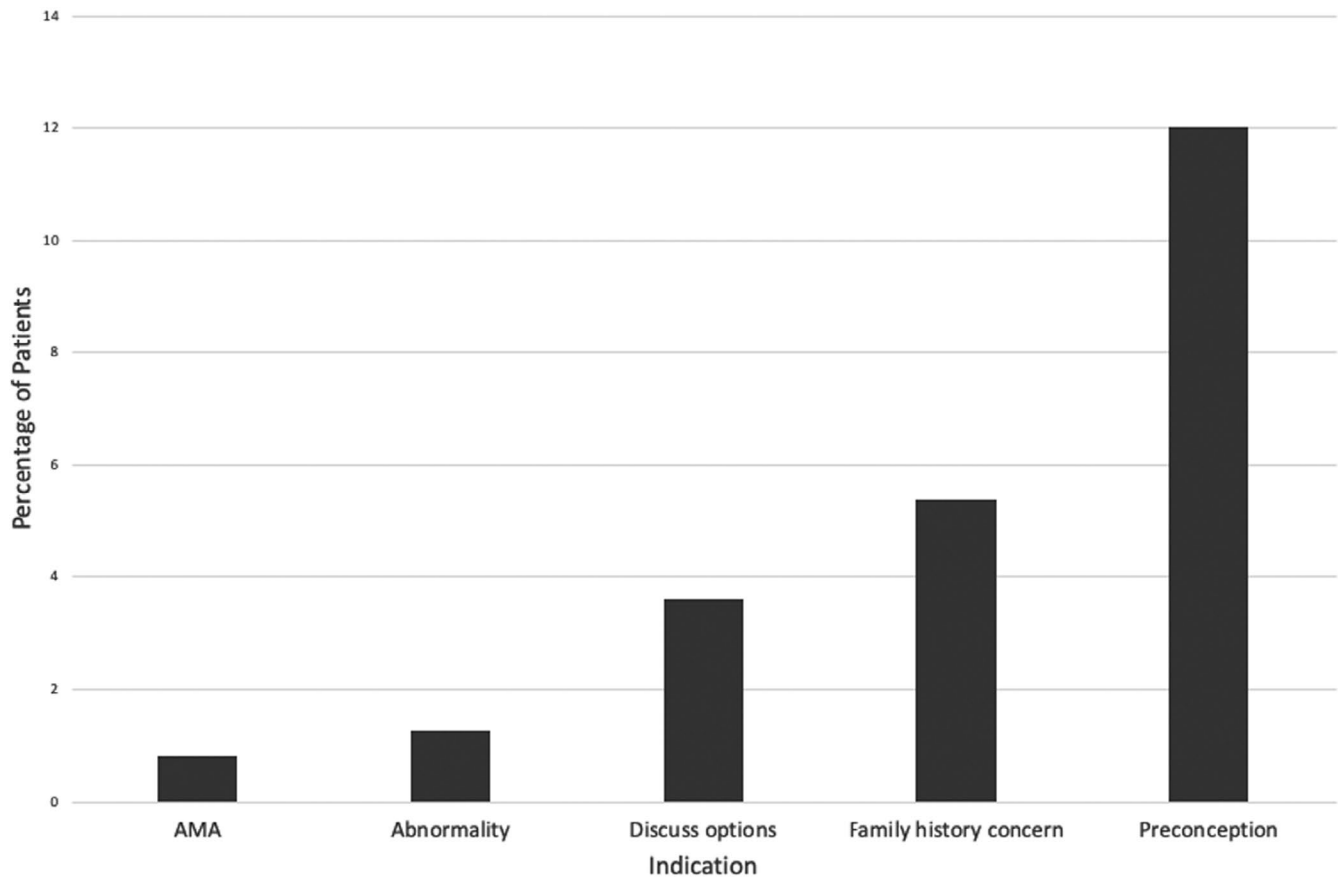
After consenting to genetic screening, patients seen during COVID-19 were more likely to not complete a sample collection for their intended screening when compared to those seen pre-COVID-19 (OR=6.18, 95% CI=1.43–26.70,  $p = .015$ ). Prior to COVID-19 0.8% (2/263) of patients did not have a sample collected for screening, and during-COVID-19 4.9% (21/425) of patients did not complete sample collection (Table 3). Compared with patients of advanced maternal age (AMA), patients <35 years of age were more likely to not have their genetic screening samples collected during COVID-19 (OR=3.91, 95% CI=1.31–11.67,  $p = .015$ ). Significantly more patients seen by a genetic counselor for an indication of a family history concern did not complete their genetic screening sample collection ( $p < .001$ ). Additionally, compared to patients are seen for an indication of AMA, patients seen for an indication of preconception were the least likely to complete their sample collection after consenting to genetic screening (OR=16.10, 95% CI=3.10–83.17,  $p < .001$ ) (Figure 2). There was no significant difference in sample collection for patients presenting for other indications, such as abnormal serum screening, fetal soft marker for aneuploidy, recurrent pregnancy loss, fetal abnormality, etc.

## 4 | DISCUSSION

Overall, the implementation of telehealth for reproductive genetic counseling services during the COVID-19 pandemic was successful. There was no lapse in the provision of reproductive genetics services and appointment scheduling was not delayed. There was a slight decline in the number of consults seen during the transition period, however, the quantity quickly returned to pre-COVID-19 metrics. With the implementation of telehealth came a change in the procedures for genetic screening. These changes, in combination with effects to the genetic testing laboratory services, caused a delay in results disclosure or lack of screening completion.

### 4.1 | How did the COVID-19 pandemic affect access to genetic counselor services?

Previous literature has shown that telehealth services may improve patient access to genetic counseling (Greenberg et al., 2020). Due to the shift to telehealth in response to the global pandemic, more patients were seen via telehealth following the onset of COVID-19. Only 7% of patients seen pre-COVID-19 were seen via



**FIGURE 2** Patients who did not complete screening during COVID-19 by indication

telehealth using the C2C model, while 88.6% of patients seen during COVID-19 were seen via telehealth using both D2P and C2C models (Table 2). Factors such as the patient's age, reason for the referral, or gestational age did not impact whether they were seen via telehealth or in-person. Additionally, there was not a statistically significant difference in the indications for referral pre- and during-COVID-19 (Table 2). This suggests that telehealth was a successful method of service delivery across many different types of patients. Two important factors did influence whether patients were scheduled in-person or via telehealth: patient language and proximity to clinic.

During COVID-19, non-English speaking patients were more likely to be seen in person than via telehealth. As the pandemic continued and telehealth usage increased, solutions to the lack of interpreter access via telehealth were continually implemented by the institution and contracted language interpretation agency. However, available languages were typically limited to Spanish and Arabic, and technological barriers often precluded interpreter use in telehealth settings. Another study (Rodriguez et al., 2021) found that patients with lower English proficiency had lower levels of telehealth use. While language did appear to be a barrier to utilizing telehealth services, 4.5% of telehealth appointments during COVID-19 were completed with non-English speaking patients, compared to 0% in the studied pre-COVID-19 timeframe. Hopefully, this trend continues over time and more solutions, such as the ability to easily add a video

interpreter to a telehealth appointment, will continue to be brought forth so that language is not a large barrier to telehealth services.

Proximity to Vanderbilt University Medical Center was another factor that impacted the scheduling of telehealth appointments. Patients who were located farther away were more often scheduled for telehealth appointments versus in-person appointments. These results suggest that telehealth increases access for patients who are located at a greater distance from the clinic, consistent with research by Cohen et al., 2016. Interestingly, the majority of patients in this study resided within a cluster of three zip codes in Middle Tennessee. It is possible that barriers to accessing reproductive genetic counseling services still exist for patients in rural areas. Aside from geographics, other barriers could include technology access and literacy (Roberts & Mehrotra, 2020; Scott Kruse et al., 2018), insurance coverage and reimbursement (Zierhut et al., 2018), cost (Molfenter et al., 2015), and patient and provider biases for appointment type (Scott Kruse et al., 2018; Zierhut et al., 2018). In all, while telehealth expanded access to reproductive genetic counseling for some populations, ways to expand access to rural populations necessitates further investigation.

Overall access to genetic counseling, assessed through patient volume, did not differ pre-COVID-19 and during-COVID-19, as there was not a significant difference in the number of consults per week (Figure 1). Therefore, the pandemic itself did not appear to be a barrier to patients seeking or obtaining genetic counseling services in



this clinic. While patient volume did not increase during COVID-19 in response to availability of telehealth services, as suggested in another telehealth study (Ashwood et al., 2017), this was likely limited by the number of available appointments. It is possible that if appointment slots were unlimited, more patients could have been seen after telehealth was expanded. Regarding scheduling time for both appointment types, there was not a significant difference in the time from referral to consultation pre- and during-COVID-19 (Table 2), but this timing was longer for telehealth appointments than in-person visits during both of those time periods. It is important to note that during COVID-19, in-person visits were reserved for those who were unable to complete a telehealth visit, requested an in-person consult, or if they were scheduled for a prenatal diagnostic procedure. Therefore, the in-person appointment templates were not always at capacity, which may explain the shortened wait time for an appointment. Similarly, patients requiring a diagnostic procedure (chorionic villus sampling or amniocentesis) were likely expedited due to the urgency of the indication. While there were many concerns that COVID-19 would impede accessibility, the present data support that the expansion of telehealth services allowed patient volume to remain the same for all indications during an unprecedented global pandemic.

#### 4.2 | How did the COVID-19 pandemic affect reproductive genetic screening?

The COVID-19 pandemic has impacted patient access to genetic screening results. Due to the increased use of telehealth, patients experienced longer time intervals between their genetic counselor consultation and results disclosure. The authors highlight this aspect because of the time-sensitive nature of pregnancy and options regarding pregnancy termination available to patients during specific gestational ages. This delay was observed both in the days to sample collection after the consult and the days to results reporting after collection (Table 3). Shifting the majority of telehealth genetic counseling consults during COVID-19 to direct to patient (D2P) visits meant establishing new protocols for collecting samples. Prior to COVID-19 and during in-person consults, patient samples were collected on-site directly following the appointment. Patients seen via D2P telehealth required a separate lab appointment or time for sample collection at home, thus increasing the time between screening consent and date of collection. Sample collection difficulties have been previously reported (Bergstrom et al., 2020) and further research is needed to address this barrier. Anecdotally, some patients were seen for telehealth consultation during an active COVID-19 infection and were not able to have blood drawn until after their allotted quarantine time. Additionally, longer turn-around-times from the laboratories were observed, likely a result of their shipping and workflow adjustments brought on by the COVID-19 pandemic (Hale, 2020; Kunjok & Zingbondo, 2020). Another potential contributing factor for results delay is the need for sample re-collection. Rates of re-draws

for prenatal cell-free DNA screening remained unchanged during COVID-19 (Table 3). However, we observed an increase in expanded carrier screening sample failures due to the shift to patient-collected saliva samples rather than blood draw. This is consistent with known data regarding increased genetic screening sample failure rates for saliva in comparison to blood samples (Yao et al., 2020). These adaptations, as well as delayed results reporting, significantly increased the time it took for patients to receive results after the COVID-19 telehealth transition.

During COVID-19, a small but significant number of patients (4.9%) failed to complete the sample collection process after consenting to screening during a telehealth consult. This lack of sample collection rate is similar to other studies involving at-home specimen collection following consent via telehealth (Sullivan et al., 2021). This rate was significantly increased in comparison with the pre-COVID-19 lack of sample collection rate (0.8%) (Table 3). The burden of an extra laboratory appointment or coordination of at-home collection may explain said increase. Importantly, there were significant differences in the demographics of the patients who did not complete their sample collection for genetic screening, with patients of advanced maternal age being the most likely to have samples collected. Patients with a referral indication of preconception or family history concern were less likely to have samples collected compared to patients with an indication of advanced maternal age (Figure 2). This finding is in agreement with previous studies showing that carrier screening uptake was lower than initial interest, especially for preconception patients as compared to pregnant patients (Van Steijvoort et al., 2020), suggesting a lack of urgency for this population. Additionally, for those with a family history concern, the value of screening results may have been lower if elected screening did not directly address said family history concerns (e.g. patient with family history of autoimmune disease concern electing prenatal cell-free DNA screening). This discrepancy may have led to a higher number of patients foregoing sample collection, though more qualitative research is needed on this topic. Increased time to results disclosure as well as a decrease in sample collection were observed during COVID-19, with possible resulting impact on patient care.

#### 5 | STUDY LIMITATIONS

This study was intentionally designed to describe the changing landscape of access to reproductive genetic counselors and genetic screening throughout the transition of service delivery in medical care during the COVID-19 pandemic. The nature of the study design has inherent limitations. The data represent patients seen by providers at one academic medical center in the United States during an eight-month period in which there were numerous challenges for patients, providers, medical systems, and the community at large. Challenges were transitory and some improved over time. For example, solutions were found for better-integrating interpreters over telehealth.



Data are from only female patients and genetic counseling sessions were for reproductive planning and conclusions should be taken in that context. The period of data collection during COVID-19 was for five of the eight total months, requiring adjustment when making comparisons. When measuring the distance of the patient from the clinic, zip code data were used as a proxy which may vary from the actual distance. The number of patients who chose not to follow through on genetic screening for which they had consented was small ( $n = 23$ ), thus creating a wide confidence interval during analysis. The reliability of these results may be diminished due to this. Additionally, the contributing factors for not completing testing are speculative and require further investigation. The data regarding indication for referral were collected consistently across the study. Multiple indications for each visit could be indicated. For the purposes of analysis the first indication was used and this may not represent the primary indication. Due to a lack of interpreter availability for telehealth appointments, many non-English speaking patients were offered in-person care which potentially made telemedicine less accessible to this population and patient preference for in-person visits should not be assumed. The major limitation is the unprecedented and uncertain time and thus other factors such as infection rates, patient's employment status, and insurance coverage may also have played a role in patient access to medical care.

## 6 | PRACTICE IMPLICATIONS

Patients desired the access to genetic counseling and genetic screening despite a pandemic. The pandemic ignited resilience, ingenuity, and perseverance to maintain access to these services across the referral base.

COVID-19 expedited the evolution of this center's genetic counseling practice by expanding telehealth. This expansion remains in place and will continue to increase access for patients. Barriers to telehealth and genetic counselor access, specifically for those who live farther away and are non-English speaking, should continue to be addressed. Obtaining samples from patients after a direct-to-patient telehealth visit will continue to need creative solutions in order to decrease time to result disclosure.

Technology support for patients and providers is crucial to ensure telehealth services are embraced and successful. While increasing access to telehealth is important, barriers remain to be solved to ensure greater access to genetic counseling and genetic screening.

## 7 | RESEARCH RECOMMENDATIONS

The clinical landscape was evolving as this data were collected and therefore is more reflective of a snapshot. Additional studies will be needed to assess the full impact of the pandemic on genetic counseling services. It is likely that telehealth will remain an essential

component of medical care post-pandemic (Topol, 2020) and more research is needed regarding the efficacy, efficiency, and patient and provider perception of this service delivery model in the genetics clinic. Future studies should continue to monitor patient compliance with sample collection in the telehealth setting and explore contributing factors to pose solutions for this obstacle.

## 8 | CONCLUSIONS

This descriptive study sought to examine differences in patient and appointment characteristics pre- and during COVID-19 for this reproductive genetic counseling service at a large academic medical center.

This transitional time highlighted barriers that had solutions, identified new barriers, and further identified access issues. By expanding the telehealth service patients continued to receive reproductive genetic counselor services without significant changes to patient and appointment characteristics, with the exception of patient proximity to clinic and language. Telehealth made these services more accessible for patients who lived further away while slightly increasing the time to obtain an appointment. The majority of non-English speaking patients were offered in-person visits and these visits, while fewer, had a shorter wait time.

Genetic screening is reliant on patients having access to phlebotomy, as well as a robust and efficient workflow for laboratories to receive, process, and report results. The many challenges imposed by the COVID-19 pandemic hindered all aspects of this process resulting in delayed results reporting.

Overall, these data support that access to genetic counseling services and genetic screening can be maintained during a global pandemic like COVID-19. An increase in telehealth services is not a panacea for ensuring all patients have access to these services. Genetic counselors are well equipped to pivot swiftly during challenging times; however, they must continue to work to address other barriers to accessing genetic services, especially for underserved populations such as rural communities and non-English speaking patients.

## AUTHOR CONTRIBUTIONS

Caitlin Mann was involved in conception and design, acquisition, analysis, and interpretation of data, drafting and revising the work, and final approval. *This author confirms full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.* Martha, Dudek, Brighton Goodhue, Arianna Guillard, Jill Slamon, and Randa Newman were involved in conception and design, acquisition and interpretation of data, drafting and revising the work, and final approval. Gianna Petrelli was involved in acquisition of data, revising the work, and final approval. Zhiguo Zhao and Tan Ding were involved in data analysis, drafting the work, and final approval. *All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in*

ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

## COMPLIANCE WITH ETHICAL STANDARDS

### CONFLICT OF INTEREST

C. Mann, M. Dudek, B. Goodhue, A. Guillard, J. Slamon, R. Newman, G. Petrelli, Z. Zhao, and T. Ding declare that they have no conflict of interest.

### HUMAN STUDIES AND INFORMED CONSENT

The study was approved by the Vanderbilt University Medical Center Institutional Review Board. No informed consent was required from subjects as data were anonymously extracted from the electronic health record, EPIC. All procedures followed were in accordance with US Federal Policy for the Protection of Human Subjects.

### ANIMAL STUDIES

No non-human animal studies were carried out by the authors for this article.

### DATA SHARING AND DATA ACCESSIBILITY

The data that support the findings of this study are not publicly available due to privacy or ethical restrictions.

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**How to cite this article:** Mann, C., Goodhue, B., Guillard, A., Slamon, J., Newman, R., Zhao, Z., Ding, T., Petrelli, G., & Dudek, M. (2021). The COVID-19 pandemic and reproductive genetic counseling: Changes in access and service delivery at an academic medical center in the United States. *Journal of Genetic Counseling*, 30, 958–968. <https://doi.org/10.1002/jgc4.1462>