COVID-19 IMPACT ON GENETIC COUNSELING FOR HUNTINGTON'S DISEASE VIA TELEHEALTH

by
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Abstract

Huntington’s disease (HD) is a severe, heritable, adult-onset neurological disorder that impairs mood, movement, cognition, and behavior, with a life expectancy of 15-20 years following onset of symptoms. HD is caused by a genetic variant of the *HTT* gene. Because the cause of HD has been isolated to a known genetic variant, predictive genetic testing is available for individuals at risk of HD. Historically, most counseling for Huntington’s disease — particularly predictive genetic testing — has been provided in-person, to assess an at-risk individual’s psychological readiness to undergo genetic testing and to monitor potential adverse reactions to test results. During the COVID-19 pandemic, many genetic counselors (GCs) were obligated to provide telegenetic counseling by phone or video conferencing despite the normative convention of in-person practice. Little is known about GCs’ experiences of providing HD telegenetic counseling (HD-TGC). To address this gap, we conducted a retrospective mixed-methods study consisting of surveys and interviews with HD GCs. Our objectives were twofold: [1] To assess the nature and extent of HD-TGC services offered during COVID-19 restrictions; and [2] To assess GCs’ attitudes toward the provision of these services as well as their perceptions of factors that favored or hindered its effectiveness. The online survey investigated both objectives and was completed by 49 GCs — 36 of whom had provided HD-TGC services during COVID restrictions, while the remaining 13 counselors provided HD genetic counseling only in-person. A sample of 16 GCs who provided HD-TGC services were interviewed to discuss their attitudes toward HD-TGC in deeper detail. Most responses from these GCs were favorable, regarding assessments of: [1] their comfort in providing HD-TGC, [2] the perceived effectiveness and [3] quality of HD-TGC services, and [4] the potential for HD-TGC to be offered as an option to the HD community beyond the COVID-19 pandemic.

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Chapter 1: Introduction

Many clinics halted in-person clinical services in response to the COVID-19 pandemic. Although the effects on clinical care varied between states, total outpatient visits in the U.S. plummeted in every state (Patel et al., 2021). On March 18, 2020, five days after a national emergency was declared in the United States, the Centers for Medicare & Medicaid Services (CMS) made a public recommendation that all elective and nonessential care either be delayed or accomplished through telehealth (TH), including clinical services delivered by phone or video conferencing (*CMS Releases Recommendations on Adult Elective Surgeries, Non-Essential Medical, Surgical, and Dental Procedures during COVID-19 Response*, 2020). To continue providing care, many clinics embraced TH as the preferred mode "*offset[ing] roughly two-thirds of the decline in in-person visit volume during the COVID-19 pandemic*" (Patel et al., 2021). Genetic counseling is one of many clinical services that adopted a mode of TH as an alternative to in-person clinical services (hereafter referred to as telegenetic counseling (TGC), versus "IPGC" for in-person genetic counseling) (Breen et al., 2021; Brown et al., 2021; Danylchuk et al., 2021; Dratch et al., 2021; Houston et al. 2021; Zoran et al., 2021).

Telehealth & Telegenetic Counseling

In a 2019 survey, two-thirds of patients indicated interest in trying TH; however, less than 10% indicated using it in the past (Amwell, 2019). Common reasons participants gave for not trying TH pre-pandemic were that they were unfamiliar with the process and unsure of their ability to access it (Amwell, 2019). Use of TH nearly tripled in 2020 for patients, increasing from 8% to 22%, while provider use of TH nearly quadrupled, increasing from 22% to 80% (Amwell, 2019; Amwell, 2020). Genetic counselors (GCs) who had not provided genetic counseling via telehealth (TGC) reported interest in trying it even before the COVID-19 pandemic (Zierhut et al., 2018). The proportion of GCs who work remotely in patient care rose from 20% in 2019 to

85% in 2020 (NSGC Professional Status Survey, 2021). In research conducted since onset of the COVID-19 pandemic, most GCs reported TGC as "*a valuable addition to patient care*," and that they "*hoped to offer it as an option beyond the pandemic*" (Mueller et al., 2021).

Notably, pre-pandemic studies of TGC found patient outcomes following TGC sessions to be equivalent or non-inferior to in-person service delivery (Athens et al., 2017; Bradbury et al. 2018; Buchanan et al. 2015; Danylchuk et al., 2021; Hawkins et al., 2013; Hawkins et al., 2011; Hilgart et al. 2012; Interrante et al. 2017; Mair, 2000; Solomons et al. 2018). TH has been described as feasible, acceptable, and effective in many settings, including those with populations with movement disorders, as well as with populations with indications of dementia and other neuromuscular symptoms — all of which are relevant to the HD community (Dratch et al., 2021; Houston et al., 2021). TGC has been found to be non-inferior or comparable to in-person services for a variety of outcomes including genetic knowledge, building trust and rapport, confidence in privacy, health behavior changes, psychosocial outcomes, and diagnostic accuracy (Brown et al., 2021; Danylchuk et al., 2021; Hatcher-Martin et al., 2020; Houston et al., 2021). Further, clinical assessments for the HD community that were historically conducted in-person such as the Montreal Cognitive Assessment (MoCA) and the Unified Huntington's Disease Rating Scale for motor assessment (excluding balance and rigidity tests), have proven validity and feasibility in pilot studies when administered via TH (Abdolahi et al., 2016; Bull et al., 2014). With proven success of TGC as well as success with additional HD-specific care via TH, providing some form of remote multidisciplinary care for the HD community appears not only possible but desirable.

Patients "*overwhelmingly endorse the use*" and feasibility of TH (Houston et al., 2021). In fact, "*the most consistent finding from [TH] studies is high patient satisfaction*" (Dorsey, 2021; Mair, 2000). Reported benefits of TH and TGC for patients include increased access to services and lower burdens of travel time and costs (Buchanan et al., 2015; Danylchuk et al., 2021; Houston et al., 2021; Hawkins et al., 2013; Menachemi et al., 2004; Solomons et al., 2018;

Terry et al., 2019). The burden of in-person clinical care can be especially high for those who live far from a clinic that provides specialized care; a full day could be spent traveling to and from an in-person clinic appointment which itself may take less than an hour. Some examples of financial burden for patients include the cost of gas or airfare to travel long distances, having to take time off from work for in-person clinic appointments, or paying for parking (Hawkins et al., 2013; Reed et al., 2020). Patient-centered care, a core tenet of genetic counseling, suggests the GC be flexible to meet the patient's needs (Dorsey et al., 2020; Houston et al., 2021). Lowering burdens for patients, increasing patient comfort, and considering convenience for the patient are all aligned with a patient-centered focus (Dorsey et al., 2020).

Recent systematic reviews of TGC across specialties suggest that patient satisfaction with TGC is comparable to — if not higher than — in-person genetic counseling (Brown et al., 2021; Danylchuk et al., 2021; Dratch et al., 2021). Most adult patients at one large academic medical center who were diagnosed with a neurological condition were satisfied with TGC services and only one patient indicated a preference for in-person services (Dratch et al., 2021). Additionally, there was no significant difference in patient satisfaction with TGC when surveyed after an initial consultation versus after a test result disclosure session (Dratch et al., 2021). Patient satisfaction does not appear to be limited to any particular element of a sequential protocol requiring more than one clinical session. This is especially relevant to genetic counseling for HD, which is recommended to include multiple sessions in the predictive genetic testing process.

Despite these findings, additional research has reported hesitation within the HD community to embrace TH services, with few HD patients utilizing an online healthcare platform and even fewer willing to commit to regular TH visits (Pfalzer et al., 2021). The authors describe uncertainty with further clarity:

> *"Potential explanations for these limitations may include socioeconomic barriers and caregiving structure … It is unclear based on our data whether inability to participate is due to unwillingness (would prefer in-person visits),*

> *inability (lack of internet access), or economic barriers (cost concerns); however, it is clear that a system based entirely on telehealth may neglect a quarter of this HD patient population" ... However, "50% of patients would participate in a monthly online support group facilitated by our social work team," which indicates interest in remote services" (Pfalzer et al., 2021).*

Barriers to TH utilization indeed exist. The primary barrier to TGC utilization commonly listed by GCs is issues with billing and reimbursement for TGC services (Mills et al., 2021; Zierhut et al., 2018). Additional barriers include privacy concerns and technical or technological issues, such as discomfort or lack of familiarity with technology, and poor internet connectivity, quality, and access (Houston et al., 2021; Mills et al., 2021). Notably, 85% of Americans own a smartphone, which increases accessibility of TGC for those patients (Pew Research Center, 2021). However, a smartphone alone does not guarantee quality internet service or technological literacy. GCs have also previously reported drawbacks of TGC related to additional administrative work, such as coordination of electronic consent and modifications to sample collection practices (e.g., direct-to-patient sample collection kits in place of clinical blood draws) (Mueller et al., 2021).

Research of TGC from GCs' perspectives is limited, and even more so when considering TGC for HD specifically (Brown et al., 2021; Danylchuk et al., 2021). Additionally, one review of the use of TH in neurology concludes that further research is needed to investigate the *outcomes, reach, adoption, and long-term use* of TH (Houston et al., 2021). The authors also note the need to investigate *how patient and physician perceptions and preferences for [TH] has changed since the pandemic* (Houston et al., 2021). Given these gaps in knowledge and the specific challenges of genetic counseling for HD, this project examined the transition to telegenetic counseling for HD during the COVID-19 pandemic.

Genetic Counseling for Huntington's Disease

Huntington's disease is a severe, heritable neurodegenerative disease. HD affects mood, cognition, memory, behavior, and movement, and has been described as having symptoms of amyotrophic lateral sclerosis (ALS), Parkinson's disease (PD), and Alzheimer's disease — all at once (Bates et al., 2015; Caron et al., 1998; Huntington's Disease Society of America [HDSA], n.d.). The symptoms of HD vary in severity and age of onset, but most often symptoms begin in middle adulthood. There is currently no treatment available to stop or slow the course of the disease (Bates et al., 2015; Caron et al., 1998).

The cause of HD has been isolated to a specific genetic variation of the *HTT* gene. HD is inherited in a dominant fashion, meaning that if an individual has inherited the causal genetic variant for HD, each of their offspring are at a 50% risk of inheriting that variant, and thus a 50% risk of developing HD. Genetic testing is available for at-risk individuals to determine whether or not they have inherited the causal genetic variant (Bates et al., 2015; Caron et al., 1998; HDSA, n.d.). Due to the severe implications of an HD risk or diagnosis, an HD-specific genetic counseling protocol was developed for at-risk individuals seeking genetic testing for HD (Bates et al., 2015; HDSA, 2016). The HD protocol has served as a model for genetic testing protocols for other severe, late-onset, and neurological diseases (Crook et al., 2021; Goldman, 2014; HDSA, 2016).

Ideally, pre-test counseling for HD predictive genetic testing explores the at-risk individual's background knowledge and experience with HD, their personal and family history, their motivations for testing, and how testing may affect the individual — personally and interpersonally, directly and indirectly — whatever the results may be. Beyond the serious physical and psychological implications for those who test positive, a positive result could also have profound effects on an individual's plans for their future such as reproductive choices, career directions, insurance coverage, and finances, to name a few. A negative result could result in “survivor guilt,” in which the individual tested may withdraw socially, or feel a heightened sense of motivation or responsibility to care for others who are at risk or have a diagnosis of HD. Those who experience distress after receiving a negative result may have made choices throughout their life as if they were HD-positive and may feel regret for not testing sooner.

Historically, genetic counseling sessions preceding and following predictive testing for HD have been recommended to take place in-person. Predictive testing protocols recommend extensive focus on pre-test counseling — sometimes as high as four sessions — before a test is ordered (Crook et al., 2021; Goldman, 2014; HDSA, 2016). One study of GCs providing service to the HD community since onset of COVID-19 restrictions revealed a mean of 2.5 pre-test visits before ordering a predictive test, with 95% of providers indicating that they implement the HD predictive testing genetic counseling protocol put forth by the Huntington’s Disease Society of America (HDSA) (Dunnington et al., 2020).

Pre-test HD genetic counseling sessions are recommended to take place in-person, largely to ensure that information is communicated clearly and to assess the at-risk individual’s motivations and readiness to learn whether they carry the causal genetic variant — ultimately *“to protect against adverse psychological effects post-testing*” (Crook et al., 2021). For post-test results disclosure sessions, the intention of in-person genetic counseling (IPGC) is to monitor and mitigate potential psychological distress the individual may experience after learning their results and to assess if the individual may require additional psychological support (Bates et al., 2015; Caron et al., 1998; Crook et al., 2021; Crozier et al., 2015; Fiedorowicz et al., 2011; Goldman, 2014; HDSA, 2016; Quaid & Morris, 1993; Wahlin et al., 2000; Wetzel et al., 2011). Examples of adverse events include a diagnosis of clinical depression, a psychiatric hospitalization, a drastic increase in alcohol consumption, divorce or break-up of a long-term relationship, or a planned, attempted, or completed suicide (Almqvist et al., 2003). Adverse reactions following predictive testing are most likely to occur within the first year post-test for those who test positive and although short-term distress surrounding genetic testing indeed exists for some, evidence suggests that "*most test participants adapt to this new information and integrate it positively into their lives*" (Almqvist et al., 2003).

In a systematic review of late-onset neurological disorders, there were few major adverse events reported (Crook et al., 2021). In a 1999 study of 4527 individuals who underwent HD testing, the "*frequency of suicide, suicide attempt, and psychiatric hospitalization following predictive testing was estimated to be 0.97% … however, half of these individuals were symptomatic at the time of the catastrophic event*" (Almqvist et al., 1999, 2003). This frequency is similar to that of the general population with no risk of HD (Almqvist et al., 2003). In another study of 202 individuals who underwent HD genetic testing, there were no reported suicide attempts for those who tested positive, but three for those who tested negative (Almqvist et al., 2003).

Studies investigating long-term psychological distress surrounding predictive genetic testing in the HD community have challenged conventional, presumptive correlations between test results and psychological distress (Almqvist et al., 1999, 2003; Crozier et al., 2015; Decruyenaere et al., 2003; Gargiulo et al., 2009; Hawkins et al., 2011). Gargiulo et al. (2009) found that a history of depressive episodes was predictive of post-test depression in those with a positive *and* negative result, suggesting that a history of depression may be a better predictor of post-test depression than test result status alone, and that individuals undergoing testing who have a history of depression should consider psychological support regardless of whether they receive a positive or negative test result. Additionally, those who have clear, specific reasons for testing (e.g., family planning) have less pre- and post-test distress and are more likely to test than those who test to reduce uncertainty (e.g., "*I just need to know*") (Decruyenaere et al., 2003; Scuffham & MacMillan, 2014). In summary, factors other than test result status may be more accurate predictors of long-term psychological distress.

There is no conclusive evidence that IPGC is superior to TGC in preventing adverse events or mitigating psychological distress. However, there are many tradeoffs between the two. In-person clinical care offers the ability to share physical space with the patient, which allows the GC to offer actions intended to console the patient (e.g., to place their hand on the patient's shoulder, hug the patient, or offer the patient a box of tissues if they begin to cry). In-person care often includes readily available access to emergency care in case the patient experiences acute psychological distress, since many genetic counseling sessions take place in hospitals or academic medical centers. However, TGC offers the patient the ability to receive the information wherever they are most comfortable and may allow support companions to join the patient in the session who may not have had the ability to attend if the session were conducted in-person.

HD genetic counseling is ideally provided alongside an interdisciplinary team that includes a neurologist. Patients may undergo a physical neurological exam and some clinics require an in-person blood draw to obtain a DNA sample for testing. With restrictions on in-person service delivery, some aspects of HD care can be challenging to perform and coordinate remotely. However, it is unclear if pre-pandemic conventions for in-person HD counseling were entirely necessary, and how GCs may feel about TGC for the HD community after the unprecedented shift to tele-care caused by the COVID-19 pandemic.

Rationale for Study

Given the limited knowledge of GCs' experiences with telegenetic counseling for Huntington's disease (HD-TGC), we designed a retrospective mixed-methods study to investigate genetic counselors' attitudes and perceptions of HD-TGC during the COVID-19 pandemic. HD genetic counseling has long been viewed as most appropriately delivered in-person despite little evidence supporting the view in terms of mitigating or preventing psychological distress in reaction to HD genetic counseling. The COVID-19 pandemic has offered an unexpected opportunity to investigate practices and perceptions of HD genetic counseling delivered through TGC. The results of this study will inform GC best practices for providing HD-TGC in the future.

Chapter 2: Study Background

Diffusion of Innovations Theory

Everett Rogers' Diffusion of Innovations theory (DOI) seeks to explain how and why behavioral changes may be accepted and adopted (Rogers, 2010). While not all of DOI is applicable to telegenetic counseling for Huntington's disease during the COVID-19 pandemic, there are elements that are useful and may provide insight in the investigation of HD-TGC as a burgeoning phenomenon. For example, Rogers describes the importance of social norms on the influence of adoption of innovation. Given the longstanding conventions of genetic counseling for HD and the paradigm set by the HD predictive test genetic counseling protocol, social norms are likely contributory to the decision of whether to adopt HD-TGC into practice.

The decision to implement HD-TGC was a *contingent innovative-decision*, in which the ultimate decision of whether to adopt an innovation is contingent upon a preceding decision (Rogers, 2010). When the U.S. Congress (acting as the *authority* in the *social system*) lifted restrictions on telehealth (TH) to allow medical institutions the option to provide TH where institutions deemed it appropriate, Congress provided the catalyst needed to allow for an expedited innovation-decision regarding telehealth. The decision of whether and to what degree to employ telehealth as an alternative mode of clinical care was then left to the collective authority of the medical institution's leadership.

According to Rogers, authority decisions generally result in the fastest rate of adoption of innovations; *collective decisions* that involve more contributors to the decision have the slowest rate of adoption (Rogers, 2010). Here, the authority's national emergency declaration followed by the recommendation to adopt TH — along with eased restrictions to allow institutions to do so — worked in concert to provide the opportunity for institutions to adopt telehealth quickly. Because the initial decision was a recommendation and not a mandate, the nature and extent of HD-TGC adoption varied between medical institutions. Almost 20 years before the COVID-19 pandemic, Walker and Whetton (2002) suggested that such a mandate may be necessary for TH to be adopted in healthcare:

> *"It might be necessary, for example, to mandate the use of technology, develop clear, precise procedures for use, and to recruit champions in strategic management positions, rather than adopt a more relaxed, voluntary adoption strategy"* (p. 74).

The authors further link DOI to healthcare institutions' innovation-decision processes by positing that the innovation-decision of whether to implement TH services is influenced by the institution's complexity and overall hierarchical structure (Walker & Whetton, 2002). In short, the delay to implement TH into regular practice in earnest may rest on two factors, both of which stem from healthcare providers holding the top of the hierarchy: [1] although TH eases the burden of travel for patients, the providers are not burdened to travel themselves; and [2] the implementation of TH would require "*changes to traditional practice and roles*," which could be met with resistance (Walker & Whetton, 2002).

In DOI, there are five main attributes of an innovation that contribute to its successful diffusion and adoption: [1] *relative advantage* over alternatives; [2] *compatibility*, such as with existing values and beliefs; [3] *complexity* or ease of use; [4] *trialability*, or ability to experiment with/try an innovation on a limited basis; and [5] *observability of results* from the innovation's use (Rogers, 2010). For HD-TGC to be implemented into regular practice, it would have to [1] have advantages over in-person care despite COVID-19 restrictions; [2] align with GCs' perceptions of what constitutes adequate care; [3] not be complex; [4] be flexible, adaptable, and not seen as a permanent commitment; and [5] result in observable success.

Specific Aims

Aim 1: Describe the nature and extent of genetic counseling services provided via telehealth to the Huntington's disease community (HD-TGC) pre- and post-onset of the COVID-19 pandemic (using a single, short online survey instrument). Examples of topics this survey assessed include elements of genetic counseling for Huntington's disease offered through telehealth, modes of delivery utilized for HD-TGC (e.g., phone, video conferencing), and conditions related to patient acceptance or decline of HD-TGC services.

Aim 2: Describe genetic counselors' attitudes and opinions toward the application of HD-TGC to various aspects of the HD counseling process (using a single, short online survey instrument). Questions probed genetic counselors' perceptions and ratings of their experience with HD-TGC. Examples of topics include comfort and confidence in providing HD-TGC, and the lasting potential of HD-TGC beyond the COVID-19 pandemic.

Aim 3: To explore contextual factors that may influence genetic counselors' perceptions of HD-TGC in detail by way of semi-structured interviews. A sub-sample selection of participants from the survey were asked to participate in a subsequent interview to describe their experience providing HD-TGC.

This retrospective, mixed-methods study involved two methods of assessment: [1] an online survey and [2] semi-structured interviews. U.S.-based, board-certified genetic counselors with 18 or more months of experience providing genetic counseling services for the Huntington's disease community were eligible for participation in both methods of study. There were no other criteria to meet for inclusion.

Chapter 3: Online Survey

Survey Recruitment

A total of 49 genetic counselors (GCs) completed the Qualtrics survey. GCs were eligible for the survey if they were a U.S.-based, board-certified GC with 18 or more months of experience providing genetic counseling services to the Huntington's disease community. A three-fold recruitment strategy was followed including the email of a detailed recruitment flyer with a link to the survey sent: [1] from the investigator to GCs within his network to participate or share the link with others who may be eligible; [2] from the Huntington's Disease Society of America (HDSA) to directors and genetic counselors of HDSA Centers of Excellence; [3] from the National Society of Genetic Counselors (NSGC); [4] from the American Board of Genetic Counseling (ABGC); and, [5] from the New York Mid-Atlantic Caribbean Regional Genetics Network (NYMAC) to subscribers of their respective listservs. Additionally, participants were provided with a link at the end of the survey and asked to forward it to any other GCs who may be eligible and interested in participating.

Survey Procedure

The online survey was constructed in Qualtrics and was open from May 4, 2021 through August 4, 2021. Although all GCs were asked about their attitudes and perceptions of HD-TGC services, GCs who indicated providing HD-TGC services at any point in the year 2020 (HD-TGC GCs) were asked additional questions to indicate the nature and frequency with which HD-TGC services were offered in 2020 and 2019, in a checkbox format (e.g., "*In 2020, I personally provided the following HD genetic counseling services for patients through the following modes of delivery: Phone, Video Chat, In-Person, N/A*") for elements of the HD genetic counseling protocol (e.g., pre-test counseling, results disclosure, follow-up counseling, etc.). HD-TGC GCs were also asked to recall the frequency in which patients may have mentioned examples of positive and negative feedback of HD-TGC services.

To assess attitudes and perceptions of HD-TGC services, HD-TGC GCs were asked to rate their agreement with 26 statements (e.g., "*I find it more difficult to respond to patients' expressions of emotion in tele-genetic counseling sessions than in in-person sessions.*") using a 5-point Likert scale ("Strongly disagree," "Disagree," "Neutral," "Agree," and "Strongly agree"). These statements were designed to assess GCs' comfort with providing HD-TGC services, their ability to establish interpersonal rapport, the quality of the service, and whether they felt GC services for HD would or should continue to be offered via telehealth, after lifting of COVID-19 restrictions on in-person clinical care. Genetic counselors who did not provide their services via TGC and instead provided their services in-person (IPGCs) were asked to rate their agreement with 18 statements similar to those described above but reworded to suit individuals who had no experience with HD-TGC (e.g., "*I think it would be more difficult to respond to patients' expressions of emotion in tele-genetic counseling sessions than in in-person sessions.*").

At the end of the survey, open text fields allowed all participants to describe portions of the survey they may have found confusing or difficult to answer accurately, or questions that were not asked that they felt should have been included in the survey. An additional open text field allowed participants to leave general comments that they wished to share about HD GC. Participants' contact information was collected and used for compensation distribution (emailed e-gift cards) and to facilitate interview recruitment.

Analytic Plan

Analysis used in this exploratory study is limited to descriptive statistics of GCs' reported perceptions and recollections of HD-TGC services they provided in 2019 and 2020. Survey prompts were different between participants who provided HD-TGC in 2020 (HD-TGC GCs) and those who did not (IPGCs); HD-TGC GCs were asked to assess HD-TGC from experience and

IPGCs were asked to assess HD-TGC as a concept. Survey response data was exported from Qualtrics into SPSS Version 28 for analysis.

Survey Results

Participant Characteristics.

A total of 49 GCs completed the Qualtrics survey. As reflected in Table 1, most participants worked for an academic medical center (76%) located in an urban area (69%) in 2020. Only one participant described the geographical setting of their institution of practice as rural. Participants practiced in 30 states and 40 cities. Most participants indicated their genetic counseling specialty as “Adult/Neuro” (32%) or “Adult/General” (19%) with other specialties reported less frequently. Note that counselors had the option of identifying multiple specialties.

Most participants had extensive experience as a GC; 57% reported more than 10 years of experience and 18% reported between 5-10 years. One-quarter of participants reported over 10 years of experience providing genetic counseling to the HD community and another third reported between 5-10 years of HD counseling experience. Half of the participants described themselves as the only GC in their practice or institution to provide HD genetic counseling. Most participants (76%) served as an HD GC for an HDSA Center of Excellence.

Table 1. Characteristics of survey participants (N=49).

		Count (n)	Column (N %)
Genetic counselor’s institution or practice setting in 2020	Academic Medical Center	37	75.5%
	Private Hospital/Clinic	5	10.2%
	Public Hospital/Clinic	4	8.2%
	Other	5	10.2%
Geographical setting of genetic counselor’s institution or practice in	Urban	34	69.4%
	Suburban/Small City	14	28.6%

2020	Rural	1	2%
Genetic counseling specialty (allowing multiple specialties)	Adult/Neuro	31	32.3%
	Adult/General	18	18.8%
	Cancer	13	13.5%
	Reproductive	1	1%
	Prenatal	7	7.3%
	Pediatrics	11	11.5%
	Cardio	5	5.2%
	Lab	2	2.1%
	Research	5	5.2%
	Other	3	3.1%
Experience as a practicing genetic counselor	More than 10 yrs	28	57.1%
	5 – 10 yrs	9	18.4%
	2 – 5 yrs	7	14.3%
	18 mos – 2 yrs	5	10.2%
Experience providing genetic counseling to HD community	More than 10 yrs	13	26.5%
	5 – 10 yrs	18	36.7%
	2 – 5 yrs	13	26.5%
	18 mos – 2 yrs	5	10.2%
Primary or head HD genetic counselor in your practice or institution?	Yes	37	75.5%
	No	12	24.5%
HD genetic counselor for an HDSA Center of Excellence	Yes	37	75.5%
	No	12	24.5%

Below, Table 2 provides an overview of participant practices regarding HD-TGC services in 2020. The majority of survey participants (74%) indicated providing HD-TGC during or since the year 2020. For clarity, GCs who provided HD-TGC will be referred to as HD-TGC GCs; GCs who did not provide HD-TGC will be referred to as in-person genetic counselors (IPGCs). The vast majority of these GCs also practiced in an academic medical center in an urban area and indicated their primary specialty to be “Adult/Neuro” or “Adult/General”.

HD-TGC GCs were well experienced with 69% reporting 10 years’ experience as a GC and 72% with more than 5 years as an HD GC, specifically. The majority of HD-TGC GCs described themselves as the primary or head HD GC in their practice or institution and served as an HD GC for an HDSA Center of Excellence. A smaller proportion of IPGCs had 10 or more years of experience as a GC (23%); a larger proportion of IPGCs (23%) than HD-TGC GCs (6%) had less than 2 years of experience . Participants who served at HD Center of Excellence and described themselves as the primary or head GC in their practice were more likely to provide HD-TGC services, whereas almost half of IPGCs did not serve at an HD Center of Excellence or consider themselves the primary or head GC in their practice.

Table 2. Characteristics of survey participants who provided HD-TGC in 2020 (HD-TGC GCs; n=36) and survey participants who did not provide HD-TGC in 2020 (IPGCs; n=13).

		Provided HD-TGC (N=36)		Did Not Provide HD-TGC (N=13)	
		Count (n)	Column (N %)	Count (n)	Column (N %)
Genetic counselor’s institution or practice setting in 2020	Academic Medical Center	31	63.3%	6	46.2%
	Private Hospital/Clinic	2	4.1%	3	23.1%
	Public Hospital/Clinic	1	2%	3	23.1%
	Other	4	8.2%	1	7.7%
Geographical setting of genetic counselor’s institution or practice in 2020	Urban area	26	72.2%	8	61.5%
	Suburban area	5	13.9%	2	15.4%
	Small city	4	11.1%	3	23.1%
	Rural	1	2.8%	0	0%
Genetic counseling	Adult/Neuro	24	35.8%	7	24.1%

specialty (allowing multiple specialties)	Adult/General	11	16.4%	7	24.1%
	Cancer	8	11.9%	5	17.2%
	Reproductive	1	1.5%	0	0%
	Prenatal	6	9%	1	3.4%
	Pediatrics	6	9%	5	17.2%
	Cardio	2	3%	3	10.3%
	Lab	2	3%	0	0%
	Research	5	7.5%	0	0%
	Other	2	3%	1	3.4%
Experience as a practicing genetic counselor	More than 10 yrs	25	69.4%	3	23.1%
	5 – 10 yrs	5	13.9%	4	30.8%
	2 – 5 yrs	4	11.1%	3	23.1%
	18 mos – 2 yrs	2	5.6%	3	23.1%
Experience providing genetic counseling to HD community	More than 10 yrs	12	33.3%	1	7.7%
	5 – 10 yrs	14	38.9%	4	30.8%
	2 – 5 yrs	8	22.2%	5	38.5%
	18 mos – 2 yrs	2	5.6%	3	23.1%
Primary or head HD genetic counselor in practice or institution	Yes	30	83.3%	7	53.8%
	No	6	16.7%	6	46.2%
HD genetic counselor for an HDSA Center of Excellence	Yes	30	83.3%	7	53.8%
	No	6	16.7%	6	46.2%

Nature & Extent of HD-TGC, 2019 v. 2020.

In 2019, the vast majority of HD genetic counseling (i.e., pre-test counseling, psychological assessment, genetic test result disclosure, and follow-up counseling) was done in-person, followed by phone, with video the minority in most categories (except test result disclosure). See Table 3 (top). In 2020, the majority of HD genetic counseling was done by video chat, followed by in-person and phone in more categories (except follow-up counseling).

Psychological assessments were performed equally in-person and over phone in 2019. See Table 3 (bottom). Almost all survey participants (92%) indicated that predictive genetic test result disclosure sessions “always” took place in-person prior to the COVID-19 pandemic, as displayed on Table 4. Trends from 2019 to 2020 for the magnitude of increased video chat services and decreased in-person services were similar between pre-test counseling, psychological assessment, test result disclosure, and follow-up counseling. However, the trend for utilizing phone services for test result disclosure sessions in 2020 was different from the trend observed for other services; test result disclosure sessions by phone increased from 2.5% in 2019 to 13.2% in 2020.

Table 3. Modality of HD genetic counseling services provided in 2019 and 2020. Responses from GCs with experience providing HD-TGC in 2020 (N=36). Participants could select more than one modality per service.

HD Genetic Counseling Service	Phone		Video Chat		In-Person		Total
	(N %)	(n)					(N)
2019							
Pre-test Counseling	20%	9	6.7%	3	73.3%	33	45
Psychological Assessment	17.4%	4	4.3%	1	78.3%	18	23
Test Results Disclosure	2.5%	1	10%	4	87.5%	35	40
Follow-up Counseling	29.3%	12	4.9%	2	65.9%	27	41
2020							
Pre-test Counseling	21.3%	13	45.9%	28	32.8%	20	61
Psychological Assessment	27.3%	6	45.4%	10	27.3%	6	22
Test Results Disclosure	13.2%	7	49.1%	26	37.7%	20	53
Follow-up Counseling	32%	16	40%	20	28%	14	50

Table 4. Frequency that predictive test results disclosures were conducted in-person before COVID-19 pandemic. No participants selected the option for "Not Often."

	Never		Inconsistent		Often		Always	
	(N %)	(n)	(N %)	(n)	(N %)	(n)	(N %)	(n)
Frequency	2.1%	1	2.1%	1	4.2%	2	91.7%	44

As displayed in Table 5, few HD-TGC GCs began providing HD-TGC prior to March/April 2020 (14%) while most indicated their initiation of TGC services occurred around or after March/April 2020. Two-thirds of HD-TGC GCs indicated that there was a period when all genetic counseling in their practice or institution was conducted through TGC. Most HD-TGC GCs reported TGC as a common mode of delivery ranging from several months to a year or more. A large majority of HD-TGC GCs indicated that HD-TGC was offered when surveyed (between May-Aug 2021) through their practice or institution, and almost all were personally providing the service. HD-TGC was often provided from home as well the GC's institutional office. Most HD-TGC GC's (63%) agreed that patients always had a support person with them for predictive genetic test result disclosure sessions provided through tele-genetic counseling, while a quarter indicated they did not (26%). Additionally, most HD-TGC GCs indicated that their workplace helped make the provision of HD-TGC to patients as easy as possible for HD-TGC GCs (n=30/36, 83%), and made access as easy as possible for their patients (n=29/36, 81%; data not represented in table).

Table 5. Miscellaneous details about HD-TGC services.

		Count (n)	Column (N %)
Time period GC started providing HD-TGC services	Before March/April 2020	5	13.9%
	Around March/April 2020	17	47.2%

	After March/April 2020	14	38.9%
All genetic counseling in GC's practice/institution transitioned to TGC for some time	Yes	24	66.7%
	No	12	33.3%
Duration HD-TGC provided as a common mode of delivery	A year or more	16	44.4%
	Several months	15	41.7%
	A few months or less	5	13.9%
HD-TGC offered in GC's practice/institution as of May-Aug 2021	Yes	32	88.9%
	No	4	11.1%
GC personally providing HD-TGC as of May-Aug 2021	Yes	31	86.1%
	No	5	13.9%
GC provided HD-TGC from home in 2020	Yes	32	88.9%
	No	4	11.1%
GC provided HD-TGC from office or institution in 2020	Yes	25	69.4%
	No	11	30.6%
“Patients always had a support person with them for HD predictive test results disclosure sessions provided through HD-TGC”	Strongly Agree	7	20.0%
	Agree	15	42.9%
	Neutral	4	11.4%
	Disagree	8	22.9%
	Strongly Disagree	1	2.9%

As displayed in Table 6, GCs recalled that in 2019, most HD services provided by a clinician other than the GC were conducted in-person. All video and phone services provided by other team members were less than 9% in 2019, except for follow-up counseling provided by phone (30%). However, even neurological exams were increasingly offered via telehealth in 2020. In 2020, most neurological exams were performed in-person (50%), followed by video chat (44%), and phone (6%). In 2020, psychological assessments performed by a clinician other than the GC were performed equally in-person as through video chat (41%). In 2020, if genetic test results were disclosed by a clinician other than the GC, they mostly took place in-person (46%), followed by video chat (44%), and by phone (11%). Most follow-up counseling provided by a clinician other than the GC was done via video (43%) in 2020, followed by in-person (36%), and phone (21%).

Table 6. Modality of HD services provided by other team members in 2019 and 2020. Responses from GCs with experience providing HD-TGC in 2020 (N=36). Participants could select more than one modality per service.

HD Service	Phone		Video Chat		In-Person		Total
	(N %)	(n)					(N)
2019							
Neurological Exam	0%	0	7.7%	2	92.3%	24	26
Psychological Assessment	8.7%	2	8.7%	2	82.6%	19	23
Test Results Disclosure	4.2%	1	0%	0	95.8%	23	24
Follow-up Counseling	30.3%	10	6.1%	2	63.6%	21	33
2020							
Neurological Exam	5.6%	3	44.4%	24	50%	27	54
Psychological Assessment	18.2%	8	40.9%	18	40.9%	18	44
Test Results Disclosure	10.9%	5	43.5%	20	45.7%	21	46
Follow-up Counseling	20.8%	11	43.4%	23	35.8%	19	53

Table 7 reports requests and offers of HD-TGC in 2019; almost a third of GCs indicated that patients had requested HD-TGC but few patients were offered the services. Most GCs indicated that if patients declined HD-TGC services in 2020, they did so with the intention to see a GC in-person and few of these patients changed their mind.

Table 7. Requests and offers of HD-TGC in 2019; declined HD-TGC services in 2020. Responses from GCs with experience providing HD-TGC in 2020 (N=36).

		Count (n)	Column (N %)
In 2019, patients requested HD-TGC	Yes	7	30.4%
	No	16	69.6%
In 2019, patients were offered HD-TGC	Yes	3	9.1%
	No	30	90.9%
In 2020, patients declined HD-TGC with the intention of seeing a provider in-person at a future date	Yes	11	91.7%
	No	1	8.3%
In 2020, patients who declined HD-TGC eventually decide to use TGC instead of in-person care	Yes	1	14.3%
	No	6	85.7%

Genetic Counselor Perceptions of Patient Attitudes Toward HD-TGC.

All HD-TGC GCs (N=36) were asked to recall the frequency of which patients may have mentioned examples of potential advantages or drawbacks of HD-TGC. As indicated in Table 8.1, the advantage most recalled was that patients did not have to travel (94%), followed by the increased ease for support persons or caretakers to attend visits (90%). Around three quarters of responses indicated that patients mentioned not having to take time off from work and being more comfortable at home. Around three quarters of GCs also indicated that patients mentioned difficulty with technology and poor connection in HD-TGC sessions, such as poor internet connectivity or phone reception. Other drawbacks such as privacy or quality of service concerns were infrequently or not at all mentioned.

All but one HD-TGC GC agreed that patients seemed satisfied with HD-TGC services, and most (83%) agreed that patients seemed comfortable (see Table 8.2 below). Most HD-TGC GCs (81%) reported positive patient feelings about HD-TGC. No HD-TGC GCs agreed that patients expressed negative feelings about HD-TGC.

Table 8.1. GC recollections of patient-mentioned advantages and drawbacks of HD-TGC. Responses from GCs with experience providing HD-TGC in 2020 (N=36).

		Count (n)	Column (N %)
Advantages			
Saved time	Yes, frequently	14	40.0%
	Yes, but not often	11	31.4%
	No	6	17.1%
	Maybe, I don't remember	4	11.4%
Saved money	Yes, frequently	8	22.9%
	Yes, but not often	8	22.9%
	No	12	34.3%
	Maybe, I don't remember	7	20.0%
Did not have to travel	Yes, frequently	27	77.1%
	Yes, but not often	6	17.1%
	No	1	2.9%
	Maybe, I don't remember	1	2.9%
Did not have to take time off from work	Yes, frequently	13	37.1%
	Yes, but not often	14	40.0%
	No	6	17.1%
	Maybe, I don't remember	2	5.7%
More comfortable to do at home	Yes, frequently	13	37.1%
	Yes, but not often	13	37.1%
	No	6	17.1%
	Maybe, I don't remember	3	8.6%
Helped due to HD-related mobility issues	Yes, frequently	11	31.4%
	Yes, but not often	5	14.3%
	No	12	34.3%

	Maybe, I don't remember	7	20.0%
Made it easier for support person(s) or caretaker(s) to attend visit	Yes, frequently	16	45.7%
	Yes, but not often	12	34.3%
	No	4	11.4%
	Maybe, I don't remember	3	8.6%
Drawbacks			
Bad internet connection / phone reception	Yes, frequently	5	14.3%
	Yes, but not often	22	62.9%
	No	7	20.0%
	Maybe, I don't remember	1	2.9%
Difficulty with technology	Yes, frequently	4	11.4%
	Yes, but not often	22	62.9%
	No	8	22.9%
	Maybe, I don't remember	1	2.9%
Concerns of internet or information security/privacy	Yes, frequently	0	0.0%
	Yes, but not often	0	0.0%
	No	34	97.1%
	Maybe, I don't remember	1	2.9%
Concern with lack of privacy due to presence of family members, roommates, or others in the home or space during session	Yes, frequently	1	2.9%
	Yes, but not often	7	20.0%
	No	26	74.3%
	Maybe, I don't remember	1	2.9%
Concern that care provided via telehealth may be inferior to in-person care	Yes, frequently	2	5.7%
	Yes, but not often	5	14.3%
	No	27	77.1%
	Maybe, I don't remember	1	2.9%

Table 8.2. GC perceptions of patients' feelings in response to receiving HD-TGC services. Responses from GCs with experience providing HD-TGC in 2020 (N=36).

	Disagree or Ambivalent		Agree		Strongly Agree	
	(N %)	(n)	(N %)	(n)	(N %)	(n)
Overall, patients have seemed satisfied with the services I provided via tele-genetic counseling.	2.8%	1	58.3%	21	38.9%	14
Overall, patients have seemed comfortable using tele-genetic counseling services for HD.	16.7%	6	63.9%	23	19.4%	7
Patients expressed positive feelings about tele-genetic counseling to the GC.	19.4%	7	38.9%	14	41.7%	15
Patients expressed negative feelings about tele-genetic counseling to the GC.	100.0%	35	0.0%	0	0.0%	0

Genetic Counselor Attitudes & Perceptions of HD-TGC.

GCs with HD-TGC Experience (HD-TGC GCs).

Table 9.1. Attitudes & perceptions of HD-TGC from GCs who provided HD-TGC services in 2020 (HD-TGC GCs; N=36).

	Disagree or Ambivalent		Agree		Strongly Agree	
	(N %)	(n)				
Comfort with HD-TGC Services						
Before the pandemic, I was uncomfortable with the idea of using tele-genetic counseling for HD sessions.	22.2%	8	52.8%	19	25%	9
I feel comfortable with providing tele-genetic counseling for the HD community.	22.2%	8	44.4%	16	33.3%	12

I am more comfortable providing tele-genetic counseling for HD now than I was before the onset of COVID-19 restrictions.	16.7%	6	44.4%	16	38.9%	14
I would prefer to provide my HD genetic counseling services in-person with the patient.	41.7%	15	33.3%	12	25.0%	9
I would prefer to provide tele-genetic counseling if in-person counseling requires the use of masks and social distancing.	41.6%	15	41.7%	15	16.7%	6
Interpersonal Rapport in HD-TGC Sessions						
I find it more difficult to respond to patients' expressions of emotion in tele-genetic counseling sessions than in in-person sessions.	57.2%	20	31.4%	11	11.4%	4
I find it more difficult to identify non-verbal cues of distress in tele-genetic counseling sessions than in in-person sessions.	45.7%	16	40.0%	14	14.3%	5
In tele-genetic counseling sessions, it is difficult to establish emotional rapport with the patient.	74.2%	26	22.9%	8	2.9%	1
Quality of HD-TGC Services						
I am confident that the informational needs of patients were met in HD tele-genetic counseling sessions.	5.5%	2	52.8%	19	41.7%	15
I am confident that the emotional needs of patients were met in HD tele-genetic counseling sessions.	41.7%	15	38.9%	14	19.4%	7
I am confident that assessment of suicidal ideation and risk can be adequately performed in tele-genetic counseling sessions.	42.9%	15	51.4%	18	5.7%	2
"In an Ideal World…"						
In an ideal world, pre-test counseling for HD predictive testing would not be provided via tele-genetic counseling.	63.9%	23	19.4%	7	16.7%	6

In an ideal world, results disclosure for HD predictive testing would not be provided via tele-genetic counseling.	41.6%	15	30.6%	11	27.8%	10
In an ideal world, neurological exams for HD would not be provided via telehealth.	16.7%	6	47.2%	17	36.1%	13
In an ideal world, psychological assessments for HD would not be provided via telehealth.	52.8%	19	22.2%	8	25.0%	9
Lasting Potential of HD-TGC						
I imagine that tele-genetic counseling for HD will routinely be offered as an option beyond the COVID-19 pandemic.	30.5%	11	52.8%	19	16.7%	6
I imagine that tele-genetic counseling for HD predictive test results disclosure will routinely be offered as an option beyond the COVID-19 pandemic.	47.2%	17	47.2%	17	5.6%	2
I feel that tele-genetic counseling for HD should continue to be offered as an option for patients.	13.9%	5	63.9%	23	22.2%	8
I feel that tele-genetic counseling for HD predictive test results disclosure should continue to be offered as an option beyond the COVID-19 pandemic.	16.7%	15	44.4%	16	13.9%	5

Table 9.1 displays HD-TGC GC attitudes and perceptions toward HD-TGC services in 2020. Most of these GCs indicated that they were uncomfortable with the idea of using HD-TGC prior to the pandemic. However, their comfort level changed by the time of the survey when over three-quarters of counselors agreed or strongly agreed that they were comfortable providing the service, indicating a positive change for a large majority. Over half of the HD-TGC GCs (58%) said they would prefer to provide their services in-person but would prefer HD-TGC over in-person services if it required the use of masks and social distancing. A quarter of HD-TGC GCs (25%) would still prefer in-person genetic counseling.

HD-TGC GCs were split on whether they found it difficult to respond to patients' expressions of emotion, with 46% indicating they did and 43% indicating they did not. Over half of HD-TGC GCs found it difficult to identify non-verbal cues of distress, while a third did not. Finally, a large majority of HD-TGC GCs (80%) did not find it difficult to establish emotional rapport in an HD-TGC session.

Almost all HD-TGC GCs (94%) were confident that patients' informational needs were met in HD-TGC sessions, and 58% of HD-TGC GCs were confident patients' emotional needs were met. A majority of HD-TGC GCs (57%) indicated they were confident that they could adequately assess suicidal ideation and risk in HD-TGC sessions, but 20% were not confident in this assessment.

HD-TGC GCs were asked to imagine providing their services "in an ideal world." A little over a third of HD-TGCs agreed that HD-TGC pre-test counseling would not be offered via HD-TGC in an ideal world, yet nearly two thirds did not agree. A little over half of HD-TGC GCs (58%) agreed that results for predictive genetic testing for HD would ideally not be provided via HD-TGC; 42% disagreed or felt ambivalent. Most (83%) agreed neurological exams would ideally not be provided via telehealth. Nearly half of HD-TGC GCs (47%) thought that psychological assessments for HD would not be offered via HD-TGC in an ideal world, while over half did not agree.

Most HD-TGC GCs believe HD-TGC — both in general (86%) and predictive test results disclosure sessions specifically (58%) — should continue to be offered as an option. A quarter of HD-TGC GCs do not think predictive genetic test result disclosure sessions should continue to be offered via HD-TGC beyond the COVID-19 pandemic. Over two-thirds of HD-TGC GCs (69%) imagine that HD-TGC will routinely continue to be offered as an option beyond the COVID-19 pandemic. Regarding HD-TGC for predictive genetic test results disclosure sessions, a little over half of HD-TGC GCs imagine HD-TGC will continue to routinely be offered as an option beyond the pandemic, while a little over a third disagree.

GCs without HD-TGC Experience (IPGCs).

Table 9.2. Attitudes & perceptions of HD-TGC from GCs who did not provide HD-TGC services in 2020 (IPGCs; N=13).

	Disagree or Ambivalent		Agree		Strongly Agree	
	(N %)	(n)				
Comfort with HD-TGC Services						
I feel comfortable with tele-genetic counseling as an option for the HD community.	30.8%	4	61.5%	8	7.7%	1
I am more comfortable with tele-genetic counseling for HD as an option now than I was before the onset of COVID-19 restrictions.	30.8%	4	53.8%	7	15.4%	2
I would prefer to provide my HD genetic counseling services in-person with the patient.	15.4%	2	15.4%	2	69.2%	9
I would prefer to provide tele-genetic counseling if in-person counseling requires the use of masks and social distancing.	76.9%	10	23.1%	3	0.0%	0
Interpersonal Rapport in HD-TGC Sessions						
I think it would be more difficult to respond to patients' expressions of emotion in tele-genetic counseling sessions than in in-person sessions.	46.2%	6	46.2%	6	7.6%	1
I think it would be more difficult to identify non-verbal cues of distress in tele-genetic counseling sessions than in in-person sessions.	7.7%	1	69.2%	9	23.1%	3
In tele-genetic counseling sessions, I think it would be more difficult to establish emotional rapport with the patient.	46.1%	6	30.8%	4	23.1%	3

Quality of HD-TGC Services						
I am confident that the informational needs of patients could be met in HD tele-genetic counseling sessions.	7.7%	1	30.8%	4	61.5%	8
I am confident that the emotional needs of patients could be met in HD tele-genetic counseling sessions.	46.2%	6	53.8%	7	0.0%	0
I am confident that assessment of suicidal ideation and risk can be adequately performed in tele-genetic counseling sessions.	53.8%	7	46.2%	6	0.0%	0
“In an Ideal World…”						
In an ideal world, pre-test counseling for HD predictive testing would not be provided via tele-genetic counseling.	53.8%	7	38.5%	5	7.7%	1
In an ideal world, results disclosure for HD predictive testing would not be provided via tele-genetic counseling.	7.7%	1	53.8%	7	38.5%	5
In an ideal world, neurological exams for HD would not be provided via telehealth.	0.0%	0	53.8%	7	46.2%	6
In an ideal world, psychological assessments for HD would not be provided via telehealth.	53.8%	7	30.8%	4	15.4%	2
Lasting Potential of HD-TGC						
I imagine that tele-genetic counseling for HD will routinely be offered as an option beyond the COVID-19 pandemic.	23.1%	3	76.9%	10	0.0%	0
I imagine that tele-genetic counseling for HD predictive test results disclosure will routinely be offered as an option beyond the COVID-19 pandemic.	69.2%	9	30.8%	4	0.0%	0
I feel that tele-genetic counseling for HD should continue to be offered as an option for patients.	30.8%	4	53.8%	7	15.4%	2

I feel that tele-genetic counseling for HD predictive test results disclosure should continue to be offered as an option beyond the COVID-19 pandemic.	53.8%	7	38.5%	5	7.7%	1

The attitudes and perceptions of HD-TGC from GCs who did not provide HD-TGC services in 2020 (IPGCs; N=13) are displayed on Table 9.2. A majority (69%) of these GCs indicated that they feel more comfortable with HD-TGC services as an option for the HD community than they did before onset of the pandemic and currently feel comfortable with HD-TGC as an option. Most of these IPGCs (85%) indicated they currently prefer to provide in-person services but only a few preferred HD-TGC if considering in-person services required the use of masks and social distancing.

Also displayed on the table are findings regarding interpersonal rapport during HD-TGC sessions and perceived quality of HD-TGC service. Almost all IPGCs thought it would be difficult to identify non-verbal cues of distress and about half thought it would be difficult to respond to patients' expressions of emotion and establish emotional rapport in HD-TGC sessions. While almost all IPGCs (92%) were confident that patients' informational needs could be met in an HD-TGC session, IPGCs were split in assessing their confidence that emotional needs of patients could be met and that assessment of suicidal ideation and risk could be adequately performed in HD-TGC sessions.

Finally, Table 9.2 displays participant attitudes toward HD-TGC in an ideal world and the lasting potential of the technology. The small group of IPGCs were largely split in imagining that HD-TGC would not be provided for pre-test counseling in an ideal world. However, almost all thought results of predictive genetic testing for HD should not be provided via HD-TGC. All IPGCs agreed that neurological exams would ideally not be provided via telehealth and again were largely split on use of HD-TGC for psychological assessments.

While the majority of IPGCs agreed that HD-TGC would continue to be offered as an option for the HD community beyond the COVID-19 pandemic, they did not agree that predictive test results disclosure sessions would. Finally, the majority of IPGCs felt that HD-TGC should continue to be offered beyond COVID-19 but were again split on whether HD-TGC should continue to be offered for predictive test results disclosure sessions.

Open-Field Prompts.

Three open-field responses at the end of the online survey encouraged participants to identify any questions they found challenging to answer, to describe any topics participants felt were absent from the survey, and finally to describe anything else about the participants' experiences with HD genetic counseling that they may wish to mention. Responses often provided additional context for interpreting survey responses, for example:

> *"There was a learning curve to tele-genetics. At the beginning, not having had any training in video counseling, I felt I was unable to provide the emotional support needed by my patients. But, after a bit of time, I was able to realize it is not that different from being in the room with a patient. I felt more comfortable with being able to provide emotional support and information and make the patient comfortable by having them in their own home. I didn't have to worry about them driving home after a positive testing result. They had their people there, and it was a safe place."*

The last open-field question asked if there was anything about participants' experience in providing genetic counseling to the HD community, in-person or via telemedicine, that they would like to tell us about. This question elicited responses from almost half of the participants. The most common topic shared throughout responses was predictive test results disclosure sessions (n=8/23, 34.8%). One GC's response illustrates the depth made possible by allowing the participant to describe their perspective on HD-TGC result disclosure sessions in a freeform capacity:

> *"Results disclosure over video was hard for me at first, but I do see several advantages to the patient and their support person being at home. I have had a few situations where the support person is also not physically with the*

> *patient but on video chat in a different location. I also make sure someone is immediately available at their physical location and we discuss the "set-up" as part of the pre-test counseling. Most people plan and take our advice. Some people don't and results disclosure is not ideal (for example, in their car during a break from their workday)."*

Discussion of Survey Findings

Genetic counseling for Huntington's disease (HD) is often treated as an exception to genetic counseling practice for most other diseases, because of the severity of the disease, the certainty associated with inheritance, and the potential for psychosocial distress that can accompany a risk or diagnosis. The caution surrounding genetic counseling for HD resulted in a disease-specific protocol recommending multiple in-person visits. COVID-19 shifted much of genetic counseling to telegenetic counseling (TGC), unfamiliar to many GCs, but especially uncomfortable for GCs when it came to pushing the boundaries of HD genetic counseling. As evident in survey findings, most GCs were uncomfortable with the idea of HD-TGC before the COVID-19 pandemic, grew more comfortable with HD-TGC over time and considered themselves comfortable with HD-TGC when surveyed.

Most of the 13 GCs who had not provided HD-TGC services in 2020 (IPGCs) indicated that their current preference is to provide HD genetic counseling services in-person. However, the proportion of GCs who preferred to provide their HD genetic counseling services in-person but *had* experience providing HD-TGC services in 2020 (HD-TGC GCs) indicated a substantially lower preference for in-person counseling. This may reflect self-selection or cognitive dissonance; those more likely to provide HD-TGC services had more positive attitudes pre-pandemic, or those who changed their behavior aligned their attitudes once their practice changed. In any event, this group is different than those who did not offer TGC services. Most HD-TGC GCs indicated that their workplace helped make it as easy as possible for them to provide their services to patients via HD-TGC; perhaps ample institutional support could influence GCs to have favorable opinions. IPGCs' institutions may not have the resources or wherewithal to support HD-TGC.

By simple comparison, providing counseling services in-person allows the counselor to gain a full view of the patient whereas video counseling offers a limited view. The view of a patient in video counseling sessions may be further restricted when poor internet quality results in low resolution of images. Phone counseling of course loses visual exchange with the patient entirely. Because psychosocial counseling can benefit from assessment of facial expressions, body language, and other non-verbal cues, it's not surprising that GCs would be hesitant to embrace an alternative to in-person care, especially for HD, where much attention is paid to the psychological state of the patient.

Most IPGCs imagined HD-TGC would hinder psychosocial interaction and assessment with patients. For example, almost all IPGCs indicated that they thought it would be difficult to identify non-verbal cues of distress and most IPGCs indicated a lack of confidence that patients' emotional needs could be met via HD-TGC. In contrast, HD-TGC GCs reported more favorably for HD-TGC regarding interpersonal rapport and psychosocial assessment, again suggesting experience with HD-TGC may afford some reassurance for providers who lack that experience. Although the majority of HD-TGC GCs felt that their ability to assess patients' potential suicidal ideation and risk was adequate in HD-TGC sessions, a sizable minority did not agree.

Future studies should focus on how best to assess for acute psychological distress and suicidal risk in virtual counseling sessions, and what interventions are best to employ for mitigation and prevention. Despite the recommendations that individuals undergoing HD genetic counseling have a support person join them for their genetic counseling sessions, some HD-TGC GCs in this survey indicated that this didn't always happen. Perhaps ensuring that a support person is with the patient for virtual sessions could help in scenarios in which the patient is highly distressed. It is worth noting that this question did not capture whether GCs experienced an HD-TGC session in which a patient was extraordinarily distressed or suicidal.

Given the availability of open field prompts at the end of the survey and the memorable nature of such a reaction, it is likely a GC would have mentioned a particularly adverse reaction there.

Most GCs seemed comfortable with pre-test counseling being conducted via HD-TGC, but many felt that predictive test results disclosure sessions would be provided in-person in an ideal world. Generally, GCs worry that a patient may have an adverse reaction in response to a predictive genetic test result. The existing HD predictive testing protocol rests on the assumption that in-person HD genetic counseling can better prevent or mitigate potential distress or adverse reactions. In this survey, almost all participants indicated that predictive genetic test results disclosure sessions “always” took place in-person prior to the COVID-19 pandemic. Despite reported preferences for providing their services in-person, the majority of HD-TGC GCs responded that they imagined HD-TGC would and *should* continue to be offered as an option beyond the COVID-19 pandemic — this was when asked explicitly about HD-TGC in general and predictive test results disclosure sessions, specifically. We interpret the positive response — despite voiced awareness of the potential for psychological distress in these situations — as indicative of a level of confidence in HD-TGC as an adequate option for HD genetic counseling.

Advantages and drawbacks of TGC have already been presented in the literature; this study confirmed that those advantages and drawbacks extend to HD-TGC, specifically. The most common advantages of HD-TGC related to increased access to HD genetic counseling: not having to travel, the increased ease for support persons to join sessions, and not having to take time off from work for appointments. Given the weight of potentially distressing information learned in an HD session, GCs may want to utilize HD-TGC to reduce burdens and increase comfort. Regarding drawbacks of HD-TGC, GCs reported that patients most often mentioned difficulty with technology and poor connection in HD-TGC sessions, such as poor internet connectivity or phone reception. It is likely that at least some of these IT problems diminished with user experience.

Overall, HD-TGC GCs' assessments of HD-TGC were consistently positive. The majority of HD-TGC GCs were confident that patients' informational and emotional needs were met in HD-TGC sessions, and that patients seemed both satisfied and comfortable with the services they received. The majority of GCs (HD-TGCs and IPGCs) reported feeling comfortable with HD-TGC and that their comfort with HD-TGC grew over time. Many GCs would prefer to provide their services in-person, particularly predictive test results disclosure sessions. Moreover, despite reservations for utilizing HD-TGC for predictive test results disclosure sessions, most HD-TGC GCs believed that HD-TGC — including predictive test results disclosures — would and *should* continue to be offered as an option for the HD community. One GC's response to one of the open-field prompts feels appropriately summative:

> *"In general, it went better than expected, although we had some technical issues from time to time, especially for those who have early HD and can't manage the technology well. Patients seem to prefer it and we're finding that this is challenging our belief that this should always be done in-person. I prefer to be in-person but recognize this may be the way services are provided in the future."*

The conventional restrictions upon genetic counseling for HD are rooted in an abundance of caution. However, the findings of this survey suggest that excellent care is still possible through HD-TGC and is not predicated on in-person clinical visits. Genetic counseling for HD is more accessible with the availability of HD-TGC as an option and is likely to continue beyond the COVID-19 pandemic.

Chapter 4: Interviews

Interview Recruitment

Survey respondents with two or more years of experience providing genetic counseling services for HD patients were eligible for a semi-structured interview. The purpose of the semi-structured interviews was to elicit more detailed accounts of GCs' experiences providing HD-TGC services. Interviewees were purposively recruited via e-mail, with the goal of maximizing the variety of the sample with respect to geographical location.

Interview Procedure

Participants gave verbal consent at the beginning of each interview. All interviews were conducted by WS. The interviews ranged in length from 35 minutes to an hour, with an average length of 45 minutes. Interviews were conducted between June 21, 2021 and July 12, 2021. Sixteen interviews were conducted via Zoom with video off; one GC left their video on as a personal choice.

Instrument Development and Data Collection.

A semi-structured interview guide was developed based on a review of background literature and feedback from two GCs who practice in adult neurology. See Appendix IV. Interviewees were first asked to describe their background as a GC and what their day-to-day practice might look like. From there, WS probed the GCs to describe [1] the impact of COVID-19 pandemic on genetic counseling services, [2] their experiences with HD-TGC specifically, and [3] their attitudes toward HD-TGC. Roughly 27 questions were asked in each interview. When the interview neared 45 minutes, WS would ask interviewees "*What haven't I asked you that you think is important for me to know about the way you think about tele-genetic counseling services for Huntington's disease?*" to capture any remaining thoughts from the interviewee.

Although the interview guide was largely developed prior to initiating recruitment, several modifications were made after the first few interviews to improve the clarity of questions and to eliminate redundant questions.

Analytic Plan

Audio was recorded through Zoom and transcribed by a third-party transcription company. Personal & institutional identifiers such as names & locations were replaced with code numbers and generic labels to maintain participant confidentiality. Transcripts were coded using a combined deductive and inductive approach using MaxQDA Standard 2018 (Release 18.2.5). The preliminary codebook was revised in an iterative manner to capture emerging themes. The first four transcripts were coded in sequentially staggered pairs: an early transcript paired with a later transcript (e.g., interview #1 alongside #10). These four transcripts were coded by WS and a second coder. After reaching consensus, all 16 transcripts were then coded by WS in early October 2021 with the full, final codebook. WS compared coded excerpts for overlapping themes and compiled them for quantification.

Interview Results

Participant Characteristics.

Interview participants' characteristics are summarized in Table 10. Seventeen interviews were conducted; 16 were included in this study. The 16 included in this study were with genetic counselors with experience providing HD-TGC in 2020 (HD-TGC GCs). The one interview excluded from analysis was with a GC who had not provided HD-TGC services and was interviewed in an exploratory manner to inform the interview guide from another perspective. No additional domains or questions were gained from this interview.

Most interviewees indicated that they were the primary HD GC in their practice (n=14), served as an HD GC at an HDSA Center of Excellence (n=14), worked in an academic medical center (n=14) in an urban area (n=11), and had more than 5 years' experience as a GC for the HD community (n=10). Interviewees mostly indicated their GC specialty to be "Adult/Neuro" (39%). Overall, interviewees represented 14 unique cities and 11 unique states of the U.S. No interviewees indicated their practice setting as rural.

Table 10. Characteristics of GCs interviewed (N=16). Participants could select more than one genetic counseling specialty.

		Count	Column N %
Genetic counseling specialty	Adult/Neuro	12	38.7%
	Adult/General	5	16.1%
	Cancer	3	9.7%
	Prenatal	1	3.2%
	Pediatrics	3	9.7%
	Lab	1	3.2%
	Research	5	16.1%
	Other	1	3.2%
Experience as a practicing genetic counselor	More than 10 yrs	11	68.8%
	5 - 10 yrs	1	6.3%
	2 - 5 yrs	3	18.8%
	18 mos - 2 yrs	1	6.3%
Experience providing genetic counseling to HD community	More than 10 yrs	7	43.8%
	5 - 10 yrs	3	18.8%
	2 - 5 yrs	5	31.3%
	18 mos - 2 yrs	1	6.3%
Primary or head HD GC in practice or institution	Yes	14	87.5%
	No	2	12.5%

Serve as an HD GC for an HDSA Center of Excellence	Yes	14	87.5%
	No	2	12.5%

Interview Results.

GCs saw both benefits and drawbacks related to HD-TGC, as well as facilitators and barriers to successful implementation of HD-TGC into regular practice. Benefits of HD-TGC included increased accessibility and reduced patient burden. GCs also faced physical, technological, and logistical barriers in the transition to HD-TGC. GCs also discussed their preferences between modalities and perceptions of whether HD-TGC would continue to be offered as an option beyond the COVID-19 pandemic. The treatment of HD as exceptional from other conditions was also a prominent theme. The following section will review these findings in more detail.

Transition to HD-TGC.

All GCs (n=16) described an interruption of their regular services in response to the declaration of the COVID-19 pandemic as a national emergency. Half of the GCs described this interruption in services as taking a month or longer; for some it took less than a month (n=4). One GC said their clinic was operational in less than a week after their institution implemented a stay-at-home order. This GC attributed the seamlessness of their institution's transition to having a video platform and telehealth system in place before COVID-19 restrictions were mandated.

Many GCs (n=7) described delaying admission of new patients for weeks to months in hope that pandemic concerns would ease, or that restrictions would be lifted. Many GCs (n=5) said that their HD-TGC protocol was similar to their HD protocol used pre-pandemic, aside from using remote/video communication in place of in-person sessions (i.e., number of genetic counseling sessions for someone undergoing predictive genetic testing remained the same).

Pre-pandemic, patients would often have their blood drawn for genetic testing, and this blood draw would most often happen in-person at the same clinic in which the patient had received genetic counseling. After onset of the pandemic, one GC mentioned using third-party saliva collection kits that could be shipped directly to patients to obtain DNA samples, despite having their own institutional laboratory that could perform DNA analysis for HD:

> *"We have the capability of ordering genetic testing in-house for Huntington's disease, but it was just a little more cumbersome to do so because our lab wasn't set up to just be able to send patient kits and things like that. So, a lot of times we were just using an outside lab just because it was easier to use."—12*

Lack of Institutional Guidance.

Some GCs (n=4) described a lack of guidance in their transition to providing TGC, that guidelines for providing telehealth varied between departments and were often unclear. GCs had to improvise and make personal judgment calls where they may have leaned on the established protocol in the past.

> *"It's also different from department to department ... I mean, it's been kind of confusing, I guess, to know who can continue to offer telemedicine services, and when they have to stop-- things like that."—12*

> *"It was kind of like the wild west at the start-- like do what you can, and just [let schedulers] know how to get links to people. And a lot of that I was doing myself just to set up the visits and get people the right link and tell them where to go and explain everything."—13*

Several GCs (n=6) mentioned that the disruption caused by COVID-19 led to an increased burden on the GC to coordinate logistics of the clinic workflow or patient contact in ways they would not have had to do in the past. One GC worried that the lack of clear guidelines or policy led to unequal care distribution to their patients, specifically in deciding who should have to come to clinic in-person and who could receive TGC:

> *"... We didn't have a set protocol of 'this is exactly how we're going to do it' for every patient. It was much more 'I, as the genetic counselor at that first visit would kind of assess how anxious I think they were, what kind of background they were coming from, did they have exposure to HD, did they really know and*

> *have good reasons for testing,' and then I was using that information to kind of decide 'does it feel safe to continue to offer them telegenetic meeting with an MD for visit number two and results disclosure,' or just in my gut do I think they should be coming in in-person for those visits? Which I feel like I did okay at making those assessments, but it's a lot harder to stand by a decision to make somebody come in when that's not your clinic's policy for every patient. That it just is harder, if you have a patient that you really are worried about but they're giving you pushback that they don't want to come in, it's harder to say they have to when you haven't been making other people.*" *—9*

GCs described frustration with not having a voice of authority to offer clear recommendations in the uncertainty of the COVID-19 pandemic. One GC felt that the guidance should have come from the Huntington's Disease Society of America (HDSA), who have established the current standard of care via their genetic testing protocol:

> "*We didn't have a lot of guidance. I'd say that was the biggest challenge with presymptomatic testing, was that we did not have a lot of guidance from HDSA, who we get funding from. We get funding from them because we follow their protocol. I mean, for other reasons as well, but probably because we follow their protocol, about what would be acceptable. Because HDSA says everything's in-person: your pre-test visit's in-person, your post-test visit is in-person. So, we really waited until our department let us have people in-person, and then we started hearing from a lot of other programs that they were doing a lot of virtual counseling. So, we're like 'Okay, if they are, we will too.'*" *—5*

Another GC felt that HDSA is an appropriate voice of authority for guidance, and should update the genetic testing protocol to incorporate telehealth going forward:

> *"I think the HDSA should-- and maybe they already are at this point, I can't say that for certain. But if they haven't already started, they should be, I think, really thinking about how we can develop a telehealth model like this moving forward, where it's written into their protocol. Specific guidelines, not just for genetic counselors, but for other team members on Centers of Excellence teams-- that they can sort of refer back to and rely on. Obviously, it goes without saying, we're living in a different world now, and I think that some centers were already doing this even pre-COVID. But for my center, personally, again, we really did not have a super strong precedence really at all" —2*

Genetic Counselors' Attitudes Toward HD-TGC.

When asked about whether they preferred HD-TGC or IPGC, responses varied. Most GCs (n=9) described a preference for IPGC in at least one element of the HD predictive testing protocol. One GC attributed their IPGC preference to a specific patient population:

> *"I think that, if I'm honest, it's the people that are new to Huntington's disease, where it just-- you know, 'Aunt Sue just got diagnosed last week, and I really have to get tested.'-- the people that don't really understand what HD is. Those are the ones I really would like to see in-person because I really want to slow them down and give them a better appreciation for what this is, rather than just-- you know, they're like a train rolling right now. They're going to get tested! They're going to... and then many of them come back later and regret that they did that, so they didn't know what they were doing." —7*

Some GCs explicitly preferred IPGC specifically for test result disclosure sessions (n=3), although one GC felt TGC was well suited for results disclosure sessions due to the often-short duration of those sessions. Some GCs (n=3) wondered if result disclosure sessions were shorter in duration than when they are held in-person. Another GC described that after having experience providing HD-TGC, they no longer felt that test result disclosure sessions should be limited to IPGC. Further, this GC felt that if there was only one session to be performed in-person, they preferred that that in-person session be when the patient consents to testing.

> *"I think that you can do that initial genetic counseling visit with the person by telehealth. And I think you can potentially do the results visit by telehealth. But that visit where you're actually getting the sample—really making that final decision… I think it's important that there's at least one visit in-person. I used to feel very strongly that the results needed to be in-person, but I think they should at least be by a Zoom visit, not just released in your portal like some other tests are." —16*

Unlike some other GCs' institutions, this GC's institution was not able to provide saliva sample collection kits during the pandemic, and thus patients were required to attend clinic in-person for their blood draw. Saliva kits can be shipped directly to the patient, negating the need to come into the clinic.

A little more than half of the GCs (n=9) described the use of protective face masks in IPGC sessions as a barrier to adequate care (e.g., masks hindered identification of emotional cues or development of interpersonal rapport). Half of GCs (n=8) mentioned a preference for TGC if asked to consider the use of masks in IPGC sessions:

> *"I think if I was given a choice between telehealth and a session with a mask, I would prefer the telehealth, just because I think you're missing out on obviously, 50% of someone's face and reading those reactions and the facial expressions, that's been kind of through all or a lot of the counseling is that's one of the downsides of phone counseling, right? You can't see someone's face and the mask kind of does that as well. You can see their eyes, which you can get some things from, but the mask definitely cuts down on what you can see about people's reactions to news or things that you are saying to them." —12*

Perceived Drawbacks of HD-TGC.

Most GCs (n=11) mentioned some discomfort with HD-TGC related to having a restricted view of the patient (i.e., the GCs' view of the patient is limited by resolution and limited to what is in frame). A few GCs (n=3) described support companions talking in sessions while out of frame, unknown to the GCs at the start of the session. Half of GCs interviewed (n=8) mentioned concern of impaired psychosocial connection (i.e., weaker ability to establish interpersonal rapport with the patient, identify emotional cues) in HD-TGC compared to in-person.

> *"It went pretty well… We were able to build rapport, I was able to get a sense for emotional cues, but it was never quite as deep. So, I could still tell how people were feeling overall, but there was still a little more kind of barrier to their emotions. So, not that we couldn't know if they were anxious or upset or sad or whatever they were feeling, but that you just couldn't quite dive into it as much. They would just have a little bit more, I don't know, reservations to opening up." —9*

Half of GCs interviewed (n=8) mentioned patients distracted during TGC sessions, such as those who may attempt to attend the session while driving or in a disruptive or public environment. A quarter of GCs (n=4) mentioned instances of patients answering calls for HD-TGC sessions in a public environment.

> *"I had one time, a patient was sitting outside with people walking by and she was asking me whether or not things are going to be confidential… and I was*

> *like ‘you're sitting in a place that I'm talking to you and people can hear.’ … it was ironic that she was asking how confidential things are going to be.” —1*

A couple of GCs (n=2) mentioned success in use of a preamble ahead of the session to set expectations that the patient should find a private, quiet space where they can concentrate for their upcoming HD-TGC session. One GC clarified that distracted sessions happened in other TGC sessions, not in HD-TGC sessions, wondering if the patients were more mindful of HD sessions than sessions for other conditions.

Many GCs (n=7) described discomfort in losing potential for physical interaction or consolation, i.e., not being able to reach out and touch the patient. A quarter of interviewees (n=4) mentioned the loss of shared physical space as a hindrance on physical assessments:

> “*Yeah, the neuro exam was a joke. … I mean, we certainly tried and got a little bit better over the six months they were doing it, but you couldn't really tell anything. And then the camera would lag, and you wouldn't actually know how to interpret somebody's jerky movements or whatever. So, I'd say that piece was a lot less useful when you're not in-person. You can't even reassure somebody with ‘you don't have symptoms,’ ‘you look great,’ when you really can't see whether they're having symptoms or not.*” *—9*

A couple GCs observed that if support companions were on the call but not in the same location as the patient, those support companions were also unable to offer their physical presence for comfort.

> “*The one thing, though, I didn't like was when a support person couldn't be there in-person with the patient when they were getting their results. So, like, they'll have their support person also on video … which was better than nothing, but it still didn't-- I don't know. I just feel like having someone to hug or hold onto is good when you're getting tough news.*” *—7*

A couple GCs (n=2) mentioned the loss of sharing physical space with colleagues and other members of their care team, perhaps a barrier to providing multidisciplinary care via HD-TGC.

> *“We were missing that multidisciplinary piece where I’m not in the same room for half the day with like the psychiatrist or the social worker. So, I can’t say like ‘I have some concerns. You mind popping in? Do you mind talking to this person just to kind of get another opinion or to have someone else take a whack*

> *at talking about those kinds of things?' So, maybe that was a little bit of a downside. If I wanted to, that would kind of extend the process and I'd have to say like 'Alright, I want you to set up a visit with this other person.' So, maybe I was missing just kind of the team member approach that we usually have in clinic." —13*

> *"It was hard because normally when you're having a bad day as a genetic counselor, your colleagues are right near you and you just pop over to their office and say, 'Oh, this just didn't go the way I wanted,' and you debrief with them or you can tell them about the great things that happened that day. 'Gosh, I had the most amazing patient today and this is what happened.' So it was just hard to do without the support of each other and any guidance." —16*

Perceived Advantages of HD-TGC.

Participants discussed several strengths of HD-TGC, i.e., perceived benefits of HD-TGC to GCs and patients. The most common strength mentioned was the reduced travel burden for the patient (n=15), a statement often accompanied by descriptions of HD-TGC leading to an increase in accessibility of care. GCs often offered examples of distances patients would have had to travel to receive HD genetic counseling in-person pre-pandemic: one patient traveled six hours, another from one coast of the U.S. to the other. Additionally, some GCs discussed how this travel burden can be especially problematic for patients with mobility issues, and for patients who may have a long distance to travel after receiving a distressing test result.

> *"It has to be there for our rural population, for our folks where it is just too hard for them to get into the van, to our clinic, out of the van, into our clinic." —5*

> *"I would like to have the flexibility just depending on the person and seeing what they prefer, especially with a lot of our patients-- They have difficulties kind of with their mobility, and it just can be hard for them to actually even come to an appointment, and I like that we're able to be a little more flexible." —4*

Many GCs felt that HD-TGC allowed for greater flexibility in scheduling patients (n=7), and that HD-TGC saved patients money (n=5), not having to take time off from work or pay for parking at a medical center. A few GCs described the effects of clinical restrictions for in-person care, in which there were limitations on who was physically allowed into the clinic.

> *"That was the other thing that we had to think about… Our hospital restricted-- you couldn't have anyone with you at the visits. So, was it better to bring someone in in-person when we started to see people back without a support person, so their support person's waiting [outside] in the car? Versus doing it all via Zoom where their person might be in their pod and be able to be with them as their partner or spouse or parent? Or is it better to postpone it until things are more back to normal when we don't know how long that's going to last? … We asked for exceptions for our patients in general and not just for Huntington's … that they needed to have a person with them. And they did lift that restriction somewhat, but not in the beginning." —16*

> *"I think I had a lot more people take me up on it because they were able to do the counseling at home or work or something more convenient, and they could have family with them where we were really restrictive in-person if anyone could be in-person." —13*

Most GCs also mentioned that having virtual appointments also allowed support companions to attend sessions with greater ease, such as support companions who may have restrictive work schedules or may live in another region or state (n=9). Many also described the ability of multiple support companions to join virtual sessions as a benefit (n=4); one GC described a session in which six family members were able to join.

> *"That has been really great. We also work with a military population. There's a military base not too far away, and so those people may not have a support person, right? In their, like, local area … they could have just moved to our state, and they want a family member to be their support person… So, that has been a big advantage… is the availability of remote support people who actually, are best situated … that might be the person that my patient talks to most about HD and what the effect would be, and they would either previously come without a support person or just with someone local as a stand-in, so that has been really important." —10*

Most GCs mentioned benefits of seeing a patient in their own home environment (n=10), that being in the comfort of their own home could help to mitigate distress or anxiety (n=5) or could allow for easier childcare management (n=2).

> "*The things that they obviously like about [HD-TGC] is not having to travel; being in a comfortable environment, so that if they do get bad news, they are right by their bed, and they can go cry. You know, that kind of just… 'comfort of home' thing. And... yeah, that the visits don't take up their whole day*." *—7*

> *"I think [restricting HD genetic counseling to in-person] was more again just making sure that in case there was some catastrophic reaction they were at*

> *least in-person, and you could help them. But that's not to say they're not going to have a catastrophic reaction a few hours later. So, I personally think if the patient is comfortable in their own home getting the results-- I think that adds a sense of security to them as well." —15*

Some GCs (n=5) also described advantages of being able to see the patient in the context of their own home, to gain insight to their living conditions and potential struggles:

> "... *You'd get a lot more insight into 'oh, that's why that's a struggle and issue for them, because this is what their bathroom setup looks like.' Or 'this is how they have their pills organized,' or whatever it was" —9*

> *"I honestly kind of preferred the telemedicine model as we've been doing it just because I think it helps provide kind of an insight as well to their homes, the condition that they're kind of living. And especially with Huntington disease, just making sure they're in a safe environment-- especially once they're affected." —15*

GCs Stated Comfort Levels with HD-TGC.

Most GCs (n=11) indicated that although they were initially apprehensive about the use of HD-TGC in place of in-person genetic counseling (IPGC), they grew more comfortable with HD-TGC over time.

> *"Before the first [HD-TGC sessions], I think we were all really nervous. In March, at the very beginning, [HD-TGC] was not something we did and we felt like we didn't do it for a reason and that it wasn't in the patient's best interest. But this kind of an exceptionalism of HD genetic counseling … that [HD genetic counseling] should be one that we continue to do in-person-- So, I think I was surprised, as we started doing it, that it actually did seem to be working pretty well for a lot of things. Maybe not everything, but I think it went better than I expected. But I was definitely nervous as we launched." —9*

> *"I think many of us who have been doing genetic counseling for a long time in-person-- getting used to doing remote counseling was an adjustment. … When I started actually seeing patients, I felt very much like I was a "genetic," not like a "genetic counselor." I was able to provide all the genetic information, I could do the education side of it, but I didn't figure out how to do the psychosocial side of things for a while. That was a learning curve. And now, I'm quite comfortable with it. I have no issues with it-- and I've been doing that, I think, very effectively over the last year. But it was an adjustment, and I think that was the biggest way in which COVID affected my practice. I think learning how to do effective psychosocial counseling was probably the best lesson I have learned from COVID." —8*

Most GCs (n=11) mentioned placing the patients' comfort as top priority, either in practice or as a consideration for future practice.

> *"I personally just think it went a lot better than I anticipated it would. Again, I think patients are a lot more resilient than we might give them credit for sometimes. And I think-- again, if it makes the patient more comfortable and if anything makes the process a little bit easier for my patient, I think I'm willing to do it." —15*

> *"I can see why in the past we would want to restrict [genetic counseling for HD] to in-person, but I feel like at this point, now that video is better-established, I feel like I don't know if such a restriction is necessary… and it really should be kind of done on the basis of the patient and what they need and what they want." —4*

Some GCs (n=5) described feeling uncomfortable with providing HD-TGC. These GCs explicitly attributed part of their discomfort to a lack of control over the session, control that they felt they may have otherwise maintained in-person. One GC described this as feeling like they were no longer able to use the educational visual aids that they would usually use in-person. Another GC described feeling uncomfortable if individuals were experiencing anosognosia (i.e., patients unaware or denying that they were showing symptoms of HD).

Lasting Potential of HD-TGC Beyond COVID-19.

When asked about HD-TGC's lasting potential beyond the COVID-19 pandemic, the majority of GCs (n=10) described a hope to continue to provide HD-TGC services as an option beyond COVID-19 restrictions and concerns.

> *"This has been a really big opportunity for us to expand access to care and for that, I can be thankful. If nothing else with COVID, there was the opportunity for us to really expand care, and I do not see a future of care for HD that does not include virtual visits or telehealth. … I think it's an invaluable resource that we can offer to our patients, so I cannot see a future where we don't have that anymore." —5*

One GC described feeling that TGC adoption was inevitable, despite the GC's preference for IPGC. Another GC mentioned feeling as if their comfort with future use of HD-TGC hinges on whether guidelines are updated:

> *“I think our virtual visits have worked as well as in-person ones. I’m a rule follower. So, just making sure we’re following the rules… I would have no qualms doing it more routinely if guidelines were to change.” —11*

Many GCs (n=9) spoke of continuing to offer HD-TGC as a service option alongside IPGC, suggesting the development of a hybrid solution that would integrate both IPGC and HD-TGC. For some, that meant providing HD-TGC for some portions of the HD GC protocol and IPGC for remaining portions. For others, a hybrid approach could be offered by having a work schedule in which they work some days in-person in the clinic and other days via TGC from home, allowing availability of both options to patients.

> *“I'm just going to reiterate that it is not like either/or — Telemedicine or in-person. It is your delivering a service to a patient, and you're integrating technology as you need it. That's what it is. And you’ve got to be flexible. Use it where it's appropriate. Manage it in terms of teeing it up, setting it up, etcetera. Manage it appropriately. And you can extend your service to patients and get away from this big barrier of having to come into the health system. Our practice, we want to use a hybrid. So, we'll do a video visit, but we'd also like to lay eyes on you. And we definitely want to do that return of results in-person. Unless there's some extenuating circumstance, I think it's better in-person.” —3*

Perceived Barriers to Providing HD-TGC.

GCs described logistical barriers to HD-TGC’s success. These complicating factors included technological limitations and concern that restrictive billing and reimbursement policies for TGC services may resurface after the pandemic.

Technological Barriers.

Various technological barriers arose in interviews. Most GCs (n=9) mentioned poor internet connection as a barrier to the success of HD-TGC. Many GCs also described limited technological literacy (n=7) and limited access to adequate technological necessities (n=6) as challenges.

> “*It’s just the technology part of it. Like, if their Wi-Fi connection wasn’t great or they had trouble connecting. Sometimes it took five or ten minutes for us to actually get the appointment started. So, they were annoyed by that… but they*

> *said ‘it’s still less time than if we had to come in.’ So, they were okay with it overall. … The only complaints were technology-related.*” *—11*

Many GCs (n=7) mentioned age in relation to comfort with technology, stating that younger patients may have been more comfortable with HD-TGC. Some GCs (n=3) mentioned concern for patients who may have been experiencing cognitive impairment from HD and how effects on their executive function may have contributed to their difficulty using technology.

Insurance / Billing & Reimbursement.

Many GCs (n=6) felt as if the potential for HD-TGC to be maintained as an option depended on whether billing and reimbursement would continue to allow TGC as an option or revert to being as restrictive and complicated as it was pre-COVID.

> *“I hope that insurance payers and people kind of learn because I think a lot of other industries, certainly commercial industries and other businesses have learned and have adapted and are kind of doing things in a different way because of COVID-- it seems like a waste to just go back to what we knew before instead of adapting like the rest of the world.” —12*

This GC further described no longer having the ability to provide HD-TGC services due to billing regulations. This GC also spoke of institutional pressure to see more patients because of billing concerns during the pandemic:

> *“Our neurology department had a huge push to meet or exceed the number of visits that we were going to have in-person, but to do them virtually. Because they couldn't charge things like facility fees and stuff at the hospital, and the reimbursement wasn't as high as someone coming in physically to the department-- they wanted us to be seeing more patients.” —12*

Perceived Facilitators of HD-TGC.

GCs described facilitators of HD-TGC’s success. Examples include having pre-pandemic familiarity with telehealth platforms or practices and having adequate institutional support to avoid potential barriers to the provision of TGC services.

Familiarity with Technology.

Many participants (n=10) described prior familiarity with telehealth tools as a major facilitator for HD-TGC's success and potential to last beyond the COVID-19 pandemic. A quarter of GCs interviewed (n=4) attributed pre-COVID familiarity with recreational video chat platforms (e.g., Facetime, Skype) as a potential contributor to their comfort with HD-TGC.

> *"I think people really caught on to telehealth for genetic counseling and really liked it — and it works well for most of the sessions. So, I was able to adopt that pretty quickly — especially because I already had so much experience with WebEx and doing the return visits that way. I already knew how to do it, whereas a lot of the people that I work with had no idea what they were doing." —13*

Institutional Support.

One GC mentioned that the loosening of billing restrictions provided the freedom their institution needed to initiate utilization of telehealth. Despite having interest in telehealth in the past, billing restrictions held the institution back from trying. Dedicated institutional support ensured a smooth transition:

> *"We were not doing a lot of telehealth prior. It was actually very difficult. We did have a couple of doctors that were interested in it. The number of hoops that they had to jump through with insurance and payment and billing to do telehealth made it next to impossible for that to be successful. Once COVID happened, all of those hurdles were flattened by the government, and the leadership at [my institution] deserves all of the credit for getting us up and running that quickly. They met I think 24/7 until they had it up and running. They had enough knowledge that they just jumped right in and made it work. They were very dedicated to making it work." —17*

GCs Preferences for HD-TGC Platforms.

Video Platforms.

In order to communicate remotely, GCs utilized a wide variety of video communication platforms. Most GCs (n=11) mentioned using Zoom for video conferencing with patients. The next most common video platform mentioned was when GCs used an extension embedded in their institution's electronic medical record system (n=7). A few GCs explicitly mentioned using Amwell (n=3) and a couple GCs mentioned using WebEx (n=2). CareConnect, Doximity, Facetime, GoogleDuo, Microsoft Teams, Skype, and VidyoConnect were mentioned explicitly by one GC each.

Video Platforms vs. Telephone.

Half of GCs interviewed explicitly mentioned a preference for video chat over phone counseling (n=8). Often, reasoning for this preference was that video allowed for assessment of facial expressions and body language (n=5). One GC mentioned that patients seemed to be more prone to distraction in phone sessions:

> "*I think I've noticed that when people are on the phone, you can definitely hear a lot more of them like making dinner or doing things that are distracting them." —13*
>
> *"So, I think the biggest difference for me is phone versus [video]. Phone, I got squat. … I did have a hard time establishing, I think, a good rapport with patients over the phone, which is why we very quickly stopped doing that." —5*

HD Exceptionalism.

HD exceptionalism refers to the tendency to treat HD genetic counseling as different than genetic counseling for other conditions. Historically, the severe implications of a risk or diagnosis of HD have been associated with a heightened sense of caution when counseling at-risk individuals seeking HD genetic testing, particularly predictive testing in the presymptomatic population. The phenomenon of HD exceptionalism (i.e., care for HD approached with more caution than other conditions) was a theme of these interviews and primarily focused upon the highly sensitive nature of predictive test results disclosure sessions. Almost half (n=7) of GCs discussed the importance of psychosocial considerations to HD genetic counseling (e.g., *"I think HD is the most psychosocial of all the genetic counseling that I do.*"—1). Half (n=8) more specifically pointed to concern of suicidal risk following test results disclosure. One GC mentioned concern about kitchen knives stored in patients' homes and how those can serve as easily accessible weapons for self-harm. Two GCs (n=2) challenged the conventional reasoning that having predictive test results disclosure sessions in-person is safer than TGC for patients:

> *"I think obviously the preferred method would always be in-person, just in the rare case there would be something dramatic or drastic. Hopefully you prepare the patient for that. I think even if you get results in-person, that patient is not — you're not going to hang out with them all day to make sure that they're okay. The visit is going to end and they're going to go home and start to process those results. I think it just kind of highlights [that] it is important to keep checking in with people and make sure they're doing okay, or [check] if there's any other needs that they have." —13*

> *"I think, as providers, we're the ones that have to get over ourselves, and trust that our patients can manage their own emotional wellbeing in their home — assuming we've done sort of a clinical assessment, where they're not emotionally fragile, or anything that's crazy is going on for them." —7*

A few GCs wondered if the restrictions of the HD GC protocol were not just in consideration of the patients and families, but also for the comfort of the GC providing care:

> *"I think we are more spun up around the mode of how we convey that message to the patient than the patients are. … I think that genetic counselors as a body are just so spun up about how we deliver the message, and the patients could care less." —14*

> *"With HD it's different and not different … That's one of the things that I've learned over time. We always talk about genetic exceptionalism, and should genetics be treated differently [from] other types of medicine, and I think more and more people say no. But there's still a little bit of this HD exceptionalism and that we have to do all these extra things for people at risk for HD that we don't do for other patients. And I think the more I work with the HD community, the more I wonder if that's something that we're doing for them, or if it's something we're doing for us and for our own comfort. … The early literature is populated with lots of concerns about suicidality and suicide attempts in the time waiting for testing and immediately upon learning results … I think the newer data is not bearing that out — people are better prepared. I think there's been some ascertainment bias about poor outcomes in the literature that made us nervous. We've never had an instance in our in-person clinics where we felt like we needed to walk someone to the emergency room for Huntington's disease learning their results." —10*

No instances of severe adverse reactions such as suicide or self-harm were mentioned in interviews. Notably, psychological concerns are not isolated to individuals who receive a positive test result; one GC mentioned that the most reactive patient she had was an individual who tested negative and experienced "survivor's guilt."

Another facet of HD exceptionalism included the higher time and travel burden of genetic counseling for HD, due to there often being more appointments for HD than for other conditions (n=5). Also, GCs admitted having a more attentive approach to care for HD (n=5) than other conditions. Many GCs (n=5) discussed that generally, individuals seeking HD genetic counseling often already have some background knowledge of the genetics and inheritance of HD, compared to individuals seeking genetic counseling for other conditions. A quarter of GCs (n=4) spoke of the near certainty involved with predictive testing for HD, due to the causal genetic variant having full penetrance if inherited. Some GCs (n=3) mentioned challenges to HD genetic counseling presented by the 21st Century Cures Act, in which patients' health records are required to be accessible to the patient "without delay," and thus genetic test results could be released to the patient's electronic medical record before a GC has been able to counsel their patient.

Discussion of Interview Findings

Overall, GCs were mostly comfortable with HD-TGC and felt it should continue to be offered as an option for the HD community, especially when it involved a video interface. HD-TGC offered an alternative to IPGC when IPGC was unavailable, and some GCs have continued to offer HD-TGC as an option even as restrictions were lifted and their clinics returned to in-person care. Most GCs who provided HD-TGC praised it for reducing burdens of attending in-person appointments that typically fall upon patients. Patients who lived in rural or distant areas who may have had to drive hours to attend an in-clinic appointment were now able to do so without having to travel, and some may have saved money by not having to take time off from work or pay for gas or parking. This confirms well-established benefits of telehealth noted in prior literature and is significant because it provides evidence that these benefits may apply in the challenging context of HD counseling specifically (Buchanan et al., 2015; Danylchuk et al., 2021; Houston et al., 2021; Hawkins et al., 2013; Menachemi et al., 2004; Solomons et al., 2018; Terry et al., 2019). For symptomatic patients with mobility issues, they and their caretakers no longer had to undergo the potentially painstaking process of traveling into the clinic. HD-TGC also offered an opportunity for multiple or preferred support companions to attend a session, even if they lived far from the patient.

GCs perceived patients' responses to HD-TGC to be largely positive and felt that patients seemed comfortable and satisfied with HD-TGC services overall. This perception is consistent with the more rigorous published evaluations of patients' attitudes to telehealth which have consistently reported that patient satisfaction is high (Breen et al., 2021; Brown et al., 2021; Danylchuk et al., 2021; Dratch et al., 2021; Hawkins et al., 2013; Houston et al. 2021; Zoran et al., 2021).

Despite largely positive assessments of HD-TGC, many GCs still preferred IPGC for some — if not all — of HD genetic counseling. This preference was informed by observations of some drawbacks of HD-TGC. Specifically, most GCs reported some discomfort with the fact that they had a restricted view of the patient when using HD-TGC, and about half of the GCs interviewed mentioned concerns about the quality of their emotional connection with patients using HD-TGC or worried that they would not be able to provide physical expressions of comfort to patients remotely. Mueller et al. (2021) recently interviewed GCs to gain insight into their experience providing TGC during COVID-19 and observed a similar split in their opinions of whether TGC impaired rapport building with patients. More research is needed to investigate patient-provider rapport building in TGC sessions.

Half of the GCs in our sample mentioned that patients seemed distracted during HD-TGC sessions, and one GC referred to physical HD neurological exams over telehealth during the pandemic as "*a joke*." This suggests that genetic counselors may need to be trained to contract with patients at the start of a virtual session about managing distractions and interruptions. Furthermore, although Bull et al. (2014) reported that motor assessment via telehealth was valid and feasible, the fact that this is not a universal view suggests that future studies should continue to compare the effectiveness of virtual neurological exams with those conducted in-person.

Taken together, these concerns about HD-TGC echo the concerns expressed by genetic counselors in other practice settings. In one recent, large survey of general practice GCs, GCs reported difficulty conducting physical examinations, challenges with translating services for non-English speaking patients, lack of visual cues, and difficulty with rapport building as some of the main challenges of the transition to telemedicine during the COVID-19 pandemic (Ma et al. 2021). Given the ethically fraught history of HD genetic counseling and the recommendations for in-person sessions in the established HD testing protocol, these types of concerns warrant closer attention and follow-up research if HD-TGC is to continue as a mainstream practice.

Two nuanced findings of these interviews have not been discussed in the literature previously. The first is that preferences for IPGC mostly reversed to a preference for HD-TGC when GCs were asked to consider the use of protective face masks for in-person HD genetic counseling sessions. Many GCs felt that the loss of visible facial expressions outweighed the loss of shared physical space with the patient, and some mentioned the loss of potential physical acts of consolation, such as offering a hand on the patients' shoulder after receiving distressing news. The second is that some GCs also observed that the patient's support companion wasn't always able to accompany the patient in HD-TGC sessions and therefore the support companion was also unavailable for physical consolation. Given the heightened concern for patients' emotional state in HD genetic counseling sessions — if HD-TGC does indeed continue to be offered as an option for the HD community — it may be especially important to adhere to the recommendation that patients bring in-person companions with them to their HD-TGC visits.

Many GCs in this sample anticipated that HD-TGC will continue to be offered alongside IPGC, suggesting the development of a hybrid solution that would integrate both IPGC and HD-TGC. This is aligned with findings of Dratch et al. (2021), in which neurogenetics patients described preference for a hybrid model combining in-person and telehealth for future clinical care. Openness to hybrid in-person and HD-TGC models of care opens the door to new possibilities that should be evaluated in future research. For example, if a patient's primary support companion is not able to physically present for an appointment, in the future, perhaps the primary support companion can still join the HD-TGC session virtually while the patient is physically accompanied by an additional support companion.

GCs most often expressed a preference for in-person genetic counseling for predictive test results disclosure sessions. Psychosocial assessments are very important in HD genetic counseling, especially in assessing an at-risk individual's psychological and emotional preparedness to undergo predictive genetic testing. However, the insinuation that IPGC is always superior to HD-TGC in mitigating or preventing psychological distress is not supported by our findings or the broader telemedicine literature (Bradbury et al. 2018; Brown et al. 2021; Buchanan et al. 2015; Danylchuk et al. 2021; Gorrie et al. 2021; Hawkins et al. 2013; Hilgart et al. 2012; Interrante et al. 2017; Solomons et al. 2018).

Many GCs reported feeling a relative "lack of control" over HD-TGC sessions, which abated over time with experience. While it is true that sharing physical space with the patient in a medical center does provide close access to resources such as emergency care, the perceived control that GCs reported feeling during in-person genetic counseling sessions is only temporary. As a few GCs in this sample mentioned, GCs will not be by a patient's side 24/7 after patients receive their results.

Some GCs described frustration with the lack of relevant guidelines when navigating the transition to TGC. This is rational given that the COVID-19 pandemic was an unprecedented global emergency. While operating without guidelines can be problematic, the lack of guidelines *did* allow GCs to bend their conventional practices which resulted in an opportunity to try TGC when they may not have done so otherwise. Future research and scholarly work should attempt to identify best practices for providing genetic counseling services virtually so that future generations of GCs will have more guidance in the future.

There were two main barriers to HD-TGC implementation described by GCs in this sample. First, some GCs noted that providers or patients with limited access to or prowess with technology were less likely to reap the benefits of HD-TGC. This has been characterized as a “digital divide” (Larson et al., 2021; Norris, 2012). While clinics cannot control the quality of internet available from the patients’ end, they *can* invest in making sure that their clinic has all the resources needed to ensure clear, interruption-free communication. A few GCs mentioned simplifying the process for patients, such as only requiring the patient to click on one link to begin their TGC session. Also, sending patients informative materials ahead of time to minimize technical difficulties and set expectations with TGC sessions could be of great benefit to the patient and GC. In HD-TGC specifically, technological concerns are of great importance, since HD patients may have impaired cognition which may affect their ability to navigate technology. Future research should comparatively evaluate the design features of different HD-TGC platforms to determine the impacts different features have on access and usability in different populations.

Second, many GCs worried that billing and reimbursement restrictions that were loosened to allow easier provision of TGC services might revert to their more complicated and exclusionary nature pre-pandemic. These concerns reflect findings in the TGC literature and suggest that the question of whether HD-TGC will continue to be offered as an option beyond the COVID-19 pandemic does not rest on whether GCs are comfortable or satisfied with HD-TGC but rather, whether pre-pandemic billing and reimbursement restrictions will be reinstated post-pandemic (Bergstrom et al, 2021; Boothe et al., 2021; MacFarlane et al, 2021; Menachemi et al., 2004; Mills et al., 2021; Terry et al., 2019; Zierhut et. al, 2017). Because the lack of telehealth services in the past has been at least partially attributed to such restrictions, a return to any semblance of the pre-pandemic status quo could impede future utilization of HD-TGC.

Chapter 5: Overall Discussion of Study Findings

If there is a silver lining to the COVID-19 pandemic, it could be that it provided an unexpected opportunity to expand the boundaries of healthcare and really explore telehealth as an option — to learn what worked, what didn't work, and what can be done to improve HD-TGC going forward. Genetic counseling for HD is often used as a model of the most extreme and complex challenges in counseling patients and families of the implications of severe adult-onset, unactionable conditions. Finding successes in the implementation of HD-TGC is encouraging for the future of TGC.

Relating to Everett Rogers' Diffusion of Innovation Theory (DOI), governmental recommendations to utilize telehealth — alongside eased telehealth restrictions — provided the initial authority innovation-decisions necessary to allow for a fast adoption of HD-TGC. Authority innovation-decisions categorically have the fastest rate of innovation-adoption. The rate of adoption is also increased when the amount of time between awareness of the innovation and the decision of whether to adopt it is decreased (Rogers, 2010). COVID-19 restrictions gave urgency to the innovation-decision of whether to implement HD-TGC. That urgency compressed the time between initial awareness of HD-TGC and the decision of whether to implement it into practice, leading to a higher likelihood of HD-TGC adoption than pre-pandemic.

GCs' assessments of HD-TGC also seem to have successfully touched upon the five main attributes of an innovation that contribute to an innovation's successful diffusion and adoption: [1] *relative advantage* over alternatives; [2] *compatibility*, such as with existing values and beliefs; [3] *complexity* or ease of use; [4] *trialablity*, or ability to experiment with/try an innovation on a limited basis; and [5] *observability of results* from the innovation's use (Rogers, 2010). Both the survey and interviews illustrated aspects of HD-TGC that GCs felt provided benefit to patients and counselors, suggesting the presence of DOI structures of *relative advantage* of HD-TGC and covering the need for [5] an *observability of results.* Providing HD-

TGC when the alternative was not providing care at all demonstrates a relative advantage — simply, patients continued to receive care. Compared to in-person genetic counseling, HD-TGC did also demonstrate other relative advantages, such as increased access for some patients and their support companions. Assessing [2] *compatibility*, most GCs responded that they were comfortable with HD-TGC and that their comfort grew over time. Finally, the tone of these interviews was in assessment of HD-TGC, a service alternative that for most was not offered before the temporary restrictions placed on in-person care due to COVID-19. As such, GCs spoke a good deal to the malleable nature of HD-TGC during this time, reflecting its [4] *trialability*.

Implications

Despite initial discomfort and uncertainty before implementing HD-TGC services into practice, most GCs were comfortable and satisfied with HD-TGC services and felt that HD-TGC would and should continue to be offered as an option beyond the COVID-19 pandemic. Given the often-perceived exceptional nature of HD genetic counseling, this evidence suggests that TGC is a viable option for the HD community and that some protocol restrictions recommended for HD genetic counseling should be revisited. Said plainly, HD genetic counseling does not have to be restricted to in-person clinic visits. A number of GCs were even comfortable with HD-TGC for predictive test results disclosures, an element of the protocol often touted as deserving highly attentive care. This evidence suggests that HD-TGC has been safely provided to multiple patients across the U.S. and could continue to be offered as an option for the HD community.

GCs reported that patients who received HD-TGC also seemed to be satisfied and comfortable with HD-TGC services. HD is relatively rare and the number of HDSA Centers of Excellence across the U.S. are limited (currently ~54). For an individual to receive HD-specialized care, they may have to travel considerably long distances. This can be a substantial time burden on the patient and potentially also their caregiver or support person. This can be especially burdensome for patients who may have mobility issues or those who may have just received distressing information. HD-TGC also allowed for greater ease for multiple and distant support persons to join the patient for sessions. Satisfactory service, reduced burdens, and increased access for patients and their support system illustrate HD-TGC as a success in patient-centered care and suggest that the HD community would like for HD-TGC to continue to be offered as an option.

Stuttgen et al. (2018) recently interviewed individuals who were enrolled in the first presymptomatic genetic testing protocol (1986-1998) for HD through Johns Hopkins. Although most interviewees saw value in a formal protocol for HD testing, some believed individuals should be able to decide for themselves whether to follow it or not. Indeed, an at-risk individual who wishes to eschew the formal testing process could find a physician to order the predictive test for them without any counseling of the myriad implications of predictive testing for HD. However, in this study, while most interviewees thought that a direct-to-consumer genetic test for HD would be inappropriate, some felt it should be allowed if the test could be cheaper or available for patients who wouldn’t otherwise be able to learn their status. These findings highlight the importance of autonomy, access, and education in the HD testing process, suggesting that the increase in flexibility and accessibility that HD-TGC can provide could benefit members of the HD community.

Despite concerns that psychosocial assessments and emotional connections with patients may be impaired in HD-TGC sessions, most GCs preferred HD-TGC over in-person genetic counseling if in-person genetic counseling required the use of masks and social distancing. Institutions and providers may want to consider how masks may impair psychosocial care and continue to offer HD-TGC while masks are required or recommended.

Most GCs still preferred IPGC for at least some of the HD GC process, and most favored maintaining IPGC restrictions for predictive test results disclosure sessions. However, a few questioned the presumptive reasoning that IPGC is superior to HD-TGC in mitigating or preventing severe distress. They suggested that patients have more resilience than they are often credited, and that GCs cannot control their patients — the patients will eventually leave the clinic after the appointment. Additionally, no GCs reported a reaction of extreme distress or suicidal ideation or behavior from a patient that warranted emergency care. What may warrant closer attention is the finding that not all patients had a support person with them for HD-TGC visits. Perhaps making a strong recommendation that someone from the patient's support system sit with them in shared physical space during results disclosure sessions is a potential solution to ease concerns of the patient's safety after ending an HD-TGC session.

Additionally, some GCs mentioned favoring a hybrid approach to HD genetic counseling. For some a hybrid approach would involve a mix of IPGC and HD-TGC for the patient for different elements of the HD genetic counseling protocol, while for others a hybrid approach meant that they hoped to work some days in clinic and some days remotely, to offer opportunities for whatever the patient preferred. The latter is less restrictive and allows for patients' preferences and values to drive decisions regarding their clinical care. This is another example of an opportunity to exercise patient-centered care, a tenet of genetic counseling. Even if GCs prefer IPGC and/or don't currently provide TGC, they should continue to familiarize themselves with best practices of TGC in case patients prefer TGC.

HD-TGC increases access for some but should be treated as an option, not as a potential replacement for IPGC. Not everyone is comfortable with TGC and more broadly, not everyone is comfortable or familiar with technology. TGC is inappropriate for anyone who struggles with technology, lacks access to necessary resources, or is experiencing cognitive dysfunction and in need of direct observation, assessment, and care.

Some GCs worried that systemic issues such as billing/reimbursement and institutional support warranted concern for HD-TGC's lasting potential beyond the pandemic. Perhaps federal, state, and institutional restrictions will ease as more research demonstrates TGC's potential for success and improved care for some patients. However, patients and providers may need to advocate for the regulatory changes needed to continue to allow TGC services.

Limitations

This study was limited by reliance on GCs' recall of attitudes and practices related to TH prior to the pandemic. Thus, this study's findings are susceptible to recall bias. It is possible that GCs' recollections are inaccurate and/or incomplete. While the interviews sought to gain a more complete picture of GCs' experiences with HD-TGC, not every HD-TGC GC was interviewed. Conversations were limited in time and scope, and the findings from these interviews are not statistically generalizable. Additionally, recalled patient feedback from the GC is a perception of the GC, not a direct account from the patient. The patients' perspective in this study is secondhand and therefore not any patient's true perspective. Further, accounts of the frequency of HD-TGC services and distinction between modalities are also subjective to the GC's recollection.

This study employed novel, unvalidated instruments for the survey and interviews. While the instruments were informed by existing literature and knowledge of HD and TGC, the questions assessed HD-TGC specifically, were developed by the research team, and were not modified from any existing validated questionnaires or interview guides about HD, TH, or TGC. Additional research should be done to validate findings of this study.

Finally, HD is rare and therefore the available population of HD GCs is considerably small. Because not every HD GC participated in this study, the sample is further limited to a self-selected pool of participants. Only some HD GCs participated, and so not all experiences with HD-TGC could be described. For example, only one GC indicated that they practiced in a rural setting. Most participants of this study served as a GC for an HDSA Center of Excellence, so the findings may not be generalizable to all HD GCs. However, HDSA Centers of Excellence undertake an annual application review process to ensure they are providing top quality care for

HD patients. It can be inferred that GCs at HDSA Centers of Excellence are particularly interested and experienced in HD care and provide an informative sample for insights into HD genetic counseling. Future expansion of this type of study to reach more HD GCs beyond those who serve HDSA Centers of Excellence will be needed to evaluate the state of HD genetic counseling practice more broadly.

Future Directions

Future studies should assess patients' responses directly from the HD community. Comparing patient outcomes between matched pairs of those who received IPGC and HD-TGC, longitudinal assessments, case studies, and in-depth interviews could be particularly informative. If studies could reach patients who decline the option of HD-TGC to investigate their reasoning, that could uncover or elucidate barriers to HD-TGC's success in the HD community. Perhaps certain demographics or characteristics may influence a patient's assessment of HD-TGC.

Additionally, given the importance of psychosocial assessment in HD genetic counseling, future studies investigating best practices for assessing risk of psychological distress in TGC sessions would be useful. While most GCs who provided HD-TGC services felt confident in their ability to assess patients' suicidal ideation or risk through HD-TGC, some GCs were not. Evidence of successful interventions or precautions could help ensure patients' safety and comfort providers. Future studies could retrospectively investigate adverse HD patient reactions that took place in IPGC and TGC environments to better characterize potential catalysts or predictors of psychological distress and assess how these situations may be handled by the GC.

Future research could also further investigate virtual neurological assessments in HD, and more broadly how clinics may have accomplished multidisciplinary HD care virtually. An undeniable benefit of IPGC for providers is not only the ability to perform physical assessments when necessary but also that often, colleagues of multidisciplinary teams are physically nearby and available for quick consults, last-minute additions, and social camaraderie.

Conclusion

Time will tell whether HD-TGC will continue to be offered as an option beyond the COVID-19 pandemic. However, findings from this study are encouraging. Despite initial trepidation, most GCs reported feeling comfortable with HD-TGC and believe it would and should continue to be offered as an option for the HD community. GCs' recollections of patient feedback suggest that most patients were also comfortable and satisfied with HD-TGC services. Benefits, drawbacks, facilitators, and barriers of HD-TGC largely mirror those found in the telehealth and telegenetic counseling literature, but many GCs continue to feel preference for maintaining some in-person aspect to the genetic counseling process for HD predictive genetic testing, usually for results disclosure sessions. It is unclear if this preference is a residual caution from conventional practice or if it is indeed to the patient's benefit, as there is currently no evidence for the superiority of in-person genetic counseling in the mitigation or prevention of potential psychological distress in HD genetic counseling sessions. Future studies should investigate this further, as well as the perspectives and preferences of the HD community firsthand.

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Appendices

Appendix I: Recruitment E-Flyer

Do you have **18+ months** experience providing **genetic counseling** for the **Huntington's disease** community?

We are looking for genetic counselors to share about their experiences providing genetic counseling services for the Huntington's disease (HD) community before & after onset of the COVID-19 pandemic.

All participants will complete an online survey about these experiences. Some participants may be contacted for a follow-up interview.

You may participate if you:

- Are a board-certified genetic counselor
- Provide genetic counseling services in the United States
- Have provided genetic counseling services for the HD community for 18 months or more

If you are interested in participating, please follow this link to a 15-20 minute electronic survey. Survey participants will receive a $25 Amazon gift card upon completion.

Select survey participants will be contacted for a 30-45 minute interview. Interview participants will receive an additional $50 Amazon gift card upon completion.

If you have questions, you may email the research team:

Wes Solem, BS
wsolem1@jhu.edu

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Appendix II: Consent Language

Thank you for your interest in participating in this study!

For more information, please read below.

If you agree to these conditions, please click the checkbox next to “Yes, I agree” at the bottom of the page.

What you should know about this study

You are being asked to participate in a research study. This consent form explains the research study and your part in the study. Please read it carefully and take as much time as you need. You are a volunteer. You may choose not to take part at all. If you join, you may skip questions, and you may quit at any time. If you skip questions, you will still receive compensation. If you decide to quit the study, you will not receive compensation.

Purpose of research project

This research is being done to understand how COVID-19 clinical restrictions have affected genetic counseling for the Huntington’s disease community, particularly the utilization of tele-genetic counseling via phone or video conferencing.

Why we are asking you to participate

You are being asked to participate because you are a genetic counselor that has provided genetic counseling for Huntington’s disease. We are interested in hearing from between 20-100 genetic counselors. You can take part in this study if you are a board certified genetic counselor in the United States who has provided more than 18 months of professional genetic counseling for the Huntington’s disease community, and if you are English-speaking.

Study procedures

You will be asked to complete a 15-20 minute survey. If you are interested in further participation, you may be contacted for a follow-up interview. If you are selected, you will be contacted to take part in a 30-45 minute interview in which the audio will be recorded, transcribed, and analyzed for emergent themes. The interview will ask about the implementation of tele-genetic counseling in your clinic (if applicable). The interview is optional and your participation in the interview is voluntary. You do not have to participate in the interview if you decided you do not want to.

At the end of this survey, you will be asked for your contact information. Your contact information will be kept confidential, and will only be kept for the duration of data collection. Identifiers will be destroyed after data collection is complete.

Your information will only be used for the following reasons:
To email your e-gift card compensation(s)
To clarify any survey responses, if necessary
To facilitate communication for a potential follow-up interview
To inform interview design, participant selection, and recruitment

Risks/Discomforts

There are no physical harms or risks anticipated with participation in this research. However, if the participant is interested in participation in a follow-up interview, their contact information (name, phone, email) will be collected and held in order to facilitate coordination of contact for the interview. Demographics collected will not include explicit mention of the participant's employer, but will include identifiable information such as city, state, and description of clinical setting. Although all of this information will be confidential, securely stored, and de-identified, there is the possibility that the security of this information may be breached.

Additionally, there is potential for the genetic counselor's reflection to conjure memories of a distressing session or detail related to tele-genetic counseling, Huntington's disease, or the implementation of telehealth more broadly. Participants are free to exit the study whenever they choose. The researcher's direct contact information is provided below, should any concerns arise. If at any point you feel upset or distressed in completing the survey or participating in the interview, you can stop participation at any point or skip questions you do not wish to answer.

Benefits

There will be no direct benefit to participants for participating in this study. However, the data collected for this research could be clinically informative to the field of genetic counseling, the Huntington's disease community, and the growing inquiry of the utility of telemedicine.

Payment

You will receive a $25 gift card for completing the survey. An additional $50 gift card will be given to participants who are interested in, selected for, and complete a subsequent interview. If you choose to exit the survey or interview early, you will not receive the associated compensation.

Data sharing and confidentiality

Any personal information provided to this research will be stored in a confidential and secure manner. A file of your demographic information shared with this study will be kept in a password-secured location. If you participate in an interview, once that interview is complete and transcribed, any personal, identifiable information will be redacted. Once interviews are transcribed and coded, audio files will be deleted. When we report results from this research, no identifiable information from individual participants will be included. If you mention any specific names or other personally identifiable information during your interview, that information will be redacted.

Protecting your privacy during data collection

Your survey will be completed using a service called Qualtrics. Interviews will be conducted over Zoom, with video off. This will allow the researcher to record the interview on an encrypted laptop. Information collected from the surveys will be saved to this encrypted laptop. Information collected from interviews will be saved to a password-protected file. If you are concerned about privacy, please make sure you are in a comfortable, private location during your participation.

What happens if you leave the study early?

If completing the survey or interview causes you any distress, you can skip questions you do not wish to answer or stop the interview at any point. If you choose to end the interview early, you will be asked whether or not you would like the parts of the interview you provided to be included in the study.

Who do I call if I have questions or problems?

If you have questions, concerns, or complaints, you may call or email either of the senior investigators:
Debra Roter, DrPH 410-446-5795 • droter1@jhu.edu
Leila Jamal, ScM, PhD, CGC 347-327-0104 • leila.jamal@nih.gov

You may also call or email the student investigator:
Wes Solem 214-235-7761 • wes.solem@nih.gov

Call or contact the Johns Hopkins Bloomberg School of Public Health IRB Office if you have questions about your rights as a participant.

Contact the IRB if you feel you have not been treated fairly or if you have other concerns.

The IRB contact information is:
Johns Hopkins Bloomberg School of Public Health
615 N. Wolfe Street, Suite E1100, Baltimore, MD 21205
Telephone: 410-955-3193 • Toll Free: 1-888-262-3242
E-mail: jhsph.irboffice@jhu.edu

What does your clicking “Yes, I agree” on this consent form mean?

Clicking “Yes, I agree” on this form means: You have been informed about this study’s purpose, procedures, possible benefits and risks. You have been given the chance to ask questions before you sign. You have voluntarily agreed to be in this study.

Appendix III: Survey Instrument for Participant with HD-TGC + In-Person HD GC Experience

Have you provided genetic counseling for the Huntington's disease community for 18 months or more?

☑ Yes ☑ No

If no →

"Thank you for your interest in participating in this study. Unfortunately, you do not meet eligibility requirements to participate. Please exit the survey."

If yes → [continue]

-- [Online consent script here; See **APPENDIX C**] --

Demographics:

As a genetic counselor, my primary specialty would be best described as:

☑ Adult/Neuro ☑ Adult/General ☑ Cancer ☑ Preconception/Reproductive
☑ Prenatal ☑ Pediatrics ☑ Cardio ☑ Other

Years as a practicing genetic counselor:

☑18 mos - 2 yr ☑2 - 5 yr ☑5 - 10 yr ☑>10 yrs

Years providing genetic counseling to HD community:

☑18 mos - 2 yr ☑2 - 5 yr ☑5 - 10 yr ☑>10 yrs

Are you the primary or head HD genetic counselor in your practice?

☑ Yes ☑ No

How many genetic counselors in your practice, other than yourself, provide HD genetic counseling? __________________

How would you describe your current involvement with HD in your genetic counseling practice?

☑ Extensive ☑ Common ☑ Intermittent ☑ Not at all

I would describe my past involvement (2019 or earlier) with HD in my genetic counseling practice as:

☑ Extensive ☑ Common ☑ Intermittent ☑ Not at all

City & state of practice: City: ____________________ State: [drop down menu**]**

Do you serve as a HD genetic counselor for an HDSA Center of Excellence?

☑ Yes ☑ No

Before the COVID-19 pandemic, how would you describe the frequency with which presymptomatic/predictive genetic testing **results disclosure** sessions took place in-person?

☑ Always ☑ Often ☑ Inconsistent ☑ Not often ☑ Never

Have you provided any tele-genetic counseling (e.g., by phone, video) for the Huntington's disease community during or since 2020?

☑ Yes ☑ No

If no → See **APPENDIX D**

If yes → [continue]

The following questions ask about genetic counseling services you provided in 2019, compared to 2020-2021 when the onset of the COVID-19 pandemic affected many practices.

For simplicity of reading, moving forward **"genetic counseling via telehealth** (e.g., video conferencing)**"** will be abbreviated as **"tele-genetic counseling"**

Is tele-genetic counseling for HD currently offered through your practice or institution?

☑ Yes ☑ No

When did your practice begin providing tele-genetic counseling for HD?

☑ Before March/April 2020 ☑ During or After March/April 2020

Was there any time period when all genetic counseling in your practice was tele-genetic counseling?

☑ Yes ☑ No

Which of the following best describes how long your place of practice provided tele-genetic counseling for HD as a common mode of delivery?

☑ ~March/April 2020 – ~June 2020 ☑ Through ~Dec 2020 ☑ Through March 2021

Currently, do you personally provide tele-genetic counseling for HD?

☑ Yes ☑ No

The following questions will ask you about genetic counseling and telehealth services offered for the Huntington’s disease community over two time frames:

- **2020; After onset** of the COVID-19 pandemic, implementation of telehealth due to COVID-19
- **2019; Before onset** of the COVID-19 pandemic, implementation of telehealth due to COVID-19

This section will ask questions regarding services provided to the Huntington’s disease community over the course of **2020, after onset** of the COVID-19 pandemic and implementation of telehealth due to COVID-19 restrictions.

Please check all applicable boxes indicating services you delivered, and the modes with which you delivered those services. More than one box per service, if applicable. For example, if in 2020 you provided pre-test counseling via phone, video chat, and in-person, please check **all 3** boxes. If you personally did not provide pre-test counseling, please check “N/A.”

In 2020, I personally provided the following services for HD patients through the following modes of delivery:

	Mode of Delivery:			
	Phone	Video Chat	In-Person	N/A
• Pre-test Counseling	☑	☑	☑	☑
• Results Disclosure	☑	☑	☑	☑

● Follow-up Counseling	☑	☑	☑	☑
● Psych. Assessment	☑	☑	☑	☑
● Intra-clinic communication	☑	☑	☑	☑

In 2020, the following services have been provided **via telehealth** by ***other*** members of my care team for HD patients:

	Mode of Delivery:			
	Phone	Video Chat	In-Person	N/A
● Neurologic Exam	☑	☑	☑	☑
● Psych. Assessment	☑	☑	☑	☑
● Results Disclosure	☑	☑	☑	☑
● Follow-up Counseling	☑	☑	☑	☑
●				

In 2020, I had HD patients **who declined** the option of telehealth for any of the following services:

	Yes	No	Don't Know
● Pre-test Counseling	☑	☑	☑
● Results Disclosure	☑	☑	☑
● Follow-up Counseling	☑	☑	☑
● Neurologic Exam	☑	☑	☑
● Psych. Assessment	☑	☑	☑
● Other or Additional ____________			

If HD patients declined telehealth during this time, did any of them do so with the intention to instead see a provider in-person at a future date?

☑ Yes ☑ No ☑ I don't know☑ N/A

In 2020, did any **HD patients who declined telehealth during this time** eventually decide to schedule a telehealth visit anyway?

☑ Yes ☑ No ☑ I don't know☑ N/A

In 2020, I provided genetic counseling services for HD patients **from my home**

☑ Yes ☑ No

In 2020, I provided genetic counseling services for HD patients in the following settings: (please check all that apply)

☑ Hospital/Medical Facility – Academic Medical Center
☑ Hospital/Medical Facility – Private (nonprofit or for profit)
☑ Hospital/Medical Facility – Public
☑ Other ___________

In 2020, the geographical setting of my institution would be best be described as:

☑ Urban area ☑ Suburban area ☑ Small city ☑ Rural

This next section will ask questions regarding services provided **in 2019, before the onset** of the COVID-19 pandemic

> Please check all applicable boxes indicating services you delivered, and the modes with which you delivered those services. More than one box per service, if applicable. For example, if in 2019 you provided pre-test counseling via phone, video chat, and in-person, please check **all 3** boxes. If you did not provide pre-test counseling, please check "N/A."

In 2019, I personally provided the following services for HD patients through the following modes of delivery:

	Mode of Delivery:			
	Phone	Video Chat	In-Person	N/A
• Pre-test Counseling	☑	☑	☑	☑
• Results Disclosure	☑	☑	☑	☑
• Follow-up Counseling	☑	☑	☑	☑
• Psych. Assessment	☑	☑	☑	☑
• Interclinic communication	☑	☑	☑	☑

In 2019, the following services were provided **via telehealth** by ***other*** members of my care team for HD patients

	Mode of Delivery:			
	Phone	Video Chat	In-Person	N/A
• Neurologic Exam	☑	☑	☑	☑
• Psych. Assessment	☑	☑	☑	☑
• Results Disclosure	☑	☑	☑	☑
• Follow-up Counseling	☑	☑	☑	☑

In 2019, as far as you know, did any HD patients **request telehealth counseling** with the intention of using this service instead of seeing someone in-person?

☑ Yes ☑ No ☑ I don't know

In 2019, as far as you know, were any HD patients **offered telehealth counseling**?

☑ Yes ☑ No ☑ I don't know

In 2019, I provided genetic counseling services for HD patients **from my home**

☑ Yes ☑ No

In 2019, I provided genetic counseling services for HD patients in the following settings: (please check all that apply)

☑ Hospital/Medical Facility – Academic Medical Center
☑ Hospital/Medical Facility – Private (nonprofit or for profit)
☑ Hospital/Medical Facility – Public
☑ Other ___________

In 2019, the geographical setting of my institution/employer's physical location would best be described as:

☑ Urban area ☑ Suburban area ☑ Small city ☑ Rural

[More on next page]

Please read each of the following statements and select the corresponding response to indicate how much you agree with the statement.

Comfort

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
Before the pandemic, I was **uncomfortable** with the idea of using telehealth for HD sessions.	☑	☑	☑	☑	☑
I feel **comfortable** with providing tele-genetic counseling for the HD community.	☑	☑	☑	☑	☑
I am more **comfortable** providing tele-genetic counseling for HD now than I was before the onset of COVID-19 restrictions.	☑	☑	☑	☑	☑
I would **prefer** to provide my HD genetic counseling services in-person with the patient.	☑	☑	☑	☑	☑

Interpersonal Rapport

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
I find it more difficult to identify **verbal** cues in telehealth sessions than in in-person sessions.	☑	☑	☑	☑	☑
I find it more difficult to identify **non-verbal** cues in telehealth sessions than in in-person sessions.	☑	☑	☑	☑	☑
In telehealth sessions, it is difficult to establish emotional	☑	☑	☑	☑	☑

rapport with the patient.					

Quality of Care

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
Overall, patients have seemed **satisfied** with the services I provided via telehealth.	☑	☑	☑	☑	☑
Patients expressed **positive** feelings about telehealth.	☑	☑	☑	☑	☑
Patients expressed **negative** feelings about telehealth.	☑	☑	☑	☑	☑
Overall, patients have seemed **comfortable** using telehealth services for HD.	☑	☑	☑	☑	☑
I am confident that the **informational** needs of HD patients were met in telehealth sessions.	☑	☑	☑	☑	☑
I am confident that the **emotional** needs of HD patients were met in telehealth sessions.	☑	☑	☑	☑	☑

In an Ideal World

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
In an ideal world, **pre-test** counseling for HD **predictive** testing would not be provided via telehealth.	☑	☑	☑	☑	☑
In an ideal world, **results disclosure** for HD **predictiv**e testing would not be provided via telehealth.	☑	☑	☑	☑	☑
In an ideal world,	☑	☑	☑	☑	☑

neurological exams for HD would not be provided via telehealth.					
In an ideal world, **psychological assessments** for HD would not be provided via telehealth.	☑	☑	☑	☑	☑

<u>Lasting Potential</u>

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
I imagine the option for tele-genetic counseling for HD **will** routinely be offered to patients as an option, beyond the COVID-19 pandemic.	☑	☑	☑	☑	☑
I imagine predictive test results disclosure sessions for HD **will** have a telehealth option beyond the COVID-19 pandemic.	☑	☑	☑	☑	☑
I feel that tele-genetic counseling for HD **should** continue to be provided as an option for patients.	☑	☑	☑	☑	☑
I feel that **predictive** test **results disclosure** sessions for HD **should** have a telehealth option beyond the COVID-19 pandemic.	☑	☑	☑	☑	☑

<u>Other</u>

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
My workplace made efforts to make the utilization of telehealth as easy as possible **for me** to provide my services to patients.	☑	☑	☑	☑	☑

My workplace made efforts to make the utilization of telehealth as easy as possible **for patients**.	☑	☑	☑	☑	☑
I would **prefer** to provide my services via telehealth, if in-person service delivery requires taking precaution, such as the use of masks and/or social distancing.	☑	☑	☑	☑	☑
HD patients always had a **support person** with them for results disclosure sessions via telehealth.	☑	☑	☑	☑	☑
I am confident that assessment of **suicidal** ideation / risk was adequate in telehealth sessions.	☑	☑	☑	☑	☑

Please estimate how frequently patients mentioned any of the following **positive** aspects of telehealth during telehealth counseling sessions you conducted since March/April 2020. (Please check box to indicate frequency):

	Yes, frequently	Yes, but not often	No	Maybe, I don't remember
• Saved time	☑	☑	☑	☑
• Saved money	☑	☑	☑	☑
• Did not have to travel	☑	☑	☑	☑
• Did not have to take time off from work	☑	☑	☑	☑
• More comfortable to do at home	☑	☑	☑	☑

• Helped due to HD-related mobility issues	☑	☑	☑	☑
• Made it easier for support person or caretaker to attend visit	☑	☑	☑	☑
• Able to include more family or support persons that otherwise wouldn’t have made an in-person visit	☑	☑	☑	☑

Please estimate how frequently patients mentioned any of the following **negative** aspects of telehealth during telehealth counseling sessions you conducted since March/April 2020. (Please check box to indicate frequency):

	Yes, frequently	Yes, but not often	No	Maybe, I don’t remember
• Bad internet connection / phone reception	☑	☑	☑	☑
• Difficulty with technology	☑	☑	☑	☑
• Concerns of internet or information security/privacy	☑	☑	☑	☑
• Concern with lack of privacy from other family members in the home	☑	☑	☑	☑
• Concern that care provided via telehealth may be inferior to in-person care	☑	☑	☑	☑

Please help us improve the quality of our survey:

Were there any questions that you felt you were not able to answer accurately? Which question(s)? **[If yes, please use this open text box/space to answer. If no, please skip.]**

Were there questions we did not ask, that you feel we should have included in the survey? If so, please let us know. **[If yes, please use this open text box/space to answer. If no, please skip.]**

Was there anything about your experience in providing genetic counseling to the HD community, in-person or via telemedicine, you would like to tell us about? **[If yes, please use this open text box/space to answer. If no, please skip.]**

END

Thank you for participating in this study!

Would you be interested in being contacted to participate in a 30-45 min follow-up interview, in which you would be compensated with an additional $50 gift card?

If no → [takes to external site]

"Thank you for your participation in this study. If you would like to receive a $25 Amazon gift card as compensation for completing this survey, please enter your email address below:

Email:____________________________
Verify Email: ______________________

Your survey responses are not linked to contact information. This contact information will be destroyed after payment/compensation is sent.

Please forward the link below to any other genetic counselors who provide services for the HD community and may be interested in participating in this study.

[*URL Link to forward to other potentially-interested genetic counselors that counsel for HD*]

If yes → [continues in survey site]

You may be selected to participate in a short, semi-structured follow-up interview (30-45 mins.) for more in-depth discussion about your experience of providing telehealth services for the HD community. If selected to participate, you would be compensated with an additional $50 Amazon gift card, delivered upon completion of the interview. Please enter your contact information below.

Contact information: **Name:_________________________**
Phone:_________________________
Verify Phone: ___________________
Email:__________________________
Verify Email: ____________________

Preferred form of contact
to schedule interview: **Phone ☑ Email**

Your information will be kept confidential and is only used for the following reasons:

- To email your $25 gift card compensation(s) for completing this survey
- To facilitate communication for a potential follow-up interview.
- To inform interview design, participant selection, and recruitment.

Please forward the link below to any other genetic counselors who provide services for the HD community and may be interested in participating in this study.

[*URL Link to forward to other potentially-interested genetic counselors that counsel for HD*]

Appendix IV: Interview Guide

30-45 min Semi-Structured Interview Guide

[Interviewer preamble and verbal consent script:] Hi, thank you for agreeing to participate in this interview. *In this interview, I'd like to discuss your thoughts and experiences with providing tele-genetic counseling services for the Huntington's disease community.*

Your participation in this interview is voluntary and your responses will remain confidential; interview data will be used for research purposes to explore tele-genetic counseling services for HD. You may refuse to answer any questions and you can discontinue your participation at any time by asking me to stop. To protect your privacy, after this interview, your name will be replaced with a code number identifier; we will not use your name, contact information, the name of your place of work, or any other identifiable information when analyzing or reporting this data. Your name, contact information, place of work, coworkers' names, or any other identifiable information that may arise will be redacted from the transcript. We also want to avoid any use of protected health information for your patients. If a patient's name or other identifiable information is discussed, all of those details will be redacted from the transcript. We will be recording the interview, which will take around 30-45 minutes to complete. Do you have any questions about the purpose of this interview or concerns about this process?

Do you consent to participation in this interview? *If no,* ***stop*** *If yes,* ***continue***

Start recording

Interviewer should begin with the interview number and date of interview for the record.

<u>DOMAIN: GENERAL BACKGROUND AND PRACTICE CHARACTERISTICS</u>

1) To get started, I'd like to hear about your practice. Can you tell me a little bit about your work background and your current work with patients?
    a. What types of patients to do see [e.g. age range of patients, race/ethnicity, rural or (sub)urban]?
    b. What types of health conditions do you see most often?
    c. Do you see patients independently or with a supervisor/colleague?
    d. Is it common for you to see a patient or family more than once for follow-up?

2) How often do you see HD patients as part of your job?
    a. What proportion of your patient volume consists of HD patients?

3) Traditionally, what has your clinic's protocol for HD genetic counseling looked like?
    a. How closely have you been following the published HD protocol guidelines?
    b. How is your approach to HD counseling similar or different to your approach to counseling patients with other conditions?

DOMAIN: PRE-PANDEMIC TELEHEALTH (VIDEO CONFERENCING) USE

In this next section, I'd like to ask you about telehealth through video conferencing broadly, not necessarily specific to Huntington's disease.

4) Can you describe for me whether and how telehealth was being used at your institution prior to the COVID-19 pandemic?
    a. How was it set up?
    b. How often was it used, and by whom?

5) What did the use of telehealth look like in genetic counseling?
    a. To what extent did genetic counselors use telehealth?
    b. What kinds of patients did genetic counselors see via telehealth?
    c. How was this going?

6) Did you personally have any experience seeing patients via telehealth prior to the pandemic?
    a. If yes, what kinds of patients did you see this way and how did it go?
    b. Did you see any HD patients via telehealth? Tell me why or why not

7) Overall, what were your feelings about using telehealth for genetic counseling prior to the pandemic?
    a. What did you think were the strengths and weaknesses of the approach?
    b. What were the barriers or facilitators of using telehealth?

DOMAIN: IMPACT OF COVID-19 PANDEMIC ON GENETIC COUNSELING SERVICE

Now I'd like to ask you a few questions about any effects of the COVID-19 pandemic on your services.

8) When did the COVID-19 pandemic begin to affect your work?
    a. How did your employer respond to the pandemic?
    b. How did the pandemic affect the way you cared for patients?

9) (IF TGC was part of the pandemic response)
    a. When did implementation of telegenetic counseling in your practice or clinic take place?
    b. Tell me what this looked like. How did this transition go?
    c. Tell me about anything you wish had gone differently.

10) How did other members of your team or institution feel about the use of TGC (e.g., neurologists, admin; supportive or resistant)?

11) What video conferencing platforms did you use for HD-TGC sessions (e.g., Zoom, WebEx, etc.)?
    a. Did you prefer any platforms over others?
    b. What features influence your preference?
    c. Did your satisfaction with the performance of this platform affect your attitude toward TGC?
    d. How did your increased use of telecommunication during the pandemic (in general, outside work) impact your feelings about using telehealth over time?

DOMAIN: EXPERIENCES WITH HD-TGC SPECIFICALLY

In this next section I'd like to hear about your experience with tele-genetic counseling for *Huntington's disease,* specifically:

12) Can you tell me how your institution made the decision to start providing genetic counseling to HD patients via telehealth?
    a. How straightforward was this decision? Was HD genetic counseling considered differently than other types of genetic counseling? Why or why not?
    b. How did you feel about seeing HD patients via telehealth?
        1. Nervous, optimistic, pessimistic? About what?

13) How closely did your approach to HD telegenetic counseling mirror your pre-pandemic protocol for counseling these patients?

14) In your survey, you mentioned that you did (X, Y, Z) components of genetic counseling for HD via telehealth. I'm going to ask you to describe how each of these went.
    a. How did (X, Y, Z) compare to in-person counseling?
    b. Would you consider doing X,Y Z via telehealth in the future, even if the COVID-19 pandemic no longer necessitates it?

15) How comfortable do you think HD patients were with having genetic counseling done via telehealth?
    a. Were certain kinds of patients more comfortable than others? Tell me more about this.

16) How did HD-TGC sessions for presymptomatic vs. symptomatic patients differ?

17) Did you get any feedback from HD patients about telehealth?
    a. Tell me what you heard from patients.
    b. What positive things did they have to say about it?
    c. What negative things did they have to say?

DOMAIN: ATTITUDES TOWARD HD-TGC

I'm going to wind down by asking for your views about the *appropriateness* of using HD-TGC:

18) Broadly, why do you think many clinics have restricted genetic counseling for HD to in-person delivery in the past?
    a. How do you feel about that reasoning?

19) How did your experience validate or contradict this reasoning?

20) How well did you feel like you could establish emotional rapport with HD patients using telehealth?

21) How well did you feel you could assess mental health concerns in HD patients over telehealth?

22) How would you describe your interactions with HD patients' family members via telehealth? What challenges or opportunities did telehealth present in this regard?

23) Can you describe any other strengths or weaknesses of using telehealth for HD genetic counseling?

DOMAIN: FINAL THOUGHTS (only ask if hasn't been addressed yet)
We're approaching the end of the interview! In this last section I'd like to hear you summarize some of the main takeaways you've learned from providing HD-TGC during the pandemic:

24) To recap, **positive reflections** regarding the use of HD-TGC in your clinical setting seem to be **[restate]**. Does anything else come to mind?

25) **Positive reflections** from the patients' perspective regarding the use of HD-TGC seem to be **[restate]**. Does anything else come to mind?

26) What **barriers** may exist that need to be addressed in order for broad acceptance and use of HD-TGC?
    a. What do you think it would take to overcome these barriers?

27) What opportunities do you think HD telehealth presents that we should learn from and continue to take advantage of in the future?

28) What didn't I ask you that you think is important for me to know about the way you think about tele-genetic counseling services for Huntington's disease?

Stop recording.

Curriculum Vitae

WES SOLEM

wes.solem@gmail.com

EDUCATION

SEP 2019 – JAN 2022

GENETIC COUNSELING, ScM, **JOHNS HOPKINS UNIVERSITY / NATIONAL INSTITUTES OF HEALTH**

Intramural Research Training Award (IRTA) recipient

CO-CHAIR, HEALTH, BEHAVIOR, AND SOCIETY STUDENT ORGANIZATION, BLOOMBERG SCHOOL OF PUBLIC HEALTH

THESIS, *"COVID-19 Impact on Genetic Counseling for Huntington's Disease via Telehealth"*

Advisors: Leila Jamal, PhD, ScM, CGC; Debra Roter, DrPH

SEP 2015 – JUNE 2019

BEHAVIORAL NEUROSCIENCE, BS, **BIOLOGY**, Minor, **WESTERN WASHINGTON UNIVERSITY**

ROTATIONS

LABORATORY	**JOHNS HOPKINS DNA DIAGNOSTIC LAB**	**OCT-DEC 2019**
PRENATAL	**HOWARD UNIVERSITY HOSPITAL**	**JAN-MAR 2020**
CONNECTIVE TISSUE	**JOHNS HOPKINS HOSPITAL**	**APR-MAY 2020**
CANCER	**NATIONAL CANCER INSTITUTE**	**JUN-JUL 2020**
NEUROGENETICS	**UNIVERSITY OF WASHINGTON**	**JUN-JUL 2020**
CANCER	**UNIVERSITY OF WASHINGTON**	**JUN-OCT 2020**
CYTOGENETICS	**UNIVERSITY OF WASHINGTON**	**SEP-OCT 2020**
PEDIATRICS	**CHILDREN'S NATIONAL MEDICAL CENTER**	**OCT-DEC 2020**
POLICY	**NAT'L HUMAN GENOME RESEARCH INSTITUTE**	**FEB, JUL 2021**
NEUROGENETICS	**NAT'L INSTITUTE NEURO. DISORDERS & STROKE**	**APR-JUL 2021**
CANCER	**INVITAE**	**AUG-OCT 2021**
PRENATAL	**MERCY MEDICAL CENTER**	**OCT-DEC 2021**
IMMUNE DEFICIENCY	**NAT'L INST. ALLERGY & INFECTIOUS DISEASES**	**OCT-DEC 2021**

OTHER EXPERIENCE

LABORATORY ASSISTANT, **WESTERN WASHINGTON UNIVERSITY**, **DR. JEFF CARROLL**

Researched the neurobiology and behavior of Huntington's disease mouse models. **MAR '16-JUN '19**

Donald A. King Research Fellow, Huntington's Disease Society of America **JUL-SEP 2018**

- ○ *"Metabolic and Molecular Effects of Adipose Tissue-Specific Knockout of Htt"*
    - Poster presentation, 34th Annual HDSA Convention, Boston, MA **JUN 2019**

Contributing Author, Scientific Reports - Nature **FEB 2018**

- ○ *"Early Detection of Apathetic Phenotypes in Huntington's Disease Knock-in Mice Using Open-Source Tools"*

Lead Facilitator, Carroll Lab Journal Club **JAN '17-JUN '19**

INTERN - GENETIC COUNSELING, **NORTHWESTERN UNIVERSITY** **JUL 2018**

VOLUNTEER CRISIS COUNSELOR, CRISIS TEXT LINE	JAN-JUN 2019
INTERN - NEURODIAGNOSTICS, PEACEHEALTH CASCADE BRAIN & SPINE CTR.	APR-JUN 2018
PEER SEXUAL HEALTH EDUCATOR, WESTERN WASHINGTON UNIVERSITY	SEP ‘18-JUN ‘19
TOUR MANAGER, **STAGE MANAGER**, **GUITAR TECHNICIAN**, VARIOUS ARTISTS	JAN ‘05-SEP ‘15