Patient uptake and satisfaction with genetic counseling service delivery models in adults with retinal dystrophies

by

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Patient uptake and satisfaction with genetic counseling service delivery models in adults with retinal dystrophies

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Genetic counseling services are essential to achieve the highest quality care management in adults with retinal dystrophies. The retinal dystrophy clinic housed within the University of Pittsburgh Medical Center (UPMC) provides the option of genetic counseling and testing to all patients through in-person and remote telemedicine options. While various genetic counseling service delivery models have been established, there is little known about satisfaction with the appointment types for patients that have a confirmed or suspected retinal dystrophy. Thirty-five patients were seen for pre-test genetic counseling between October 3, 2022 and February 6, 2023. Patients self-selected one of the following four appointment types: in-person in-coordination with another appointment in the department, in-person genetic counseling only, telemedicine video genetic counseling only or telemedicine phone genetic counseling only. All patients were contacted within one week of completing their appointment with an invitation to participate in a satisfaction survey consisting of the Genetic Counseling Satisfaction Scale (GCSS) and independently developed questions. The majority of patients completed genetic counseling in-coordination with another appointment (N =15) or via telemedicine video (N =14). Eighteen individuals completely the survey, for a response rate of 51.4% (18/35). The results suggest that patients were highly satisfied with genetic counseling regardless of service delivery model. There was no significant difference in uptake of service delivery model and/or satisfaction between respondents that did and did not meet the criteria for legal blindness, meeting a tenant of public

health by providing equitable care. This study demonstrates that telemedicine genetic counseling is an acceptable service delivery model for adults with confirmed or suspected retinal dystrophies. Genetic counseling should continue to be offered in-person and via telemedicine to help ensure that all patients have the ability to access care by their preferred appointment type.

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Preface

I would like to thank my thesis committee for their support and contributions to this project. This research would not have been possible without their guidance and expertise. Specifically, I would like to acknowledge my thesis chair, Michelle Alabek, for her assistance and endless encouragement from the conception of the project.

I would also like to thank the retinal dystrophy clinic team at UPMC for the time they put into scheduling and conducting the appointments. I am grateful to the patients who participated in the survey for giving me the opportunity to learn from their experiences.

My success through my graduate school career as a whole is due to the support I have received from coworkers, instructors and program directors. To my classmates, friends and family – thank you for being with me every step of the way.

1.0 Introduction

The implementation of telemedicine in genetic counseling has increased due to the COVID-19 pandemic. The ability to meet with a genetic counselor through virtual video visits has proven to be beneficial for many patients by increasing their access to care. Telemedicine has provided the greatest benefit to individuals that have trouble accessing transportation, medical conditions that make for challenging clinic experiences, and time constraints due to work and other commitments (Uhlmann et al., 2021). While there are many advantages to telemedicine platforms in genetic counseling, the availability and opportunity for in-person genetic counseling appointments should not be overlooked. Virtual visits may not be practical for patients with limited access to devices with a camera and internet issues causing complications with video quality and connectivity (Uhlmann et al., 2021). It is conceivable that this could lead to difficulty building rapport and reduce the psychosocial interactions that are a key component of genetic counseling. The benefits and limitations associated with telemedicine genetic counseling have been primarily derived from studies conducted in cancer clinics (Breen et al., 2022; Buchanan et al., 2015). The large majority of patient satisfaction with telemedicine has also been measured by surveying patients receiving cancer genetic counseling. Results of these studies have shown that there is evidence of a growing trend towards and satisfaction with telemedicine genetic counseling services in oncology (Breen et al., 2022). There is limited information, however, on patient interest and satisfaction with telemedicine genetic counseling in specialty clinics (Dratch et al., 2021).

To date, there have been limited studies that examined patient uptake and satisfaction with telemedicine vs. in-person genetic counseling appointments in adult ophthalmology. A previous study in pediatrics suggested that genetic eye disease clinics are suitable for remote service

delivery and have proven to be acceptable to families of children with inherited eye disease (Bell et al., 2021). There is no data to support or refute if this is also the case among adult patients. It has been noted that there is a lack of information on comparisons between in-person and telemedicine appointments related to satisfaction amongst patients with eye diseases (Macarov et al., 2021). The ability to better understand patient uptake with the type of genetic counseling service delivery in ophthalmology will allow for a higher quality of care. The use of genetic testing is especially valuable in ocular subspecialities such as retinal dystrophies due to the high number of genetically identifiable conditions (Garafalo et al., 2020; Lam et al., 2021). The results of genetic testing for retinal dystrophies can be used to confirm diagnoses and determine eligibility for gene specific trials and therapies (Macarov et al., 2021; Sutherland & Day, 2009). This population is unique in that an individual's diagnosis of a retinal dystrophy will lead to a decrease in vision over time and may impact their ability to navigate telemedicine platforms. Specifically, a need for retinal dystrophy patient uptake and satisfaction with telemedicine genetic testing has been called for in recent literature (Al-Moujahed et al., 2021). This data could help to determine if telemedicine is an acceptable service delivery model for genetic counseling among patients seen for this indication.

The purpose of this project is to develop an understanding of retinal dystrophy patient uptake and satisfaction with telemedicine genetic counseling. To best learn about patient service delivery model preferences, patient data will be extracted from an existing clinical database housed in the Research Electronic Data Capture, REDCap. This includes demographic information, diagnosis and visual acuity as reported in the most recent clinical note. These measures are considered to be a standard of care and are readily available. A quality improvement survey approved by the UPMC Quality Review Committee (Project ID #4046) will be conducted to

collect information on patient satisfaction with their appointment. The survey will consist of the genetic counseling satisfaction scale (GCSS) and open-ended questions that were developed with the goal of improving patient care (Tercyak et al., 2001).

Together, the study aims to evaluate patient uptake of and satisfaction with telemedicine and in-person genetic counseling appointments. The patient satisfaction survey responses and self-selected service delivery model will be matched with their demographic information, diagnosis and visual acuity. The data will be analyzed to determine if telemedicine genetic counseling is a satisfactory service delivery model when compared to in-person appointments for adult patients with retinal dystrophies.

2.0 Manuscript

2.1 Background

The onset of COVID-19 in the United States during the spring of 2020 rapidly altered the use of telemedicine in genetic counseling. Genetic counselors, along with the majority of the medical field, were forced to evolve quickly in order to provide quality care to patients while minimizing the exposure risk to all. Unlike many other medical professionals, genetic counseling appointments do not require a physical exam. Armed with this advantage, genetic counseling via telemedicine had been established at many institutions prior to the pandemic. As the use of telemedicine in genetic counseling has increased, more information on the benefits and limitations of the service delivery model have been presented. Collectively, patients have reported that the most significant benefits include overcoming geographic barriers, lower cost, the opportunity to have family members join in from other locations and a reduction in stress that can be caused by planning for a medical visit (taking off work, finding childcare, etc.) (Uhlmann et al., 2021). Patients also reported on what they felt to be the greatest limitations of telemedicine, which included challenges forming a relationship with the genetic counselor and receiving emotional support, technical difficulties (video quality, internet access, etc.), lack of privacy and surrounding distractions (Uhlmann et al., 2021). These factors have helped to provide a better understanding of the patient experience with telehealth and what may be driving their decisions when selecting a genetic counseling service delivery model. Given the previously described benefits and limitation, there is little surprise that past studies have suggested the preference for telemedicine is strongest in patients that prioritize convenience over personal connection (Allison et al., 2022).

Many clinics will likely adapt to provide a hybrid model of care, allowing for both in-person and telemedicine genetic counseling appointments, depending on the purpose of the visit (Uhlmann et al., 2021). The goal of the hybrid approach is to provide quality care while reducing the time that patients are required to spend in clinic, ultimately limiting potential exposures. Prior to the development and implementation of the hybrid model, patients that were seen in multidisciplinary eye clinics could spend up to two days in clinic undergoing testing, examinations and meeting with various doctors (Rali et al., 2021). While this was widely accepted as the standard of practice prior to COVID-19, the shift in modalities was essential to allow for ongoing care and management of new referrals. The clinical hybrid model consists of two parts, the necessary in-person testing and the telemedicine encounter with the physician(s) and other members of the medical team. To provide a full evaluation and monitor patient status of inherited retinal diseases, the diagnostic testing that must be complete in-person is essential. The remainder of the standard clinical appointment is then conducted at a later time via telemedicine. A study at Emory Ophthalmic Genetics Service examined patient completion of diagnostic testing and satisfaction using a hybrid model. The study reported that variables such as visual acuity did not have an impact on telemedicine favorability and that the majority of patients "agreed" or "strongly agreed" that they like seeing the doctor in this way (telemedicine) as much as seeing him/her the ordinary way (in-person) (Rali et al., 2021). As hybrid models continue to develop, it will be important to recognize the advancements in virtual care and be mindful of its growth to ensure the best practices are being instituted.

Another study completed with a pediatric population in the UK found that remote service delivery models are appropriate for ocular genetics (Bell et al., 2021). It was reported that 96% of patients felt comfortable communicating health information via a phone consultation, but that

64.3% would have preferred a video consultation to receive services as opposed to a phone consultation (Bell et al., 2021).

Patient reported satisfaction with genetic counseling services allows providers to better understand the patient experience. This information can be used to determine if any adjustments to patient management could increase future quality of care. A study completed in the setting of prenatal genetic counseling addressed psychosocial outcomes for patients that had an appointment with a genetic counselor about amniocentesis decision making. The satisfaction was measured using a survey that has since become known as the Genetic Counseling Satisfaction Scale (GCSS) (Tercyak et al., 2001). The GCSS is a validated six-item Likert scale survey. The goal is to assess the levels to which patients agree with the positive statements about their genetic counseling experience. The topics include perceived understanding and recognition of stress the patient was facing, perceived value of the session, levels of reassurance and length of appointment (Tercyak et al., 2001). The survey has been used in other genetic counseling specialties including cancer and neurology (DeMarco et al., 2004; Dratch et al., 2021).

There are several studies that examine patient satisfaction with telehealth; however, the majority have occurred in the setting of cancer genetic counseling. One study conducted at Memorial Sloan Kettering Cancer Center evaluated patient experience with technology, emotional response and preferences for future visits while measuring patient satisfaction with the genetic counseling appointment using the Genetic Counseling Satisfaction Scale (GCSS) (Breen et al., 2022). It was reported that most participants were highly satisfied with their telehealth appointment and that 78.6% of participants would recommend scheduling a telehealth appointment to others (Breen et al., 2022). A separate study conducted in cancer genetics cited that while patients were

willing to attend telemedicine appointments, 32% stated they would have preferred to be seen by a genetic counselor in-person (Solomons et al., 2018).

Little is known about telegenetics satisfaction in other specialties or subspecialties. One study conducted in neurogenetics showed that 87% of participants strongly agreed that the telemedicine genetic counseling was useful, with 71% reporting they would use this service delivery again (Dratch et al., 2021). This study utilized data from telegenetic video visits and phone calls for pre and post-test counseling. Overall, each study concluded that patients were willing to participate and found satisfaction in telemedicine genetic counseling services.

To our knowledge, patient uptake and satisfaction with telemedicine genetic counseling for inherited retinal dystrophies has not been reported in current literature. With the increasing development of gene therapies for retinal dystrophies, it is especially important that patients have access to genetic counselors that are trained in the specialty. There can be phenotypic overlap amongst various retinal dystrophies, amplifying the importance of genetic testing, as it may help provide a definitive diagnosis that guides care management. In many cases the gene therapies are gene- or variant-specific, making genetic testing a common pre-cursor for determining eligibility. Genetic counseling appointments for adults with inherited retinal dystrophies have occurred through various modalities including telemedicine (phone and video), as well as in-person. Information on patient uptake and satisfaction with genetic counseling will provide insight as to the needs of patients and if they are being met similarly across various service delivery models.

2.2 Methods

2.2.1 Study Design and Participant Uptake

This study was approved by the Quality Improvement Committee at the University of Pittsburgh Medical Center (UPMC) (See Appendix A.2). Potential participants included patients with suspected retinal dystrophy referred for genetic counseling as part of their routine care at UPMC Eye Center from October 3, 2022 to February 6, 2023. Patients were given the option to self-select one of the following service delivery models: in-person – in coordination with another appointment in the department (IP-C), in-person – genetic counseling only (IP-GC) and telemedicine visit via secure video platform – genetic counseling only (TM-V). If specifically requested by the patient and approved by the genetic counselor, telemedicine visit via phone– genetic counseling only was accommodated (TM-P).

The retinal dystrophy clinic at UPMC functions as a multidisciplinary clinic. During these extensive appointments, patients who are referred for genetic counseling are given the option to initiate genetic counseling same day or schedule an appointment at a later time, either in-person or through telemedicine. Patients that elected to remain in clinic are considered to have had genetic counseling IP-C. The appointment availability was the same across all genetic counseling only service delivery models.

Information on patient age, diagnosis and visual acuity was obtained from their most recent clinic note from an ophthalmologist in the retinal dystrophy clinic. When available, the patient's visual field test at the time of their genetic counseling appointment was also interpreted.

2.2.2 Survey Development and Distribution

A nine-question survey consisting of both validated and independently developed questions was designed to measure patient satisfaction with service delivery models in genetic counseling. The survey is a combination of the Genetic Counseling Satisfaction Scale (GCSS) followed by three questions that are meant to identify potential barriers patients may face when scheduling an appointment (See Appendix A.1 Table 1). The GCSS consists of six items that are used to assess participant satisfaction with the genetic counselor and their perceived value of the genetic counseling session (Tercyak et al., 2001). Participants were instructed to respond to the GCSS using a 5-point Likert scale. The questions following the GCSS addressed the logistics and patient influences on scheduling telemedicine versus in-person appointments. Specifically, patient access to the resources needed for a telemedicine appointment and reliance on assistance to attend the appointment was assessed. Examples included assistance with traveling to clinic or help navigating the video visit platform. All participants were given the same survey regardless of service delivery model or status of vision. Participants included individuals that met criteria for legal blindness. Legal blindness is defined as having a visual acuity of 20/200 or less in the better eye with correction, or a visual field of 20 degrees or less.

Patients were contacted by phone one time within seven days of completing their pre-test genetic counseling appointment by a trained member of the retinal dystrophy clinic team to complete the survey. Participation was garnered through verbal confirmation that the patient was willing to share their experience and satisfaction with the genetic counseling appointment. If they were unable to be reached, a brief voice message with a return phone number was left, when possible. All participants were aware they could end the survey at any point. Upon conclusion of the survey, participants were given the option to share any additional information about their

genetic counseling experience. An individual was considered a participant if they completed the entire survey.

2.2.3 Data Collection and Analysis

Patient uptake and satisfaction survey data was collected and managed in a REDCap project designed specifically for this study. Demographic and phenotypic information was obtained from the UPMC Rare Ocular Disease Database, also housed in REDCap. All descriptive statistics were generated in Microsoft Excel. In addition, *t*-tests were performed to determine if there was a significant impact of appointment type on satisfaction score and to compare the satisfaction of respondents that did and did not meet criteria for legal blindness. Open-ended responses at the conclusion of the survey were individually reviewed and summarized.

2.3 Results

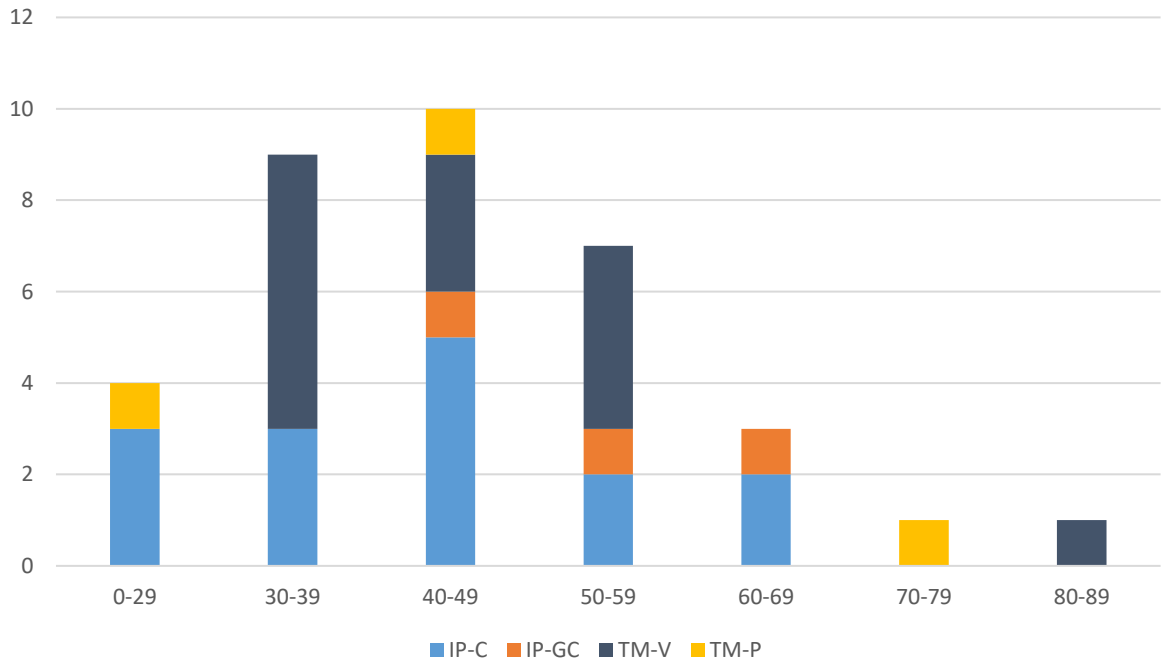
A total of 35 patients completed a pre-test genetic counseling appointment for an indication of retinal dystrophy from October 3, 2022 to February 6, 2023. Included patients ranged in age from 17-82 years old and 22 (62.9%) identified as female. Indications for referral included retinitis pigmentosa (34.3%), unspecified retinal dystrophies (25.7%), macular dystrophy (11.4%), pattern dystrophy (11.4%), cone dystrophy (5.7%), cone-rod dystrophy (5.7%) and family history of a retinal dystrophy (5.7%). Of the patients that had a genetic counseling appointment through the duration of the study, 14 of the 35 met the criteria for legal blindness.

2.3.1 Service Delivery Model Uptake

The majority of the patients (*n*=15, 43%) were seen for IP-C, followed by those that elected to schedule TM-V appointments (*n*=14, 40%). The remainder of patients were split in the self-selected IP-GC (*n*=3, 8.5%) and when appropriate, TM-P appointments (*n*=3, 8.5%).

For the 14 patients that met criteria for legal blindness, six were seen for IP-C (42.9%), six chose TM-V appointments (42.9%), one appointment was conducted via TM-P (7.1%) and one chose an IP-GC appointment (7.1%) (Figure 3).

Figure 1. Service Delivery Model by Age Group



2.3.2 Satisfaction Survey Responses

Eighteen patients completed the survey, giving an overall response rate of 51.4% (18/35). All participants who answered the initial phone call consented to and completed the survey (*n*=18, 100%). The remainder of patients did not return the voice message. While most of the patients in the full cohort of 35 completed their genetic counseling appointment IP-C, the survey response rate among this group was only 33.3% (5/15), compared to 71.4% (10/14) who selected a TM-V appointment and participated in the study (Table 1, Figure 2).

Table 1. Survey Response by Genetic Counseling Appointment Type

	Completed survey	Did not complete survey	Total
IP-C			15
N	5	10	
%	33.3	66.7	
IP-GC			3
N	2	1	
%	66.7	33.3	
TM-V			14
N	10	4	
%	71.4	28.6	
TM-P			3
N	1	2	
%	33.3	66.7	
Total	18	17	35

The survey response rate for individuals that met criteria for legal blindness was 64.3% (9/14) (Figure 3). All respondents that met the criteria for legal blindness and chose a TM-V appointment completed the survey (6/6), while the survey response rate for those that did not meet

criteria for legal blindness and chose a telemedicine was 50% (4/8). The IP-C response rate for individuals that do and do not met criteria for legal blindness was the same at 33.3%.

Figure 2. Survey Completion by Genetic Counseling Appointment Type

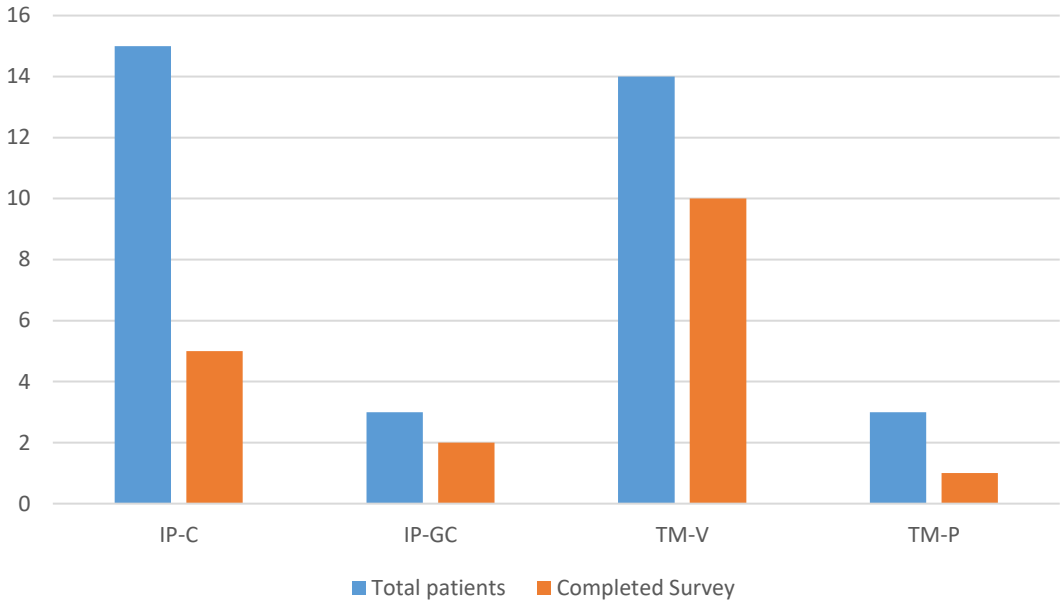
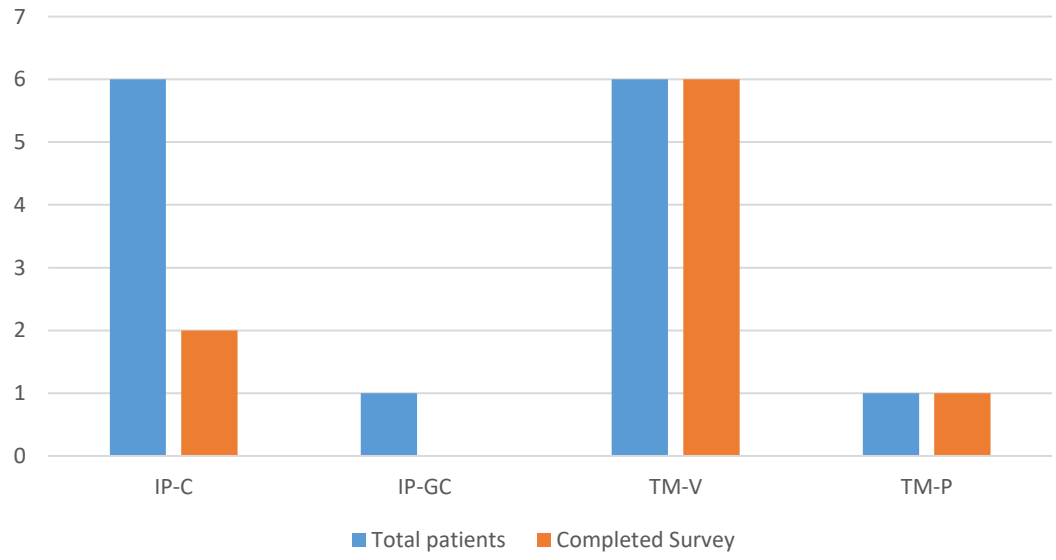


Figure 3. Survey Completion for Patients Meeting Legal Blindness Criteria



Of the 18 respondents to the satisfaction survey, the majority completed their appointment through TM-V (55.6%, n=10). One-third of survey respondents (n=6) were 50-59 years old, with only one respondent in each of the following age groups: under 29, 70-79 and 80-89 years old (Table 2).

Table 2. Survey Completion by Age and Appointment Type

	0-29	**30-39**	**40-49**	**50-59**	**60-69**	**70-79**	**80-89**
IP-C	1	1	-	2	1	-	-
IP-GC	-	-	-	1	1	-	-
TM-V	-	4	2	3	-	-	1
TM-P	-	-	-	-	-	1	-
Total	**1**	**5**	**2**	**6**	**2**	**1**	**1**

2.3.3 Satisfaction with Genetic Counseling

A majority of the 18 survey respondents reported that they "agree" or "strongly agree" to the six-items that were evaluated in the GCSS (Table 3). The two respondents that had IP-GC appointments and the one respondent with a TM-P appointment all "strongly agreed" to each of the six-items. When the survey addressed the respondents belief that the genetic counselor seemed to understand the stresses they were facing, 14 respondents "strongly agreed" and 4 "agreed". Of the 14 respondents that "strongly agreed", 8 out of 10 (80%) were scheduled as TM-V while 3 of 5 (60%) respondents had their appointment IP-C. For the 2 respondents that had IP-GC and the 1 individual that had TM-P, all 3 reported that they "strongly agree" with the statement. Furthermore, 94.4% of respondents (17/18) "strongly agreed" that their genetic counselor was truly concerned about their well-being. All respondents that had appointments TM-V and TM-P reported that they "strongly agreed" while 80% of those seen IP-C (4/5) "strongly agreed" with the statement regarding the genetic counselor's concern. Overall, 88.9% of respondents "strongly agreed" that

the genetic counseling session was valuable to them (16/18). Respondents that "strongly agreed" that the appointment was valuable consisted of 90% TM-V, 80% IP-C, with IP-GC and TM-P at 100%.

Table 3. GCSS Responses

	IP-C	IP-GC	TM-V	TM-P
My genetic counselor seemed to understand the stresses I was facing				
Strongly agree	3	2	8	1
Agree	2	-	2	-
Neutral	-	-	-	-
My genetic counselor helped me to identify what I needed to know to make decisions				
Strongly agree	2	2	9	1
Agree	2	-	1	-
Neutral	1	-	-	-
I felt better about my health after meeting with my genetic counselor				
Strongly agree	-	2	7	1
Agree	3	-	2	-
Neutral	2	-	1	-
The genetic counseling session was about the right length of time				
Strongly agree	2	2	7	1
Agree	3	-	2	-
Neutral	-	-	1	-
The genetic counselor was truly concerned about my well-being				
Strongly agree	4	2	10	1
Agree	1	-	-	-
Neutral	-	-	-	-
The genetic counseling session was valuable to me				
Strongly agree	4	2	9	1
Agree	1	-	1	-
Neutral	-	-	-	-

Four of the nine participants who met criteria for legal blindness "strongly agreed" to all six items of the GCSS, three of whom completed a TM-V appointment and one who completed a TM-P appointment. Six individuals that met criteria for legal blindness and chose to schedule a

TM-V appointment “strongly agreed” that the genetic counselor was truly concerned about their well-being and “strongly agreed” to finding value in the appointment.

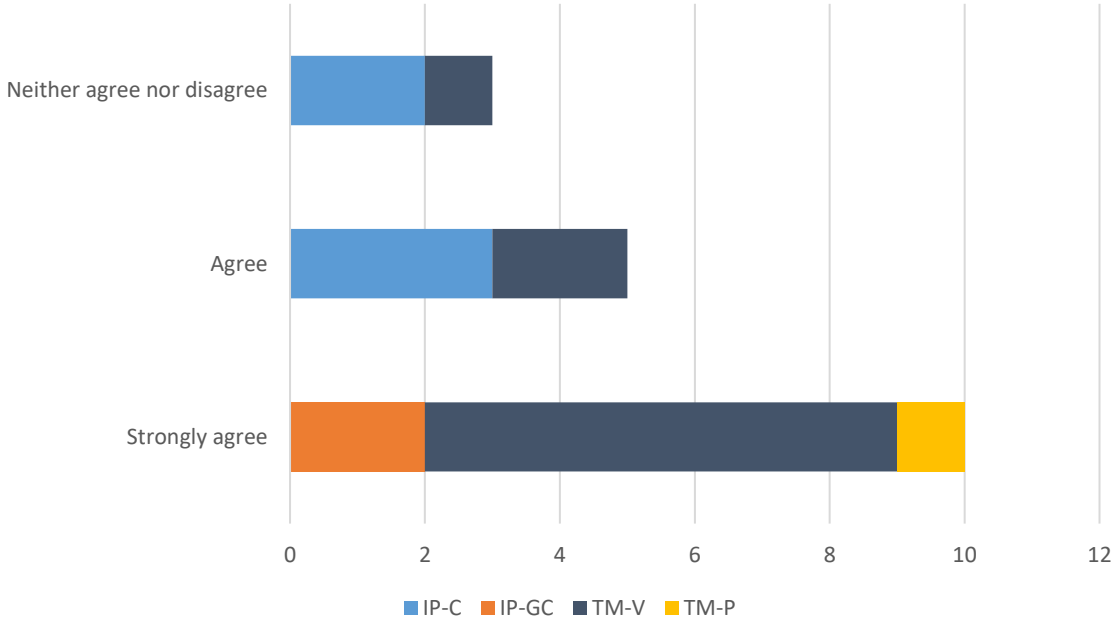
The survey respondent’s perception of the genetic counselors understanding of their stress shows that 27.8% of respondents both fall within the ages of 50-59 and “strongly agree” (5/18) (Table 4). Of these five individuals, one chose IP-GC, one was seen IP-C and three had TM-V appointments. The two oldest patients completed their appointments via TM-P and TM-V, and both “strongly agreed” that their genetic counselor understood the stresses they were facing. Regardless of age, there are four participants that “agreed” with the statement, all of these individuals had genetic counseling IP-C.

Table 4. GCSS Preceived Stress by Age

	0-29	**30-39**	**40-49**	**50-59**	**60-69**	**70-79**	**80-89**
Strongly Agree	1	4	1	5	1	1	1
Agree	-	1	1	1	1	-	-

When reviewing how patients felt about their health following their genetic counseling appointment, three stated that they “neither agreed nor disagreed” that they felt better about their health (Figure 4). None of the 5 participants that were seen in-coordination with other appointments reported that they “strongly agreed” to feeling better about their health following the genetic counseling session.

Figure 4. GCSS Health Assessment



To further understand the results of the GCSS, the responses have been coded 1-5 with 1 being strongly disagree and 5 for strongly agree. Each respondent was given a total satisfaction score that included their answers to the six items on the GCSS (Table 5). The highest possible score is 30, meaning the respondent "strongly agreed" with all statements. The average satisfaction score for all respondents suggests that they were highly satisfied with their genetic counseling experience (*M*=28.22, *SD*=1.66). The lowest combined satisfaction score resulted from individuals that had IP-C appointments (*M*=26.4, *SD*=1.34). The two lowest scores in this section came from individuals that met the criteria for legal blindness. The greatest number of respondents had TM-V appointments and also reported high satisfaction (*M*=28.7, *SD*=1.25). A majority of these individuals met criteria for legal blindness (60%) and of these respondents, half had satisfaction scores of 30. A comparison between IP-C and TM-V appointments indicated that respondents who selected TM-V appointments were significantly more satisfied $t(13) = -3.28$, $p = .003$; however,

both groups are still categorized as highly satisfied. Respondents with legal blindness that had an IP-C appointment were significantly less satisfied than those with IP-C appointments that did not meet criteria for legal blindness $t(3) = 5.42$, $p = .006$ (M=25, SD=0). Again, it should be noted that both groups still fall within the highly satisfied category. When examining the influence of legal blindness on satisfaction in TM-V appointments (M=29, SD=1.26), there is no significant difference $t(8) = 0.92$, $p = .192$.

Table 5. GCSS Total Satisfaction Score

Appointment Type	GCSS Total Score	Mean (*SD*)
IP-C	28	
(n=5)	27	
	27	
	25*	
	25*	
		26.4 (1.34)
IP-GC	30	
(n=2)	30	
		30
TM-P	30*	
(n=1)		30
TM-V	30*	
(n=10)	30*	
	30*	
	30	
	29*	
	28*	
	28	
	28	
	27*	
	27	
		28.7 (1.25)

*Met criteria for legal blindness

Across all service delivery models, the average respondent satisfaction scores for individuals that did meet the criteria and did not meet the criteria for legal blindness were 28.22 (*SD*=2.11) and 28.33 (*SD*=1.32), respectively. There was no significant effect of legal blindness on the collective patient satisfaction score with genetic counseling $t(16)=-0.13$, $p=.448$.

After being scored, the survey results showed that the average response score for combined IP and combined TM service delivery models was between 4-5 (Table 6). This means that on average, all respondents "agreed" or "strongly agreed" to each of the six items on the GCSS (See Appendix A.1 Table 2).

Table 6. GCSS Average Satisfaction Score per Question

	IP-C, IP-GC (n=7)	TM-V, TM-P (n=11)
My genetic counselor seemed to understand the stresses I was facing	4.71 (0.49)	4.81 (0.4)
My genetic counselor helped me to identify what I needed to know to make decisions	4.43 (0.79)	4.91 (0.3)
I felt better about my health about meeting with my genetic counselor	4 (0.82)	4.64 (0.67)
The genetic counseling session was about the right length of time	4.57 (0.53)	4.64 (0.67)
The genetic counselor was truly concerned about my well-being	4.86 (0.38)	5
The genetic counseling session was valuable to me	4.86 (0.38)	4.91 (0.3)

2.3.4 Logistics of Service Delivery Models

Fifteen of the eighteen respondents indicated that they had adequate access to the resources needed for a telemedicine video appointment. Of the three participants who stated they did not feel that they had access to telemedicine resources, two of these patients received genetic counseling IP-C and the remaining patient completed their appointment through TM-P. The participant that had the TM-P appointment and one of the participants that had IP-C met the criteria for legal blindness.

A total of four respondents stated that they relied on assistance to attend their genetic counseling appointment. The three previous participants that stated they do not have the resources for a telemedicine video appointment also stated they relied on assistance. The additional patient that reported they relied on assistance had a TM-V appointment and met the criteria for legal blindness. Three out of four respondents that required assistance to attend their appointment met the criteria for legal blindness. The remaining six respondents that met the criteria for legal blindness stated they did not rely on assistance.

2.4 Discussion

The study functioned as a quality improvement measure to inform genetic counseling practices in the setting of a retinal dystrophy clinic. Patients were seen for genetic counseling both independently and in coordination with other providers in the department. When given the opportunity to select their preferred service delivery model, the majority of respondents selected TM-V. Across all service delivery models, most respondents reported that they "agreed" and

"strongly agreed" to the statements in the GCSS. The responses aimed to evaluated the respondent's feelings towards their genetic counselor's ability to provide adequate information to make decisions on testing. The responses also analyzed their feelings about their genetic counselor's capacity to recognize and understand their stress and display concern over their well-being. Respondents that had IP-C appointments, and those that had genetic counseling only appointments all "agreed" or "strongly agreed" to finding value in the appointment.

While the largest number of patients were seen for IP-C, it is important to note they were still given the opportunity to schedule genetic counseling at a later time through a second in-person visit (coordinated or independent) or via telemedicine. Participants that selected to be seen for IP-GC and those that were seen for IP-C both showed satisfaction with their genetic counseling experience. Several of these participants reported that they did not have a device with the required technology for a telemedicine appointment, a frequent limitation seen in other telemedicine studies (Uhlmann et al., 2021). It is essential to recognize that the ability to access a genetic counselor in methods that were common practice before the pandemic should not be eliminated in favor of telemedicine entirely.

Of all surveys completed, the largest response rate came from those that had TM-V appointments. Patients have previously been reported to be highly satisfied with telemedicine across other disciplines, primarily prenatal and oncology (Breen et al., 2022). In our study, all participants that had TM-V appointments "strongly agreed" to feeling as though their genetic counselor was concerned about their well-being. TM-V participants showed high satisfaction, as all but one "strongly agreed" to finding value in their genetic counseling appointment. Individuals seen in specialty clinics differ from those seen in prenatal and oncology clinics, because they often do not have the same care management and treatment options available (Dratch et al., 2021). This

is consistent with the retinal dystrophy patient population, as many participants meet criteria for legal blindness. Through this study it was found that the individuals meeting the criteria for legal blindness were comfortable scheduling telemedicine video appointments and found them to be a satisfying experience. The utility of the participant responses from the GCSS confirms that telemedicine genetic counseling is an acceptable service delivery model for this indication.

2.4.1 Study Limitations

Limitations to this study include small sample size and response bias. Throughout the four-month study period, all patients that were seen for pre-test genetic counseling for a retinal dystrophy indication. Given that retinal dystrophies are rare, we did not expect to see the same volume of patients as has been published in similar studies conducted in oncology and prenatal settings; however, collecting this data for a larger sample size over a longer duration would be beneficial. In addition, this study was conducted at a single center in an urban setting and therefore may not be generalizable to all retinal dystrophy clinics.

The survey was conducted entirely by phone, and while anonymous, there is potential for acquiescence. Given the structure of the GCSS, a participant may respond to the Likert scale in a way by which they believe is desired. This would not reflect their true experience or feelings toward their genetic counseling appointment and may be difficult to discern. As with any study involving a survey, there is also concern for non-response bias. All of the individuals that did not answer the phone, did not return the voice message. It is possible that non-respondents did not have a high level of satisfaction with genetic counseling and or their service delivery model.

The survey participants consisted only of individuals that had pre-test genetic counseling, individuals that had post-test counseling were not contacted. The post-test counseling typically

involves the disclosure of genetic testing results. When contacted to schedule a post-test appointment, patients are given the option to schedule with their preferred service delivery model (similar to pre-test). We did not examine the post-test data. This could have provided a more complete picture of uptake with all service delivery models.

2.4.2 Future Directions

Telemedicine genetic counseling will continue to exist as a service model. While our data helps to inform the uptake in service delivery models among patients seen for this indication, there is additional data that could be collected on next steps in the genetic counseling process. It would be interesting to gain an understanding of the turnaround time from initial appointment to when results are received from the laboratory. The patients that are seen in-clinic have their sample collected during the appointment as opposed to those that have telemedicine appointments and are responsible for collecting and shipping their own sample. This data could also provide information on adherence and follow-through with the plan that was agreed upon during the genetic counseling session. It may also be valuable to address the patient's feeling towards their genetic counseling service delivery model by asking questions direct toward their appointment type. For example, discussing if the patient would choose to have this genetic counseling service delivery model again and if it is something they would recommend to a different individual. This would provide information beyond what the GCSS is able to capture. Further research is needed to expand our knowledge of uptake and satisfaction with genetic counseling service delivery models for inherited retinal dystrophies in the pediatric population.

2.5 Conclusion

Access to genetic counselors and testing is especially valuable for patients with inherited retinal dystrophies due to the rapid advancements in gene therapy trials and approved treatments. Without access to a genetic testing, identification of patients who may be eligible for these therapies would not be possible. Genetic counseling service delivery models are expanding beyond the traditional in-person appointment to improve patient access, including telemedicine; however, it is important for providers to be confident that these novel service delivery models do not compromise the level of care. Our study found that patients with suspected retinal dystrophy have varied preferences for genetic counseling service delivery model. However, across all four service delivery models included in this study, participants were highly satisfied with their genetic counseling appointment. Collectively, the results suggest that telemedicine is a satisfactory service delivery model when compared to the traditional in-person genetic counseling. Telemedicine and in-person appointments for genetic counseling in adults with inherited retinal dystrophies should continue to be offered to all patients. Providers should attempt to ensure patients with legal blindness and other disabilities have the assistance they need to access their preferred appointment type.

3.0 Research Significance to Genetic Counseling and Public Health

Genetic counseling as a field has made significant contributions to public health (Khoury et al., 2022). This study is focused on quality improvement through building an understanding of patient access to and satisfaction with genetic counseling in adults with inherited retinal dystrophies. The various aspects of the three core functions of public health; assessment, policy development and assurance, align with the aims of this study (CDC, 2020). Specifically, the pillar of assurance as it applies to actively working to ensure communities remain healthy through access to innovative and equitable healthcare. The results of this study will contribute to the understanding and utility of the service delivery models that provide access to genetic counseling for patients with retinal dystrophies.

A key public health function is to improve and innovate healthcare by evaluating the processes that are in place. While telemedicine has long been an option in many medical settings, it has become more readily available due to the onset of the pandemic in 2020 (Uhlmann et al., 2021). Hospital systems have established secure video platforms to provide remote services, including genetic counseling. A strong infrastructure has been built to support the growth of telemedicine services as they become more popular across various specialties. Given that the foundation for telemedicine genetic counseling appointments is in place at UPMC, it is essential to understand if patients are equally satisfied with this type of appointment in comparison to the traditional in-person genetic counseling appointment. This study recognized that patients are highly satisfied across all service delivery models, proving that there is value in telemedicine genetic counseling and that it should continue to be a mainstay service delivery model.

Telemedicine comes with both benefits and limitations to equitable access. Patients that live in remote locations, are unable to drive or have difficulty finding transportation to an appointment benefit greatly from telemedicine and having the ability to meet with their providers from home. Unfortunately, telemedicine is also limiting, as many individuals do not have access to or know how to use a device necessary for a telemedicine appointment. The study showed that telemedicine video and phone appointments were valuable service delivery models, but that the option for in-person appointments cannot be eliminated. It is also important to note that just because an individual has access to and is comfortable using a device capable of a telemedicine appointment, it does not mean that telemedicine is their preferred appointment type. In addition, many individuals with retinal dystrophies meet/will meet the medical criteria for legal blindness. This added challenge gave further reason to explore patient satisfaction with genetic counseling delivery models. A majority of the patients that met criteria for legal blindness and completed the satisfaction survey stated that they do not require assistance to attend their appointments. This shows that telemedicine is both accessible and acceptable for some individuals with legal blindness.

An essential service of public health is the ability to communicate effectively to provide information and education (CDC, 2020). When offering patients the option to schedule in-person or telemedicine genetic counseling appointments, it is important that the patient has realistic expectations as to what each appointment type involves. This includes having an understanding of travel distance/parking versus how to navigate the secure video platform and knowing a saliva sample will be collected and managed by a provider in-person versus having to collect and submit the sample and documents independently. While data on sample collection and patient follow-

through with testing after telemedicine genetic counseling appointments was not collected for this study, it is a future direction to further comprehend the utility of telemedicine genetic counseling.

Public health involves the protection and promotion of health in all communities. The goal of this study was to understand patient uptake with genetic counseling appointments in-coordination with another appointment, in-person genetic counseling only and telemedicine genetic counseling only through secure video platform or phone call. The results showed that patients chose appointments in each of the service delivery models, suggesting that patients value having different appointment options available. This data confirms the importance in offering genetic counseling through multiple modalities, protecting patients' access to care. It was also found that respondents were satisfied with all service delivery models. Knowing that patients are having positive experiences with genetic counseling regardless of the appointment type is valuable in that it allows for the promotion of multiple service delivery models in adults with retinal dystrophies.

Appendix A Appendices and Supplemental Content

Appendix A.1 Tables and Figures

Appendix Table 1. Survey

Genetic Counseling Satisfaction Scale					
	Strongly Disagree	Disagree	Neither agree nor disagree	Agree	Strongly agree
My genetic counselor seemed to understand the stresses I was facing	1	2	3	4	5
My genetic counselor helped me to identify what I needed to know to make decisions	1	2	3	4	5
I felt better about my health after meeting with my genetic counselor	1	2	3	4	5
The genetic counseling session was about the right length of time	1	2	3	4	5
The genetic counselor was truly concerned about my well-being	1	2	3	4	5
The genetic counseling session was valuable to me	1	2	3	4	5
Logistics					
Did you have access to resources needed for telemedicine appointment	Yes	No			
Did you rely on any assistance to complete your appointment?	Yes	No			
What influenced/would influence your decision when scheduling an in-person vs telemedicine genetic counseling appointment?	Open response				

Appendix Table 2. GCSS Satsisfaction Score per Question by Appointment Type

	IP-C (n=5)	IP-GC (n=2)	TM-V (n=10)	TM-P (n=1)
My genetic counselor seemed to understand the stresses I was facing	4.6 (0.55)	5	4.8 (0.42)	5
My genetic counselor helped me to identify what I needed to know to make decisions	4.2 (0.84)	5	4.9 (0.32)	5
I felt better about my health about meeting with my genetic counselor	3.6 (0.55)	5	4.6 (0.7)	5
The genetic counseling session was about the right length of time	4.4 (0.55)	5	4.6 (0.7)	5
The genetic counselor was truly concerned about my well-being	4.8 (0.45)	5	5	5
The genetic counseling session was valuable to me	4.8 (0.45)	5	4.9 (0.32)	5

Appendix Table 3. GCSS Total Satisfaction Score and Legal Blindness

Appointment Type	GCSS Total Score - NLB	GCSS Total Score - LB
IP-C	28	25
	27	25
	27	
	27.3 (0.58)	25
TM-V	30	30
	28	30
	28	30
	27	29
		28
		27
	28.25 (1.26)	29 (1.26)

NLB – Did non meet criteria for legal blindness, LB – met criteria for legal blindness

Appendix A.2 Quality Improvement Study Approval

The Quality Improvement Review Committee is pleased to inform you that your QI project has been approved.

We have also notified your local quality department of this approval and encourage you to share updates on the project's progress.

Please note that results of QI projects must be reviewed by local quality directors and approved by the Chief Quality Officer prior to dissemination (via presentation or publication) outside of UPMC. UPMC has adopted the Standards for Quality Improvement Reporting Excellence guidelines, SQUIRE 2.0 as the suggested reporting format.

For multi-center projects, the QRC **approval** refers only to that **part of the project being performed at UPMC facilities** and the sponsors are responsible for obtaining approval from other non UPMC facilities participating in the project.

We suggest that you share your findings on this project with the QRC. When your project is complete, please navigate to the Quality Improvement Project Portal and go to "My Projects." Select the project and go to the "Project Summary" tab, add the findings in the "Project Results" field, and click "Submit Project Results to QRC."

Projects reviewed and approved by the UPMC Quality Improvement Review Committee do not meet the federal definition of research according to 45 CFR 46.102(l) and do not require additional IRB oversight.

Project Submission Details:

Project ID: 4046

Project Title: Patient uptake and satisfaction with genetic counseling service delivery models in adults with retinal dystrophies

Project Sponsor:

Michelle Alabek ****** Genetic Counselor II ****** POP14 EYE Cln Main Office

Project Co-Sponsor(s):

Jose Sahel ****** Chairperson ****** UPP14 EYE Cln Main Office

Andrew Williams ** Faculty - Clinician, Physician ** UPP14 EYE Cln Main Office
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Submitted By:

Michelle Alabek ** Genetic Counselor III ** POP14 EYE Cln Main Office

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