**Barriers to Access, and Time to Genetic Testing in the**

**Provincial Medical Genetics Program**

**Newfoundland and Labrador**

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**Abstract**

**Objective**: To determine the wait times for service for hereditary cancers and cardiac disease, as well as the barriers that exist that impact wait time.

**Background**: Access to genetic testing and services is currently largely restricted for practitioners outside of the medical genetics specialty within Newfoundland and Labrador. The current practice may delay timely access to care that is required to diagnose and develop a plan of care.

**Methods:** Data was extracted from 78 patient referrals received in the Provincial Medical Genetics Department (PMGP) from January 1 through December 31, 2013. Charts were evaluated to establish the timelines for access to services, with an examination of the timelines for each phase of the process. Semi-structured interviews with nine key stakeholders were completed to gain an understanding of their role in the patient pathway, as well as the expectation of the service.

**Results**: There is a non-significant difference in the median wait time to receive service between cancer and cardiac referrals with a wait-time of 45.7 months (95% CI 41.8 – 49.5) for cancer versus 36.5 months (95% CI 22.7 – 50.3) for cardiac referrals. Of the 78 patients in the sample, 9 (12%) did not require an appointment but remained in the queue, and 11 (14%) were a "no show" or refused an appointment. The referral triage process did not impact time to receipt of services, with a median wait time of 43.6 months (95% CI, 38.9 – 51.3) for referrals that were triaged and 45.7 months (95% CI 40.9 – 50.4) for those not triaged. To assist with the triage process additional information was required for 33 of the 78 referrals, 17 (51.5%) patients returned this information, while 16 (48.5%) did not. Wait time for service was not impacted, as all referrals received an appointment. There was no significant difference in wait times between internal or external referrals, median wait time 45.7 months(95% CI, 38.2 – 53.1) for internal referrals, versus 44.1 months (95% CI, 36.9 – 51.3), however, referrals from families had a wait time of 3.4 months (95% CI, .18 – 6.6). Qualitative analysis of semi-structured interviews that were

completed with nine health care providers suggests that the major barriers for access were the current referral process, prolonged wait times, lack of referral appropriateness knowledge, and the inability of the provider to order baseline genetic testing.

**Conclusion:** Genetic counseling and treatment are complex processes. The current referral process is prolonged. Survey respondents indicate a low satisfaction, and frustration with the disjointed process. A streamlined standard process would enable the PMGP to better control the flow, to ensure time to access and service is decreased for the highest priority patients.

**General Summary**

This study aimed to understand the barriers to access genetic testing within Newfoundland and Labrador. The current practice of accessing genetic testing requires patients to be assessed and counseled by a clinical medical geneticist or a genetics counselor prior to any test being offered. This potentially causes significant delays in care, increased anxiety while the patient waits, as well a significant backlog within the health care system. Wait times for access to the services range from 6 months to 3 years, (excluding emergent referrals) followed by a further delay if a specialist appointment is necessary. Currently, there is minimal published research documenting barriers to access specifically to Genetic Counsellors. This research project will focus on access to two specialized services within medical genetics; cardiac and hereditary Cancers. Upon the completion of this study, it was noted that the medical genetics department in Eastern Health had improved the referral and intake process that decreased overall wait times.

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This thesis is dedicated to my parents, Jack and Lorraine Dooley who have always provided me with endless support in whatever I choose to do, especially my academic endeavors. To my husband Dennis, and sons Nicholas, and Christopher. Thank you for your patience and ability to wait while I was reading, writing, and attending class, thanks for taking on the extra. I couldn't have done this without you.

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**List of Tables**

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**List of Abbreviations**

Canadian Foundation for Healthcare Improvement .....CFHI

Canadian Institute of Health Information .....CIHI

Canadian Medical Association ..... CMA

Canadian Medical Protective Association .....CMPA

Cardiac Genetics Clinic.....CGC

Centre for Health Informatics and Analytics ..... CHIA

Direct to consumer.....DTC

Eastern Health.....EH

Excerpta Medica dataBASE .....EMBASE

Genetics Counsellor.....GC

Health Care Practitioner.....HCP

Health Research Ethics Board ..... HREB

Medical Practitioner.....MP

Memorial University of Newfoundland .....MUN

National Society of Genetic Counselors.....NSGC

Newfoundland and Labrador ..... NL

Newfoundland and Labrador Centre for Health Information ..... NLCHI

Provincial Medical Genetics Department.....PMGP

Regional Health Authority ..... RHA

**LIST OF APPENDICES**

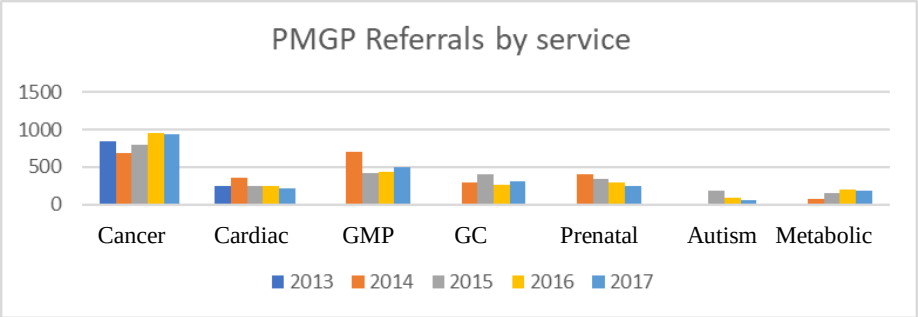
# Chapter 1 Introduction

## 1.1 Background and Rationale

Access to genetic testing is currently largely restricted for practitioners outside of the medical genetics specialty. Within Newfoundland and Labrador, the current practice for accessing genetic testing requires patients to be assessed and counseled by a clinical medical geneticist or a genetic counselor before any testing is offered. The exception to this was implemented in 2017 for breast cancer patients, as the medical oncologist can order initial testing sequences to expedite the treatment while waiting for the patient's appointment with the Provincial Medical Genetic Department (PMGP). The current referral process is paper-based and manual, potentially causing considerable delays in care, increased anxiety for the patient as they wait, as well as backlog within the health care system. It can also create health care issues for those “at-risk” family members of the proband. Data retrieved from the PMGP for the calendar year 2013 indicate that there were 2,479 referrals received. It is estimated that the wait times for access to the genetic counselors range from 6 months to 3 years, (excluding emergent referrals) followed by a further delay if a specialist appointment is necessary. There are minimal published research documenting barriers to access to genetic counselors. The number of overall referrals received within the PMGP has steadily risen by nine to 10 percent year over year. Figure one displays the referrals received by disease presentation. Referrals for hereditary cancer continue to be the most requested service. The number of cardiac referrals has fluctuated year to year, but the wait time remains long. This proposed research project will focus on access to these two specialized services within PMGP with increasing referrals numbers and wait times; Hereditary Cancers and Cardiac Disease.

**Figure 1**:

*Referrals received within PMGP and logged, identified by service for 5 years. (2013 – 2017)*




Note: GMP – Genetics Medical Practitioner / GC Genetics Counsellor

### 1.2 Current Knowledge

With the increasing complexity of the genetic patient population, related to the growing genetic testing technology, the uncertainty in the processes required and needs for counseling patients makes it difficult to manage patient access. The actual current wait times to access genetic services are not known and vary depending on many factors, including human resources, population, service area, internal and organizational processes, individual practice and experience, patient engagement, and referring physician engagement. Inconsistent data collection methods, lack of clear standard processes, and unclear roles and responsibilities may have impacted the referral flow process. As referrals accumulate (see figure 1)and patient wait times appear to grow, it becomes a seemingly daunting task to overcome. To fix access we must first understand the barriers and the etiology of the processes.

### 1.3 Research Questions

1. What is the current overall wait time, and the wait time for each phase in the process for access to genetics services?
2. What are the clinical and non-clinical barriers to access for genetic counselors and geneticists?

# Chapter 2 – Literature Review

## 2.1 Introduction

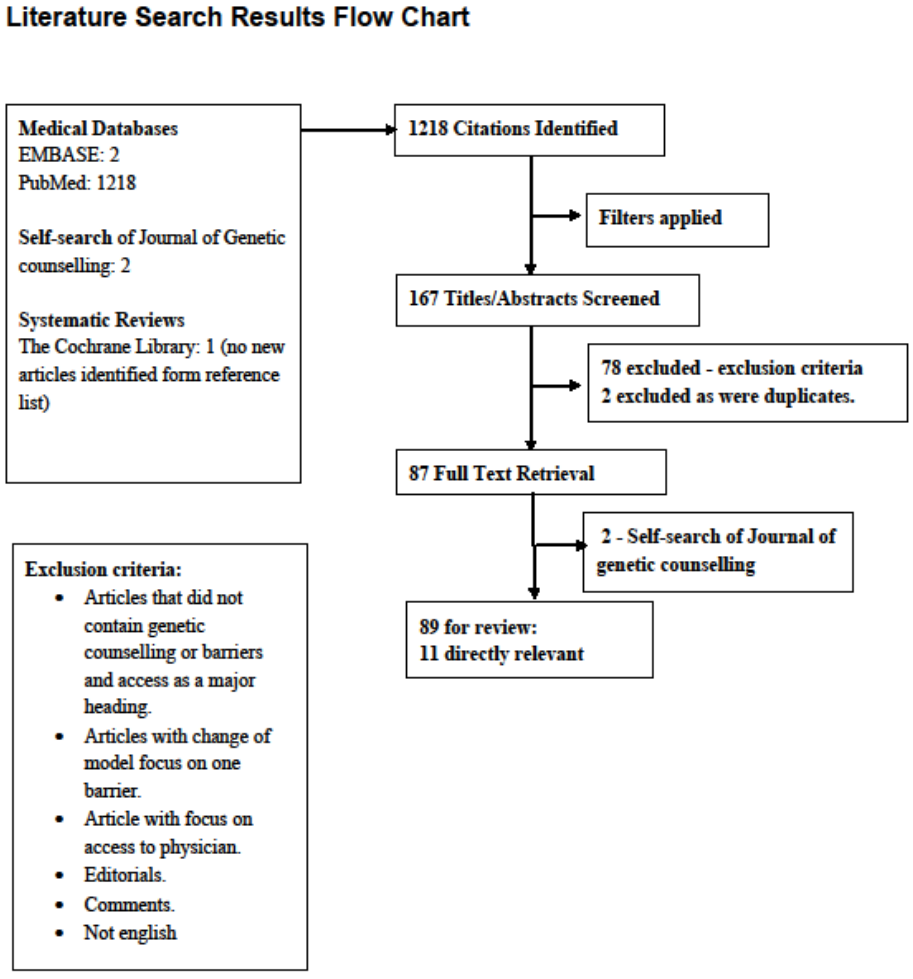
This literature review examined clinical literature related to the barriers and time to access genetic counseling services and genetics testing services, as well as access and barriers to general medical services. The clinical literature has revealed several gaps in current knowledge related to access to genetics services, specifically with limited available research related to access to genetics counseling services.

## 2.2 Search Strategy

Publications were located through searches in PubMed, EMBASE, and Google Scholar. A combination of terms: Genetic Counselling, genetic counselors, wait List, barriers, access to health care, availability of health care, model of care yielded 36,958 articles (see Figure 2). These articles were very broad in topic, language, and scope. Further search terms were applied: health knowledge, attitudes, practice, health service accessibility, genetic testing, accessible health care, efficiency, and appropriateness. Filters were applied for English only and time frame 2010 to 2019. 1218 articles were returned. Further filters were applied with the addition of the search terms “Newfoundland OR Labrador OR Canada OR Canadian” yielded a return of 89 articles, with 11 being most relevant.

**Figure 2:**

*Diagram of the search strategy used to identify peer-reviewed research related to the topic barriers for care and access to genetics care.*




## 2.3 Genetics

The study of genetics is the study of our heredity, of genes and factors related to all aspects of genes (Durmaz, 2015). In the mid-19th century, Gregor Mendel discovered the basic principles of "heredity and laid the mathematical foundation of the science of genetics" (Durmaz, 2015).

The study of and understanding of genetics has historically seemed foreign to the average person. With the advancing technology and popularity of social media, there has been an increasing

interest in genes, genetics, and the impact on human connections and health. Referrals for genetic services increase when celebrities tell their story of making health decisions using their genetic information. A study by (Raphael, 2016) demonstrated a 90% increase in referrals for BRCA 1 gene mutation, and more specifically for genetic counseling 6 months after Angelina Jolie shared that she had decided to proactively have a double mastectomy after learning she carried a mutation in the BRCA 1 gene in 2013.

The need to access genetic testing can be time-sensitive. A genetic predisposition is the increased likelihood of developing a certain disease based on an individual's genetic makeup. Does this mean they will develop the disease; this is the unknown, and the reason why timely access to testing and counseling is so important. Knowledge of predisposition to a disease can help individuals control other contributing factors that may also increase the risk of developing the disease.

Recent scientific advances have enabled direct-to-consumer (DTC) companies to offer the ability to realize personal genetic information, without leaving home. Individuals can now discover where they came from, but also if the family history has evidence of hereditary disease (Radford, 2014) or gather information related to their genetic make-up for their health (Peterson, 2018). It is important to note that DTC genetic testing is not comprehensive. The evolution of DTC genetics has motivated consumers to need to know more about their genetic makeup, and the request for further genetic testing has been impacted. Requesting genetic testing and follow-up does not require the support of a health care provider. Individuals can request the service themselves, based on confirmed family history, as a follow-up from home testing or sometimes for the sheer purpose of knowing. However, knowing one's genetic makeup, and understanding the medical implications are two very different issues. So, in addition to an increase in requests for testing, there is also an increased need for counseling. Genetic counselors play a critical role in understanding genetic information. (Battista, 2011; Riley, 2012) and help individuals make

health care and lifestyle decisions. A delay in access to testing and/or counseling can increase the risk of disease progression and have a negative impact on health outcomes.

With an increase in referrals for all aspects of genetic care being experienced, it has become even more important to have a robust evidence-based referral process in place.

Genetics has been often misunderstood, villainized in mainstream media, or romanticized by the potential ability to pick and choose designer babies, genetically modified to filter out the weak, "thus flying in the face of survival of the fittest" (Savulescu, 2006). Cultural beliefs and fear of losing one's privacy also impact the attitudes toward the benefit and utility of genetic testing (Etchegary, 2014). For this study, DTC genetics is not included, but it needs to be acknowledged as an impact on the number of referrals as well as a potential barrier as this may confuse the average person when faced with a medical decision of "do you want genetic testing." (Molster, 2009; Henneman, 2013; Haga, 2014).

**2.4 Genetics Counsellors**

Genetic counselors (GC) work with patients, families, and other healthcare providers to advise on the availability of testing based on medical concerns and the impact genetic testing may have on their health, family, and future planning when a patient decides to proceed with testing. (Andrews LB, 1994). Genetic "counseling should provide the patient with information about the testing process, medical management of the disorder, and possible impact of the test results on patients and their families." (Andrews LB, 1994). The clinician must ensure that proper explanation of both the pre-test information, testing results, and impacts occur (Lu, 2014; Riley, 2012). Genetic counseling is available in most countries; however, the access and provision of the service vary greatly depending on funding and service delivery models. (Beene-Harris, 2007; Trepanier, 2014). The ability to access genetic service is impacted by the referral pattern or service delivery model. Within EH the service delivery is a combination of the GC seeing and assessing the patient independently, and with the medical genetics

physician (MGP) if required. The referral pattern to EH's medical genetics service has not been previously studied. Currently, the process for a referral is nonstandard and varies by specialty. Further review is required to identify the appropriateness of referrals and establish a process for follow-up.

Three types of service provision arose from the literature. First, provision of counseling service without a physician, second, provision of counseling service with a physician, and third, providing service using a combination of both approaches. Wham (2010) studied the models of service provision and found that half of the studies "respondents (50.3%) indicated providing service independent of a physician, 22% of respondents indicated providing service with a physician" (Wham, 2010). In 2012, (Hannig, 2014), reviewed a service delivery model for GCs only, indicating that 321 patients were seen in a 24-month period, where 61% (n=194) of those seen did not require further follow-up, and 10% (n=32) were sent for a medical genetics physician (MGP) appointment. The ability of the GC to assess, counsel, and establish a treatment plan for follow-up allowed improved access to the MGP, allowing care to happen at the appropriate level of service and decreasing the overall wait-time.

A previous study by Cohen (2013) (using the 2010 *Professional Status Survey for the National Society of Genetic Counselors (NSCG)*), focused on the service delivery models either in person, by phone, in a group, or telemedicine. The patient referral is matched with the best delivery model, being mindful of geographic location, and cost. Of the respondents, 65.3% indicated they always book appointments face-to-face, and 30.4% often book appointments face-to-face. Other studies, from a patient's perspective, reported low uptake of new emerging models of care, such as telemedicine and group sessions, highlighting the importance of ensuring individual patient needs should be included when deciding on a pathway. (McDonald, 2014; Pieterse, 2007)

The vast majority of the GCs used more than one service model, (Cohen, 2013) in-person or face-to-face appointments were the most popular service models. The use of the telephone or telehealth methods was supported by the GC groups, and uptake, as expected, was greater for patients

living further away from the point of care provision. It was also noted that time spent with patients using these service models was significantly shorter than face-to-face visits. The requirements and associated costs for new technology may contribute to the low or slow uptake of the telehealth service model (Wham, 2010). A large proportion of respondents of both Cohen (2013) and Wham (2010) indicated they would use telephone appointments for disclosing patient results, with a face-to-face follow-up appointment to discuss outcomes, findings, and future planning. A small number of respondents indicated they book a face-to-face for all aspects of the appointment regardless of the test results.

The third model was providing service using a combination of both approaches. This model was used when the initial booked appointment with a GC required more in-depth follow-up with the MGP, a new diagnosis that required follow-up, or at the request of the patient/family. (Hannig, 2014)

**2.5 Wait Time**

Acknowledging prolonged wait times within EH, it was important to identify processes that may lead to delays and explore potential improvements. The literature did not uncover any standard access benchmark times for genetic-based appointments. A study by Knapke S, (2015) based on the 2014 *Professional Status Survey for the National Society of Genetic Counselors (NSCG)*, identified that urgent access to genetic counselors (GC) is meeting established benchmarks: “46% of genetic counsellors indicated they could accommodate a new patient appointment within a week, with 27% reporting the ability to provide service within 1-3 days” (Knapke S, 2015; National Cancer Institute, 2018). These benchmarks were based on local information and past performance within the individual departments. There was no evidence found identifying a universal, evidence-based benchmark, or standards for access times for genetics services, testing, and counseling.

A component of the process of care is preparing for the initial appointment. This takes time for the GC to review the provided information, piece together the patient’s medical story, complete the

pedigree, research the diagnosis and establish a plan of care. The literature suggested this time varies with the service model and the requirements of the organization. Wham (2010) concluded that the time for case preparation ranged from 30 – 90 minutes, with McPherson et all (2008) identifying an average of seven hours for each new patient appointment. “Time spent on risk assessments, case preparation and follow up were all less than recommended standards and varied with the practice” (Nelson HD, 2019).

It is suggested that adequate human resource relieves the pressure of access on the system by balancing demand and capacity (Kabene, 2006). However, before human resources can be identified as the main barrier, standardized processes within the organization must be developed, implemented, and evaluated (Kabene,2006; Liker, 2011).

Minimal literature was uncovered related to the consistent triage process for genetics referrals. The need to triage patients for care is not uncommon but is generally seen in an emergent care area, or a service that is under capacity and over demand. The need for an appropriate standardized triage process must consider the ability to flow patients through the system and not increase the wait time for non-urgent referrals while ensuring the most urgent patient gains access to service as quickly as possible (McVeigh, 2019).

**2.6 Barriers**

An unexpected theme that emerged from the literature was practitioner reluctance to refer (Delikurt, 2015). Several studies indicated barriers with uptake for genetic services including the often-perceived siloed approach (Mikat-Stevens, 2015), lack of inclusion and understanding of genetic care (Starfield, 2005; Kegley, 2003), timeliness of information (Watson, 1999), and lack of referral coordination of care (Greenwood-Lee, 2018). The limited ability for a physician to order the initial test, independent of the genetics service, was also noted as a barrier (Prochniak, 2012) . The authors indicate a large proportion of responders felt confident in ordering and providing appropriate follow-up care to

their patients, with 41.6% of the physician's stating they prefer to order their testing; however, they do not feel confident providing emotional follow-up, as "they lacked the requisite expertise to counsel patients about genetic testing" (Hallowell, 2019; Andrews LB, 1994). Staying current is difficult for general practitioners, with the rapidly changing availability of genetics testing, and as Prochniak (2012) found, only 18.6 % of study participants reported receiving formal genetics training in medical school, residency, or fellowship. The authors also note that in a previous study by Brandt (2008), 73% of physician respondents indicated that the biggest motivator for referring to genetics is patient request, with 54 % of physicians indicating the biggest motivator for not referring is patients' lack of interest.

Other barriers to referral were noted to be lack of awareness of the program (Brandt, 2008; Delikurt, 2015; Prochniak, 2012), patients insurance coverage (Brandt, 2008; Prochniak, 2012), and potential discrimination toward the patient for future job and health insurance (Prochniak, 2012; Radford, 2014).

There is the suggestion that there is a connection between the low rate of referrals and the health care providers' perceptions of the relevance of genetics to their practice exist (Mountcastle-Shah, 2000). The availability of up-to-date accurate risk assessments was noted as a potential for inaccurate initial referral (Mikat-Stevens, 2015; Riley, 2012), and more available support is needed for the PHC physicians to improve their comfort level of utilizing genetics in their practice (Greiner AC, 2003).

**2.7 Genetics in NL**

Historically, NL has been an attractive locale for genetics research due to the small gene pool and the ability to trace back many generations. "The population of Newfoundland and Labrador is genetically isolated" (Rahman, 2003), and serves as an excellent resource for identifying genes implicated in common diseases (Zhai, 2016 ). A founder effect has been observed in Newfoundland for many genetic disorders, including a variety of hereditary cancers and cardiac diseases. "Almost 90% of the current inhabitants of Newfoundland and Labrador are descended from immigrants, mostly English

and Irish, who settled here before the mid-19th century" (Young & Leonard, 1999). Due to this unique history, considerable interest exists in Newfoundland and Labrador as a location for human genetics research.

In Newfoundland and Labrador, medical genetics services are based in St. John's, with two outreach clinics; one in the central area and the second on the west coast of the province which is staffed by Registered Nurses with genetic counseling education. Since the completion of this study, both of these positions have been retired and have not been replaced, all patients are referred to the St. John's site. There is no genetics program staff present in the Labrador region. The practice of a genetics counselor offering assessment and support before testing is offered may impede the referral flow, but as demonstrated in the literature it is a practice that supports patient safety and quality of care (Riley, 2012). Providing evidence-based information before, during, and after the offering of test results supports the patient with decision-making throughout the process.

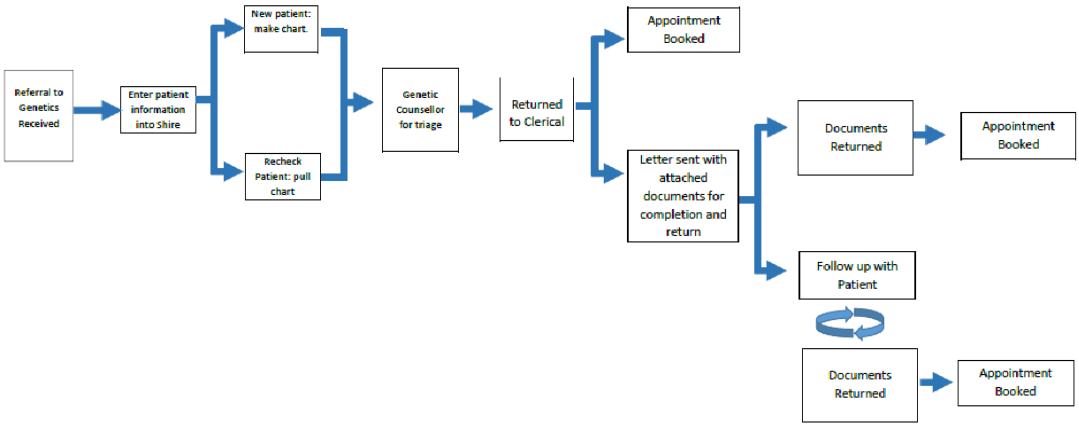
# Chapter 3 – Methodology

## 3.1 Methods

This research was conducted using a retrospective cohort study design. A standardized data collection tool was developed and used to collect the following: referral date, date referral received in PMGP, date referral sent for triage, date referral triaged and assigned triage code, date of appointment, service requested, gender, age, geographic location, appointment attended status (attended, cancel or no show), refer to table one. These data points were extracted from EH's patient information system, Meditech, the Provincial Medical Genetics Shire database, and patient paper charts located at the PMGP. The de-identified dataset and the manually retrieved data were formatted in a .xlsx format (Excel software). The master file of de-identified data included the entire population. Standard security practices established by EH and Memorial University of Newfoundland were followed to ensure data integrity, confidentiality and that the data was stored securely.

**Figure 3:**

*High-level flow diagram for referral to PMGP.*




*Note:* The time for each step in the process is impacted by several factors, and is not standard.

Once a referral is received in the PMGP, it is processed and entered into the Shire database. The referral is then assigned to the genetics counselor who is responsible for triage duties for that current period. The process for referrals is standard as depicted in Figure 3, but individual practice and lack of standard documentation process hider the flow.

The wait time data from point of service request (referral date) to service provision, suggested that 2479 patients were waiting for first access to genetic service. Completion of a standard patient pathway identified all the major steps for the referral as it progresses through the PMGP e.g.: referral date, time to triage, time to appointment attended. This flow process provided the variables required for the study, see Table 1. Referrals are received from both internal i.e., within the EH system, and external i.e., from outside of the EH system.

These referral types are categorized by:

1. Consult sheet, internal health care providers,
2. Doctor letter, which would be from the community or outside of EH, and
3. Family connections of a previous patient.

**Table 1:**

*List of variables included in the study which were retrieved from charts, Meditech, and Shire database.*

| Variable | Example |
|---|---|
| Appointment date | (dd/mm/yyyy) |
| Service requested | Hereditary cancer or Cardiac |
| Gender | M or F |
| Age | Years |
| Referral Resource | Specialist/GP/NP other |
| Referral Date | (dd/mm/yyyy) |
| Referral Received Date | (dd/mm/yyyy) |
| Triage Status | routine/semi-urgent/urgent |
| Triage Date | (dd/mm/yyyy) |
| Information Required/ requested | Yes or No |
| Final Documents Received | (dd/mm/yyyy) |
| Attended | Yes or No |

*Note*: Due to inconsistent charting practices, not all variables were available in the charts included in the study.

**3.2 Sample Size**

For this research study, an exploratory approach was taken to identify trends and barriers. The data collection required manual chart reviews and getting data from external sources. The eligible population for this chart review was all referrals for Hereditary Cancer and Cardiac genetic services received within the PMGP with a referral date between January 1, 2013, and December 31, 2013. As the focus was on the adult population, referrals received for patients under 18 were excluded.

A systematic sampling process was established where every 8$^{th}$ chart was identified for further review and study inclusion. A total of 123 charts were identified for manual review. Due to a large number of referrals and allowing for a manageable number of referrals, upon consultation with a statistician, it was decided to review 10% of the total referrals from each subgroup (hereditary cancer and cardiac) over 1 year (2013) which was 120 charts.

After the initial electronic file review, 45 charts were excluded: 19 referrals had incorrect diagnoses and did not require genetics follow-up, nine referrals were for follow-up by the clinics outside of St. John's where a different process for referrals exist, eight referrals for under 18 were excluded as the focus of this study was on the adult population, nine referrals did not have sufficient clinical information to be included in the study. A total of 78 charts were identified for in-depth chart review (44 Cancer/34 Cardiac). Although the number of charts reviewed was less than initially identified, it was felt to be an appropriate number as themes were recurring.

A semi-structured interview (Appendix B), was completed with health care practitioners (HCP) to gather information regarding the referral process. The invite (Appendix C), was sent to 21 HCP who were identified by referral source and included those who had referred more than three patients to the PGMD from 2013 – 2014 for either cancer or cardiac services. The initial group consisted of 16 specialists and five general practitioners. Of the nine respondents, five were specialists, three were

general practitioners, and one chose not to identify their profession. There was representation from across the province with six participants from Eastern Health, two from Western Health, and one from Central Health.

**3.3 Procedure**

Following submission and approval by the Health Research Ethics Committee (HREB) (Appendix A), a mortality report was requested from Newfoundland and Labrador Center for Health Information (NLCHI), which was used to validate deaths associated with the available waitlist, these events were censored at the time identified. An initial review of the charts was completed to confirm the referral date, that an appointment had not been scheduled, and that the patient was not discharged from the PMGP care. For this thesis, the sample size is 78.

Using the developed standardized collection tool, the paper chart was reviewed, and the relevant information was collected. Eastern Health's Meditech system and the PMGP Shire database were used to cross-reference the data, and to locate missing information and confirm dates (i.e., referral, received, triaged, appointment if booked). This data was entered into the prepared spreadsheet for further analysis.

The semi-structured survey consisting of standard questions was developed and used to guide the interview (See appendix B). I was responsible for the consent, (Appendix D) data collection, and analysis of the nine interviews and discussions. The study overview was provided to the participant ahead of the interview so they could review and familiarize themselves with the study objectives. Participants were informed that written notes would be taken during the interview and used in the report, and direct quotes could be included. Once consent was received, the interview began and lasted approximately 30 mins. At the end of the interview, participants were allowed to add additional

information that may not have been discussed during the formal interview or to elaborate on further points.

### 3.4 Outcome Measures

The primary outcome for this study was to measure the time from the referral date to the service provision date. To confirm this, the chart required information for two specific date points 1) Referral date 2) Service provision date (appointment). Factors that may influence the outcomes were also assessed, including the triage status, the need for additional information, and the turnaround time for the information requested from the patient and /or family.

#### *3.4.1 Thematic Analysis*

The qualitative component of this project included a semi-structured interview with key stakeholders, (referring practitioners, genetic counselors, geneticists, decision-makers, and leadership), to provide a better understanding of their role in the patient pathway, as well as the expectation of the service. Thematic analysis is provided, drawing on information from the data and the semi-structured interviews

For the survey portion of the study, potential participants (physicians and other health care practitioners) An email (See appendix C) was sent by the PGMP program on my behalf, inviting them to take part in the face-to-face interview. If the invitee was interested in participating, they were to contact me via email, and a date and time to meet were agreed upon.

Nine of the 31 invited participants that had been invited agreed to take part in the semi-structured interview. Transcripts from the interviews were reviewed and used to formulate a thematic analysis. Thematic analysis was selected as it offers an accessible and theoretically flexible approach to analyzing and generating themes from qualitative data (Braun, 2006). The information collected from these interviews identified barriers and facilitators to access from the referring HCP perspective.

### *3.4.2 Ethical Issues*

Usage of all data held by NLCHI, and Eastern Health's data system followed strict ethical guidelines for de-identifying data, no patient identifiers were included. Research ethics approval from the Health Research Ethics Board of the Memorial University of Newfoundland was obtained before beginning this project. The project lead is also certified in the Tri-Council Policy 2 Statement of conducting research in human populations

### *3.4.3 Statistical Analysis*

Data was entered into excel spreadsheets and statistical analysis was performed using Statistical Package for Social Science (SPSS) software version 24 (IBM, Chicago, IL, USA).

### *3.4.4 Time to Event*

Kaplan Meier time to event analysis was used to calculate the time to the event (appointment). This is unique for each referral, based on the date of the individual referral and the date of appointment

### *3.4.5 Time to Event by Service*

Referrals included for review are from one of two services, either hereditary cancer or cardiac disease. Kaplan Meier time to event analysis was used to calculate the time to the event (appointment) by the requested service.

### *3.4.6 Time from Referral to Triage*

Analysis of the length of time required to triage a referral was evaluated using the Kaplan-Meier method, with further analysis of the assigned triage status' impact on wait time.

### *3.4.7 Time to Event by Referral type*

As the PMGP is a provincial program, referrals are received from both within EH, or externally from community HCP or other Regional Health Authorities (RHA). Referrals are also received from family

members of a patient with a diagnosed genetic condition, requesting assessment and testing. Analysis of the time to the event, based on referral source, using the Kaplan-Meier to assess if there is a difference in wait time between these two groups.

### *3.4.8 Baseline Demographics*

Descriptive statistics were used to create a profile of patients by gender, age, service, and method of request. Normality of distribution was checked for all variables using Kolmogorov-Smirnov and Shapiro-Wilk's tests and homogeneity of variance ($p < 0.05$) along with visual inspection of histograms and P-P plots

A comprehensive literature review of relevant academic and non-academic sources of evidence-based practices, programs, and initiatives that currently focus on access to Genetic services and testing, access to health care, the role of the genetics counselor, and current practices in genetics counseling service provision.

# Chapter 4 Results

**4.1 Demographics**

Table two displays the demographics of the population included in the study. Of the 78 referrals, 66.7% were female and 33.3% male. As the study focused on the adult population, excluding anyone under 18, the age distribution was 18 - 78 years, with 50% (n=39) of referrals coming from the 41 – 65 age group

**Table 2:**

*Demographics: Age and Gender of referrals for PMGP included in this study*

| Age Group (years) | 19 -24 | 25 - 40 | 41 - 65 | 65 + | Total |
|---|---|---|---|---|---|
| Female | 5 (6.4%) | 15 (19.2%) | 27 (34.6%) | 5 (6.4%) | 52(66.7%) |
| Male | 2 (2.6%) | 7 (9%) | 12 15.40%) | 5 (6.4%) | 26 (33.3%) |
| Total | 7 (9%) | 22 (28.2%) | 39 (50%) | 10 (12.8%) | 78 (100%) |

*Note:* N=78. Average age 46.9, a higher percentage of referrals were female for both cardiac (70%) and cancer (86.3).

**4.2 Analysis of the Research Question**

***4.3.1 Overall Access Times***

Of the 78 referrals, nine did not require appointments but contributed to the overall wait times. Of these nine referrals, four did not require an appointment as there was no available testing to offer at that time. These referrals were "closed" after this was identified. Three patients died and two moved out of province, these referrals were removed after they were a 'No Show' for a booked appointment.

The median access (wait) time for all referrals was 45.1 months (95% CI, 38.9 – 51.3), (Table 3), the mean wait time was 39.8 months. As the median was different from the mean, it was decided to add the interquartile range as the primary descriptor. A portion of referrals, 15% attended an appointment within 12 months. The interquartile range is 22 months.

**Table 3**:

*Time to Event reported in months for all referrals to the PMGP for 2013*

| Median | | | | Percentiles | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| | | 95% Confidence Interval | | 25% | | 50% | | 75% | |
| Estimate | Std. Error | Lower Bound | Upper Bound | Estimate | Std. Error | Estimate | Std. Error | Estimate | Std. Error |
| 45.1 | 3.2 | 38.9 | 51.3 | 52.1 | .583 | 45.1 | 3.2 | 30.1 | 8.1 |

*Note:* Nine of the 78 did not require an appointment to see either a GMP or a GC, these contributed to the waitlist time until this decision was made. Another 11 (14.1%) were no-shows or refused an appointment, and these contributed to the wait times until the date of the appointment.

**Table 4:**

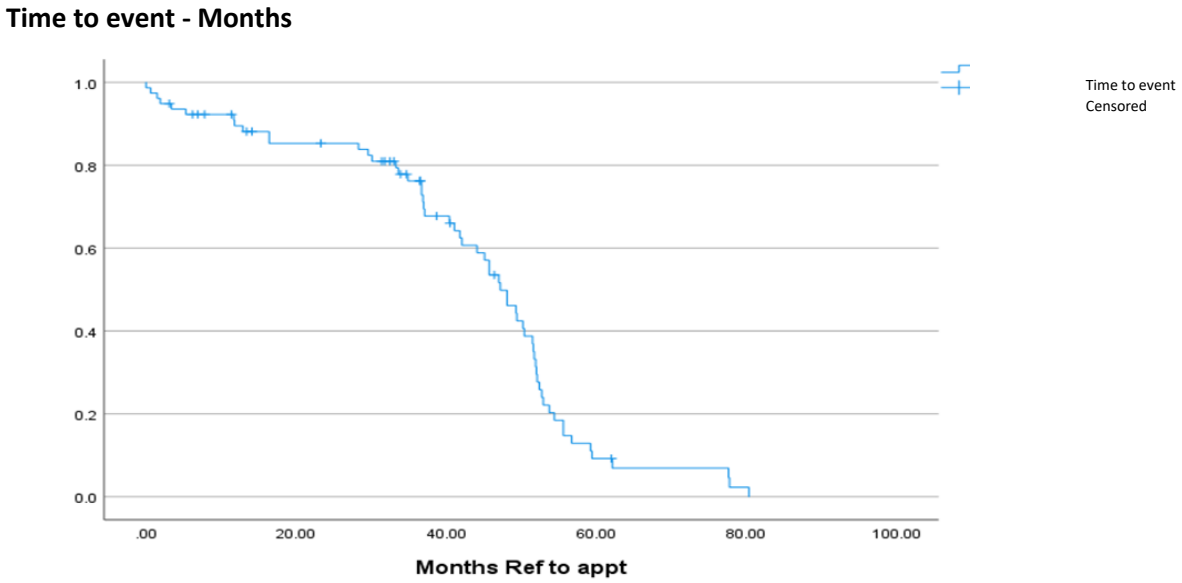
*Number of Appointments by year for all referrals received into the PMGP for 2013, categorized by appointment outcome.*

| Appointment year | Appointment Not required | No Show | Refused appointment | Appointment Attended | Grand Total |
|---|---|---|---|---|---|
| 2013 | 3 | 1 | 1 | 8 | 13 |
| 2014 | 3 | | | 3 | 6 |
| 2015 | 1 | 5 | | 6 | 12 |
| 2016 | 2 | 2 | 1 | 15 | 20 |
| 2017 | | | | 22 | 22 |
| 2018 | | 1 | | 4 | 5 |
| Grand Total | 9 | 9 | 2 | 58 | 78 |

Table 4 depicts the year an appointment was booked if required, and the status of that referral. In 2013, 13 referrals were closed, with eight having an appointment booked and attended, one refused the appointment, one did not show and three did not require an appointment at all.

**Figure 4:**

*Time to the event (appointment).*




*Note*: The above data is reported as months to access genetics services at PMGP, for all referrals from 2013.

### *4.3.2 Access Times by Service*

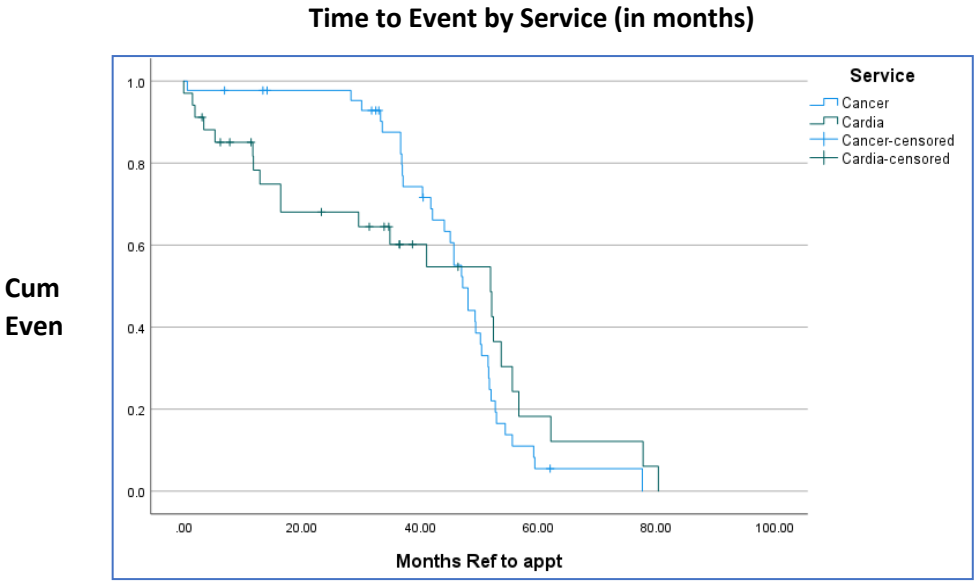
Some referrals did not have an appointment but impacted the overall wait time and these are censored in the analysis. Censoring refers to " the end of follow-up events other than the primary study outcome." (Bland, 2015). For this data analysis, 11 referrals were censored as they did not have an appointment: due to death (n=3), refusal of appointment (n=2), did not require an appointment (n=4), or moved out of province (n=2).

The median access (wait) time shown in Table 5 for cancer referrals was 45.7 months, and cardiac referrals were 36.5 months, the 95% CI overlapped, (cancer 41.8 – 49.5 versus cardiac 22.7 – 50.3) indicating that the difference in wait time is not statistically significant at 95% confidence level. A greater proportion of cardiac genetics referrals received an appointment within 24 months (Table 8), 44.1% (n=15), versus cancer 9% (n=4). The reason for this difference may be related to the request for

the patient to provide additional information, (cancer referrals 26/44, (59.1%) versus cardiac 7/34, (20.5%)).

**Figure 5**:

*Time to the event (appointment).*




*Note*: The above data is reported as months to event, identified by service for all referrals included in this study

**Table 5:**

*Time to event identified by service and reported in months for all referrals included in this study*

| Time to event by Service | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | Median | | | | Percentiles | | | | | |
| Service | Estimate | Std. Error | 95% Confidence Interval | | 25% | | 50% | | 75% | |
| | | | Lower Bound | Upper Bound | Estimate | Std. Error | Estimate | Std. Error | Estimate | Std. Error |
| Cancer | 45.7 | 1.9 | 41.8 | 49.5 | 51.6 | 0.98 | 45.7 | 1.9 | 36.9 | 1.9 |
| Cardiac | 36.5 | 7 | 22.7 | 50.3 | 53.7 | 2.1 | 36.5 | 7 | 11.8 | 4.7 |
| Overall | 45.1 | 3.1 | 38.9 | 51.3 | 52.1 | 0.58 | 45.1 | 3.2 | 30.1 | 8.1 |

*Note:* The event is defined as the date first access to genetic service was provided (appointment).

**Table 6:**

*Percentage of referrals seen by year and service***.**

| | Year | | | | | |
|---|---|---|---|---|---|---|
| Service | 2013 | 2014 | 2015 | 2016 | 2017 | 2018 |
| Cancer | 4.50% | 9.10% | 25.00% | 59.10% | 95.50% | 100.00% |
| Cardiac | 32.40% | 44.10% | 58.80% | 73.50% | 91.20% | |

*Note:* Cumulative percentage of patients seen by service year over year.

### *4.3.3 Access Time by Triage Status*

Triaging referrals is a part of the referral process within the PMGP and takes time to review the relevant history and determine urgency. It is hypothesized that referrals classified as "urgent" would have a shorter wait time to the appointment than other, less urgent triage codes.

Referrals are identified as one of three categories. Routine, Semi-Urgent, or Urgent. Of the 78 referrals, 49 had an assigned triage code, with 29 not having any indication that a triage process had taken place (Table 7).

**Table 7:**

*Identification of referrals by Triage Outcome*

| Referrals by Triage Outcome | |
|---|---|
| Triage Outcome | Total |
| Not assigned | 29 |
| Routine | 26 |
| Semi Urgent | 15 |
| Urgent | 8 |
| Overall | 78 |

*Note:* This table demonstrates the outcome of the triage process for each referral.

Of the 49 referrals with an identified triage code, 33 had required additional information, to assign an appropriate triage code. Figure 6 shows the time to appointment by triage outcome. Statistical comparison of the 3 levels of triage, as well as those that did not have an assigned triage code, shows no statistical difference in the wait time for an appointment. Those assigned an urgent triage

code (45.1 months, CI 11.4 - 69.4), did not access service before referrals with an assigned routine triage code (36.7 months, CI 31.2 – 42.2), or no triage code (45.7 months, CI 40.9 – 50.4).

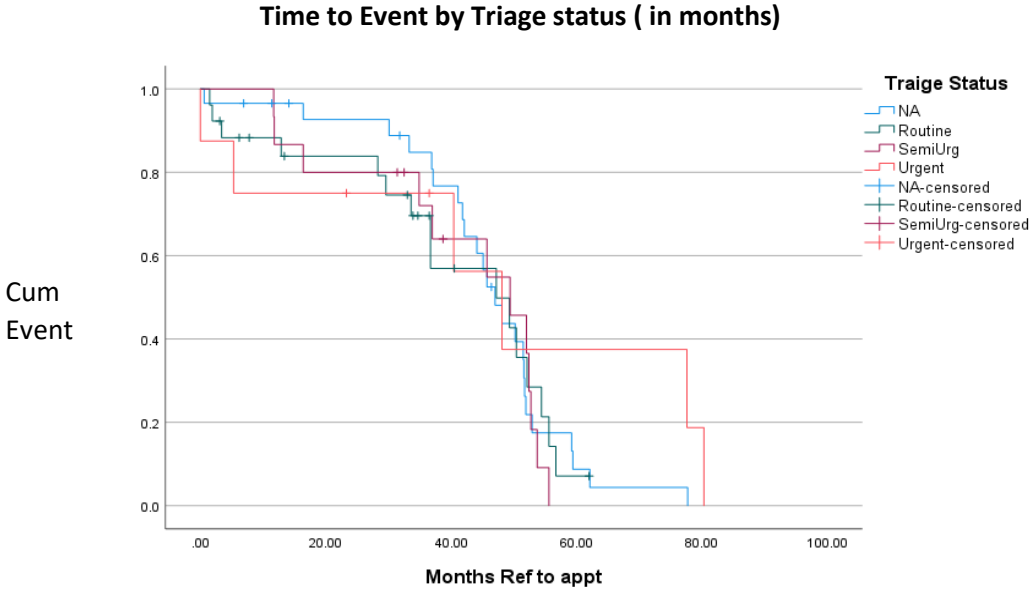
**Table 8:**

*Median time to event (Appointment) for all referrals identified by triage status and reported in months*

| Medians for Time to Event (Appointment) | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| Triage Status | Median | | | | Percentiles | | | | | |
| | Estimate | Std. Error | 95% Confidence Interval | | 25% | | 50% | | 75% | |
| | | | Lower Bound | Upper Bound | Estimate | Std. Error | Estimate | Std. Error | Estimate | Std. Error |
| NA | 45.7 | 2.4 | 40.9 | 50.4 | 51.7 | 0.73 | 45.7 | 2.4 | 36.9 | 6 |
| Routine | 36.7 | 2.8 | 31.2 | 42.2 | 52.1 | 2.9 | 36.7 | 2.8 | 13.4 | 16.1 |
| Semi Urgent | 49.4 | 10.1 | 29.6 | 69.2 | 52.7 | 0.45 | 49.4 | 10.1 | 31.4 | 15.1 |
| Urgent | 45.1 | 14.8 | 11.4 | 69.4 | 77.6 | 26.8 | 40.4 | 14.8 | 5.3 | 14.3 |
| Overall | 44.3 | 3.2 | 38.9 | 51.3 | 52.1 | 0.58 | 45.1 | 3.2 | 30.1 | 8.1 |

*Note:* a. Estimation is limited to the largest Time to Event if it is censored.

**Figure 6**:
*Time to event (appointment)*




*Note:* The above data is reported as months to the event, by triage status, for all referrals included in this study.

### *4.3.4 Requesting Further Information*

The need for the GC to request further information from the patient to establish a triage status requires a deeper look into the data. 33 referrals required extra information. A request was sent to the patient being referred, including the forms to be completed. Of the 33, 26 were cancer, and 7 were cardiac (Table 9). The overall return rate was 51.5%, with 48.5% having no documented indication that requested information had been returned.

Table 9 indicates the number of referrals that required additional information, while Table 10 shows the number of referrals that had this additional information returned.

There was considerable documentation related to follow-up calls and letters requesting the information be returned. An average of three (1-6) calls per outstanding chart, which required a substantial investment of human resources to complete, with little response. It was unclear if the delay in returning the documents impacted the overall wait time of the individual referral. Additionally, all referrals were offered an appointment despite not having returned the requested documentation.

**Table 9:**

*The number of referrals requiring additional information from, identified by service, and assigned triage status*.

| | NA | Routine | Semi Urgent | Urgent | Grand Total |
|---|---|---|---|---|---|
| Cancer | 12 | 8 | 4 | 2 | 26 |
| Cardiac | 2 | 2 | 2 | 1 | 7 |
| Grand Total | 14 | 10 | 6 | 3 | 33 |

*Note:* This data indicates referrals that required further information to complete the triage, requiring extra work, and may indicate an incomplete referral or lack of relevant information.

**Table 10**:

*Number and percentage of requested forms for additional information that were returned, identified by service, and assigned Triage status.*

| Service | NA | Routine | Semi Urgent | Urgent | Total |
|---|---|---|---|---|---|
| Cancer | 33.3% (n=4) | 41.7%(n=5) | 16.7% (n=2) | 8.3% (n=1) | 12/26 (46.2%) |
| Cardiac | 0.0% | 40.0% (n=2) | 40.0% (n=2) | 20.0% (n=1) | 5/7 (71.4%) |
| Grand Total | 23.5% | 47.1% | 56.7% | 28.3% | |

*Note*: This data indicates that not all requested information was received.

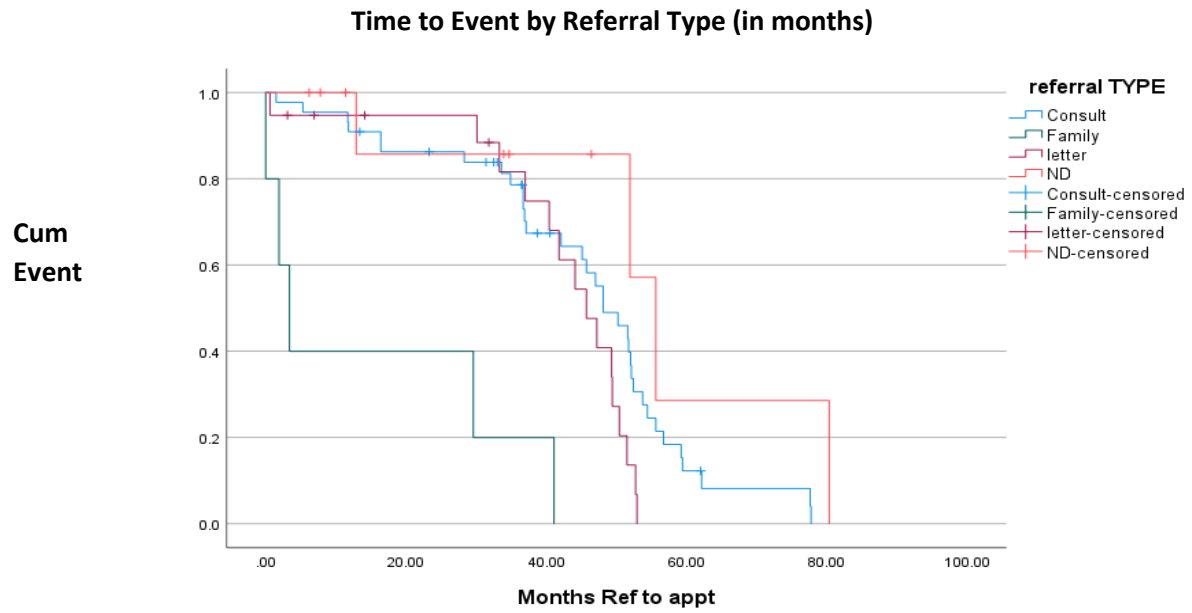
### *4.3.5 Access Time by Referral type*

There was a noted decrease in wait times for referrals with family connections, (Figure 8) with the median wait time of 3.37 months, as compared to the median greater than 44 months for the internal consults or external doctor's letters (Table 15). A family connection referral is a family member of an existing proband. The number of charts for this category was rather low (five), however, this is likely true for family referrals that are received with the PMGP, as there is a pre-existing relationship with the department, and the family may already be known.

There was no significant difference (Table 11) in the time to appointment for referrals received from within the hospital system (Consult) and those referred by physician requests external to the hospital system (Dr. Letter).

**Figure 7**:

*Time to the event (appointment)*




*Note:* Identified by referral type with censored data identified, and reported in months for all referrals included in this study.

**Table 11**:

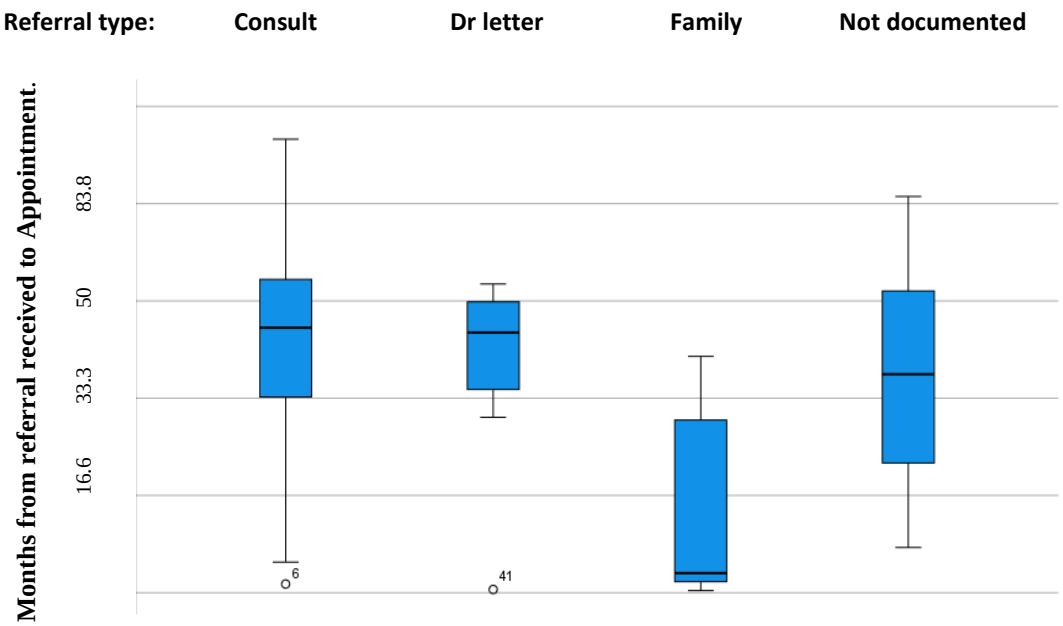
*Time to event identified by referral type, reported in months for all referrals include in this study*

| | Median | | | | Percentiles | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | 95% Confidence Interval | | 25% | | 50% | | 75% | |
| Referral TYPE | Estimate | Std. Error | Lower Bound | Upper Bound | Estimate | Std. Error | Estimate | Std. Error | Estimate | Std. Error |
| Consult | 45.700 | 3.807 | 38.238 | 53.162 | 53.700 | 1.873 | 45.700 | 3.807 | 33.600 | 5.239 |
| Family | 3.400 | 1.643 | .179 | 6.621 | 29.600 | 23.434 | 3.400 | 1.643 | 1.900 | 2.081 |
| Dr letter | 44.100 | 3.655 | 36.936 | 51.264 | 49.400 | 1.869 | 44.100 | 3.655 | 30.100 | 16.469 |
| ND | 55.600 | 17.883 | 20.549 | 90.651 | 80.300 | . | 55.600 | 17.883 | 51.900 | 27.358 |
| Overall | 45.100 | 3.170 | 38.887 | 51.313 | 52.100 | .583 | 45.100 | 3.170 | 30.100 | 8.129 |

*Note:* Medians for Time to Event By referral type (Months)

**Figure 8**:

*Months to Appointment by referral resource*




### 4.4 Interview Results

The results of the interviews are consistent with the data analysis, indicating a complicated process, and frustration with prolonged wait times. All nine respondents indicated that they were not satisfied with the wait time for their patients to access services, while 78% indicated they were not satisfied with the referral delivery process. The identified barriers are noted in Table 12 and can be categorized as process or system issues as identified in Table 14. All respondents identified the commitment and professional integrity of the PMGP staff, and the knowledge and compassion of the GCs.

**Table 12:**

*Barriers to access and time to service by PMGP, identified by the interview respondents.*

| What do you see, or have experienced as barriers to accessing services with PMGP |
|---|
| • No direct access |
| • If a specialist suspects a genetic connection, should be able to order baseline testing |
| • Need to be seen before testing provided |
| • Inconsistent referral process (2) |
| • Inability to track referral (2) |
| • No rule-out criteria, so unsure when a referral is appropriate |
| • Lack of coordination of care with specialist |
| • Travel - need to go to the city for an appointment, should use telehealth more. (2) |
| • My patients wait, and then never see a doctor. |
| • Lack of connection of referral state |
| • Disjointed process, needed my patient to see genetics but they ended up seeing a cardiologist |
| • Having to test the initial family member makes sense, but the delay for the others is too long. |

*Note:* Table 12 displays direct quotes from the interview participants.

The suggestions for improvement as proposed by the respondents in Table 13 are in keeping with a standard, evidence-based referral, and access process noted in the literature and recommended by the CMA.

**Table 13**:

*Survey respondents' suggestions for improvements to the PMGP referral process.*

| **Suggestions on improvements** |
|---|
| Better access to GC |
| Quicker, streamlined consult process |
| More timely response |
| A response from the program when referral received (2) |
| Connect with the specialist (2) |
| Provide an update on time frames |
| Provision of some information to the patient before the appointment so they can prepare |

| |
|---|
| Look at the process of having all consents done at the initial appointment, so family information can be shared, and you do not have to go back for it. |

*Note:* Response from nine participants, included duplicate statement identified by the number behind the statement

# Chapter 5 - Discussion

## 5.1 Genetics Services

Medical Genetics represents a specialized, unique, and developing field of healthcare. Emerging technology and testing provide the ability for individuals to make proactive decisions to mitigate the risk of developing a hereditary disease. How this impacts the service delivery for medical genetics varies by the demand. Access to basic health care is complex, requiring four factors; is the service availability adequately resourced, can the population access the service and how is the organization facilitating this (Gulliford, 2002). Access to healthcare in rural areas is further complicated by distance, weather, transportation and community accessibility, and the social determinants of health. This is a global concern since half the world's population resides in rural areas (UN Department of Economic and Social Affairs Population Division, 2005). It is certainly a concern in NL where 40% of the population lives in rural areas (Newfoundland and Labrador Department of Finance, 2020). While the challenges to access basic health care exist, for both the patients and primary health care practitioners, it is even more complex to access specialized healthcare services, such as genetics (Harding, 2019; Johannessen, 2018)

The purpose of this study was to determine the wait times to access services for hereditary cancers and cardiac disease, as well as identify the barriers that exist that impact wait time and access.

To fix access we must first understand the processes and the etiology of the barriers. As reported by the Canadian Medical Association (CMA), 2004, waiting for care has become part of any health care system and has often been built into the practice. Lack of understanding related to the demand and the capacity of the service often leads to ineffective processes such as overbooking a clinic to balance the no-show rate (Johannessen, 2018).

With increasing technology and the ability to receive a private genetic testing kit, discovering your genetic makeup is advertised as exciting and fun and has become more mainstream. As the results

of the testing are shared directly with the customer, a decision may be made to seek further testing or treatment, or exploration of a genetic condition without the guidance of specialized genetics medical practitioners.

It is recommended that a genetics professional be involved with the initial assessment, (Khouzam, 2015), and that a patient receives genetics counseling before being offered testing options. Providing information and interpretation related to testing results that may indicate an increased risk or predisposition to a genetic syndrome, will empower patients to make an informed decision regarding the next steps. The patient may require psychological support to assist with decision-making and sharing of the information with other family members who may be or have been impacted. Often the decision to have life-changing surgery, major medical treatment, or to have children requires consideration. The GC must be educated and experienced in the fields of need, which is why GCs are often assigned specific roles within a department such as cancer, cardiac and prenatal genetics. (Shugar, 2017), This is the approach EH's PMGP has taken.

Within the EH PMGP, the wait time to see a GC is lengthy, and despite the desire of the staff to see those most urgent, and those in need of the service first, the current process is a barrier. This impacts the team as they see the waitlist grow and question if they are efficiently seeing the most urgent patients. This added stress can lead to an unhealthy work environment and a lack of engagement in the workplace (Rasool, 2021). Additional factors that have been identified as barriers to the access of service and are out of the control of the PMGP staff are those built-in processes and the culture of the organization. (Valaitis, 2018).

**5.2 The Referral Processes**

The referral process within the PMGP has been inconsistent and often arduous on both the patient and the clinician. Within each step of the process, there are many secondary steps, which may or

may not be required depending on the referral content and available information. This study has verified that the referral process is manual, fragmented, and vulnerable to lost, misfiled, or overlooked referrals. Once a referral is received within PMGP, the information is entered in the Shire database, and a new chart is developed, or if the referral is for an existing patient, their current files/chart would be used. Unfortunately, if a referral is lost or misplaced, the team may not know until the patient or referring physician calls the PMGP to inquire on the status of the referral (PMGP, Personal Communication, 2016). Even then, the process to find this information and provide an accurate update is antiquated and cumbersome. Patient charts are paper-based and stored within the PMGP department. All patients are assigned a unique pedigree number which is entered in the Shire database and attached to the paper chart. A chart locator search function within the Shire database is used to track the locations of the paper chart. The concept of this is sound, and if used in a standardized and consistent approach would allow the PMGP staff to document where and with whom the chart is currently located. Unfortunately, due to inconsistent practices, it is often difficult to locate a chart or referral using this process. This leads to extra work, and often workarounds. One such workaround is frequently seen as the documentation of information is written on a single sheet or form and then placed into a loose filing folder for placement in the chart when located. (PMGP, Personal Communication, 2016). This leads to a potential delay in information reaching the patient chart and increases the risk of pertinent information being misplaced or misfiled.

Various themes emerged that may prolong the referral process or delay access to services that are outside of the department's control. The efficiency of the referring practitioner, response time for additional information requested from the patient, the referred patients' interest in testing, comfort with the process, or the support they receive from the family. (Khouzam, 2015; Delikurt, 2015), When a patient is not interested in receiving services or testing, they may not return a requested form or

information, or advise the PMGP. Further study into this aspect would provide better insight into the reasons for this and allow the department to enhance patient engagement.

The study results show that the average time for initial processing of the referral is two days, however, some referrals had an initial processing timeline of up to 111 days. As not all referrals had documentation of the date received, and there was no documentation on the referral indicating if there was a reason for the delay, it is unknown if this delay occurred on the sending or receiving end. A standardized process that begins the moment a referral arrives in the PMGP would identify why a delay, if a delay, and would mitigate the risk associated with the omission of information that may stall the referral when not received. (Boettcher, Hunter, & McGonagle, 2019). Within a standardized process, each step of the referral's pathway would be mapped out and roles and responsibilities would be assigned. When one step of the process is completed, there would be no uncertainty about what the next step would be. Standard documentation processes would allow for all PMGP staff to review a chart (referral) and know what has been completed, what is outstanding, and what may be stalling the referral from reaching the next step.

There are strong evidence-based guidelines within the literature to support the patient's eligibility for genetic testing (Dos Santos Vidal, 2016; Blair, 2020; Vasen, 1999; Gupta, 2019). Criteria to support an appropriate referral is available, however, the PMGP did not have established guidelines. The study results found that all referrals were accepted and processed, as it would not be known at the point of entry if there was a need for genetic testing or follow-up. Development of a standard referral would ensure the referral was appropriately completed, allowing the PMGP to determine if the referral is accepted for service or returned for more information at the point of entry. Providing updated guidelines for referring to HCP would also assist with the identification and potential redirection of an

inappropriate referral while allowing efficient communication back to the referring HCP and patient. (Pieterse, 2007).

Patient referrals were received from health care practitioners in various formats, such as physician notes, prescription pads, and consult sheets. Electronic referral processes have been shown to improve the access time to specialty services (Azamar-Alonso, 2019), and should be explored to improve efficiency and the quality of the process. In NL the lack of province-wide high-speed internet must be considered as implementing an electronic referral process could be a barrier to care. Referrals from rural family physicians and specialists, as well as referrals from family members, could be negatively impacted. Extra up-front investment may be required of the organization and the province, this must be seen as supporting a more streamlined process that will result in improved patient care and outcomes. Patients would receive the right services in a timely and efficient manner (Figure 9)(Components of appropriate care, adapted from Canadian Medical Association, 2015; Consortium for Genetics, 2009). The ability of an electronic process to locate and confirm receipt of a referral, determine wait time, and confirm that all supportive or requested documents have been received, are additional benefits to the PMGP team and the patient. Important to note that introducing a new work process would require knowledge translation and ongoing support for both the PMGP and the referring HCP.

**5.3 Triage**

A triage system is necessary where there is a greater demand than the current service capacity can accommodate (McVeigh, 2019). A program's triage system or urgency classification system should be established to ensure that the most urgent in need of care patient is seen before less urgent or routine required care. This should be standardized based on the service and require minimal information to complete. This study did not uncover any evidence of a robust, best practice-based triage category system used within PMGP. There was an understanding amongst the team that the triage

process used in PMGP was based on EH's established categories using three separate urgency classifications: Routine, Semi Urgent, and Urgent (EH waitlist Management Initiative 2013). Patient referrals may have an urgency category noted on the referral, assigned by the referring HCP. Once received within PMGP each referral is reviewed and given a triage status based on urgency, indication, and prior family history. This allows each patient to be individually assessed, prioritized, and placed on the waitlist accordingly. The assessment for triage is completed by the GC that is assigned to the triage process during that time. Informal discussion with the GC confirmed that the referrals are reviewed and assigned a triage status based on the assigned GC knowledge, experience, and "review with another colleague if unsure" (PMGP, Personal Communication 2015). There is also the option of having the referral triage by, or with the Medical Geneticist.

As previously indicated, the study results showed that all referrals are accepted for service and processed for triage. It was noted during the analysis that some referrals had been discharged before receiving an appointment, but lack of consistent and standard documentation prevented the ability to determine if this decision was part of the triage process, or if the decision to discharge the patient came about from the review of outstanding patient referrals that had not received service. These referrals remained active and impacted the overall wait time data.

The average number of days for triage to commence was 20, with a median time of seven days. As there were no written or established standard triage documentation processes, start and end dates were not captured, the time involved for the triage process to occur was not able to be verified.
Of the 78 included referrals, 37.2% (n=29) did not have an identified triage code, nor was there documentation indicating if any triage discussion took place. 19% (n=15) had a triage code of semi-urgent, 10% (n=8) urgent, and 33% (n=26) routine. Based on the data analysis, there was no significant difference between the wait times by assigned triage code. Those with an assigned triage code of routine (36.7 months) did experience a trend towards shorter wait time than the semi-urgent (49.4

months), and urgent categories (45.1 months). This was unexpected. When a standardized referral triage process is developed and used, urgent categories would be seen before routine.

For the provision of health care, the value for the patient is in receiving the services (Johannessen, 2018). Based on study outcomes, the existing triage process adds little value to the patient and does not have the desired outcome of the most urgent patient accessing service sooner than the less urgent patient. This process is questionable, as it is not evidence-based, and supports the need for the development of a robust standard triage process that works for the patients who require services and supports the PMGP in making patient care decisions.

### *5.3.1 Request for Additional Information*

The triage process requires a review of current medical information, existing genetic information, as well, there may be a need for additional medical and/or family history information. Current and accurate information assists the team in assessing and assigning the triage/urgency status and supports a reliable equitable access process for the patients.

The study results show that of the 78 patient referrals for triage, 42% (n=33) required extra information to be provided by the patient, and or family to complete the triage assessment, and assign the appropriate triage status. The remaining 45 referrals did not require further information and once triaged were entered into the Shire system until an appointment date was identified and booked. The Community-Wide Schedule module (CWS) available in the Meditech system can track patients' appointment requests by triage status, allowing for the patient to be scheduled for the next available appointment that fits the triage status parameters. As the PMGP did not utilize the full functionality of the CWS within Meditech, there was no guarantee that the patient received the next available appropriate appointment based on the assigned triage status. CWS would also allow the team to review a report on the waitlist more efficiently. The implementation of this module would require upfront work

and an evidence-based triage process but would save time on the other end with the need to follow-up of referrals and ensure the referral does not get misplaced or unnecessarily delayed.

Once the request for additional information is sent to the patient, the process and flow are placed in the hands of the patient, and the PMGP no longer can monitor the referral progress as it is dependent on the return of the requested documents. Of the requests for additional information, 51.5% (n=17) were returned, this was confirmed by the documents identified in the file and noted in the Shire system. The remaining 48.5% (n=16) did not have any indication of receipt of the requested information, no dates documented in the Shire system, nor in the chart. In previous studies, the response rate for this request within NL has been low (Parfrey, 2017). The reason why these requested documents were not returned is unknown, however, it is noted in the literature that lack of knowledge of family health history is a barrier when completing the family history (Lu, 2014). Reading level, and ability to easily complete the documents is a noted barrier (Safeer, 2005), suggesting a review of the reading level of current documents should be completed. The clinicians should also be mindful of the accuracy of the information, as the reliability of the health history of more than two generations may not be clear or available (Goergen, 2016). As there was no documentation of the dates the information was returned, or if at all, the ability to calculate the average time for document return could not be measured. It is worth noting here, for the patients who did return the documents, 81% (n=13) were called, and/or sent letters or reminders two to six times, equaling 30 additional tasks required by the GC and clerical staff. As previously indicated, the triage process times were not studied, so the impact of this additional workload on the PMGP team could not be measured but should be acknowledged.

Prior studies have noted the importance of family history (Goergen, 2016; Bennett, 2012) as a required element of the triage and case review processes. If these are not returned, they will be completed during the initial appointment “we (sic) will fill it out with them, as they don’t know how to

answer some of the questions" (Personal communication, GC 2015). This leads to the question if the patient receives an appointment with or without returning these documents, what is the value in adding this task to the process at that point, and can it be completed more efficiently? The implementation of an electronic process has been shown to increase the return rate and decrease the length of time it takes to complete and return the documents. (Azamar-Alonso, 2019; Bell DS, 2012) From a patient safety perspective, this would allow documents to be sent and received via an electronic system, with tags and reminders for follow-up, decreasing the burden and inefficiencies of a manual process.

**5.4 Overall Wait-times**

Wait times are an important indicator of measuring access to organizations. While the work or internal process is important for the organization, they are not important to the patient (Johannessen, 2018) and the patient trusts that the service providers have appropriate workflows that ensure efficiency and safety. The developed processes require continuous review and a consistent methodology, based on best practices. The PMGP lacked written standardized procedures, policies, and standard operating procedures. The results of this study show that the median access (wait) time for an appointment for all referrals was 45.1 months, (Table 3), the mean wait time was 39.8 months. 15% (n=13) of all referrals attended an appointment within 12 months. While 11 patient referrals did not require an appointment, they did proceed through the system, requiring a chart review to determine this, thus adding to the wait time and department workload. As noted earlier a standardized referral would allow early assessment of the appropriateness of a referral. Identification of an inappropriate referral would allow it to be removed from the workflow earlier in the process thus minimizing unnecessary work. Communication of an inappropriate referral back to the referring HCP in a more efficient time frame would allow them to adapt the plan of care and alleviate the anxiety the patient may be experiencing while waiting for an appointment that may not be required. (Johannessen, 2018)

There are situations when a patient may not be able to make a scheduled appointment, forget or just dismiss it as not necessary. Within the PMGP from 2013 to 2016, the no-show rate was 23% (Meditech Data). This means that the booked patients did not show up for their scheduled appointment, nor did they call and cancel. This leads to wasted appointment slots and the need for follow-up and rebooking an appointment, as necessary. There is a “no show” policy with EH, this policy needs to be implemented and consistently followed by PMGP. The team spends time following up with a patient who does not show for an appointment, which includes a minimum of one call. We were unable to quantify the amount of work invested in this process as it was not consistently documented.

**5.5 Wait Time by Service**

Analysis of the wait times demonstrated a trend towards difference in the time from referral to the appointment between hereditary cancer and cardiac patient referrals, but the results are not statistically significant. The median wait-time for a cardiac patient referral was 36.5 months, and for a cancer patient referral, was 45.7 months. From 2013 to 2017 there has been a steady growth of referrals for both cancer and cardiac genetics, with an increase of 9 – 10 % increase year over year. As the number of referrals increased, a human resource plan to match demand was not in place.

For cardiac genetics patient referrals, depending on the identified level of risk, the patient’s medical information may be required to be reviewed with the Cardiac Genetics Clinic (CGC) team. There are three paths the patient may be directed towards; 1. GC only, 2. GC and cardiac MD, 3. Registered Nurse appointment to establish baseline testing. These services are offered by different programs, and the internal program processes differ, such as triage/urgency assignment and clinic availability. The coordination of the appointments is very challenging as the booking of the appointments is the responsibility of each program's clerical support. These disjointed processes cause delays in the genetics appointment and can artificially elevate overall wait times. This has been identified as a barrier by both programs. The cardiologist requesting to see the patient at the same time as the GC may further delay

the genetics appointment, as the cardiologists assigned to the genetics clinic have limited capacity outside of their cardiology practice. Unfortunately, the electronic and manual chart documentation systems do not capture the complexities of the CGC process, therefore identifying the impacts on wait times is not possible. Seeing the Genetics MD occurs after these initial visits, and only if needed.

Patients who are referred to the PMGP for potential hereditary cancer diseases are provided information by the genetic counselor on the availability and need for testing, family history, and inheritance of hereditary cancer syndromes, and should be provided with a cancer risk assessment and preventative screening recommendations. (Gupta, 2019). There is also the practice of offering cascade testing, genetic screening, and follow-up to the family members of the proband, adding additional pressure on the sparse resources. A note of caution, not all of these referrals are captured individually, therefore may not be represented in the referral data.

The complexity of the changing genetics landscape has led to increasingly complicated referrals, placing extra demand on the GCs and the organization. The resources assigned to each of these specialties have remained unchanged since 2003. In 2012 a request was approved to increase GC resources, but due to limited graduates, shifting responsibilities, and availability of current staff, the benefit of the extra HR was not realized. The number of available new graduates has not met the growing demand, which places further strain upon the incumbent GCs (Hoskovec, 2018). The stabilization of the department HR was not achieved until 2016, all the while the waitlist continued to grow.

**5.6 Wait Time by Referral Type**

How a patient referral enters the system should not impact wait time. The literature demonstrates that the mechanism for entry is not the deciding factor for how a patient referral is placed in the queue. Based on this, we would have presumed to have seen no significant difference in time to event for referrals received from within the EH system versus those received from outside of the EH

system. These referral types are categorized by: Internal health care providers, (consult sheet), doctor letter, (which would be from the community or outside of EH), and family connections of a previous patient. The study found that there was no significant difference in the time to event for the internal and external referrals. Both groups had a prolonged wait time of greater than 44 months.

Interestingly, there was a noted decrease in wait times for referrals with family connections, ($p<0.05$) with a median wait time of 3.4 months. A family connection referral is a family member of an existing proband. Caution must be applied when interpreting these results as the number of charts for this category was low (five), it is difficult to establish if this is the case for all family referrals received with the PMGP.

**5.7 Barriers and Facilitators**

Both barriers and facilitators were identified during chart reviews and from the stakeholder interviews. Most topics could be identified as a barrier or facilitator, but a few could be classified as either. For example, medical staff support was identified as easily accessible by one group, and difficult to access by another group. The overall themes that emerged could be classified into four categories: internal processes, organizational practices, patient involvement, and referring HCP. (Table 14).

**Table 14**:

*Barriers and facilitators from data analysis and response from interview participants*

| Barriers | Facilitators |
|---|---|
| **Internal processes** | |
| Triage process<br>Inconsistent referral form<br>Requirement of more information<br>Lack of flags to track referrals<br>Duplication of information<br>The inability of referring HCP to order baseline testing | Staff dedication and knowledge<br>Medical Staff support<br>Awareness of the need to provide efficient service<br>The willingness of the team to try new approaches |

| | |
|---|---|
| Medical Staff support | |
| **Patient Involvement** | |
| Lack of established follow-up processes for information request<br>Lack of interest in pursuing testing | Willingness to participate in testing and screening. |
| **Organizational Practices** | |
| Number of electronic systems<br>The inability of electronic systems to "talk"<br>No show practices<br>Complicated telehealth process | No show practices |
| **Referral Process from referring HCP perception** | |
| No direct access to baseline testing<br>If a specialist suspects a genetic connection, should be able to order base testing<br>Need to be seen before testing provided<br>Inconsistent referral process (2)<br>Inability to track referral (2)<br>No rule-out criteria, so unsure what is appropriate to refer<br>Lack of coordination of care with specialist<br>Travel - need to go to the city for an appointment, should use telehealth more. (2)<br>My patients wait, and then never see a doctor.<br>Lack of connection of referral state<br>Disjointed process, needed my patient to see genetics but they ended up seeing a cardiologist | Need to be seen before testing provided<br>Knowledge of the counselors |

*Note:* The data contained in Table 14 are responses from the health care providers' interviews and presented in themes for both barriers and facilitators.

**5.8 Recommendations**

Internal organizational processes are often seen as an insurmountable barrier. Lack of training, attitudes toward change, and fear of change are noted as the main reasons for resistance (Fischer, 2016). With the right support and engaging the care providers on the referring and service provision end, a solid process can be developed and implemented. Organizational quality and process improvement must be patient-centered. The Canadian Medical Associations Dimension of Appropriate Care (CMA, 2015) identifies five (5) rights for care (see Figure 11). These 5 rights, evidence, provider, care choices, place, and time support appropriate and timely access to service. (Figure 9). Patient-

centered care is why continuous process improvement cycles are needed. Having the patient be part of these process improvements can improve clinical adoption and the quality of the process (Fisher, 2016).

A robust evidence-based guide to the appropriateness of referrals would allow the early assessment of referrals to establish appropriateness and urgency. An educational strategy is needed for HCP across the province. The ability to access updated guidelines for an appropriate referral will improve access to PMGP.

Utilizing a standard referral form, based on evidenced-based appropriateness of a referral, in an electronic form would decrease errors and omissions in the referral completion, (Azamar-Alonso, 2019), decreasing the effort related to gathering information from the referring practitioner. Thus only those patient referrals that require the specialized care of the PMGP enter the system. Since the close of this study, the PMGP has implemented a standard referral form.

**Figure 9:**

*Dimensions of appropriate care adapted from Canadian Medical Association, 2015*.

**Dimensions of Appropriate Care**

1. Right care is based on evidence for effectiveness and efficacy in the clinical literature and covers not only use but failure to use;
2. Right provider is based on ensuring the provider's scope of practice adequately meets but does not far exceed the skills and knowledge to deliver the care;
3. Right patient acknowledges that care choices must be matched to individual patient characteristics and preferences and must recognize the potential challenge of reconciling patient and practitioner perceptions;
4. Right venue emphasizes that some settings are better suited in terms of safety and efficiency to delivering a specific type of care than others;
5. Right time indicates care is delivered in a timely manner consistent with agreed upon

Triage guidelines should be developed to improve and support decision-making, which would ensure that the patient in most need of service receives care quickly. These guidelines should be

evidence-based and built on best practices with the consensus of the team. An auditing process would ensure the guidelines are meeting the needs and identify when a change is required.

At the time of this study, there were no target times for the return of forms sent to the patient and/or family for completion. Most of these forms are sent to patients to gather further medical information on family members' health status. The investment by the organization in an electronic process to track referrals, create patient pedigree (Family tree), and collate documents would decrease the manual work that occurs in the department while mitigating the risk of human error or omission (Azamar-Alonso,2019; Bell DS, 2012).

Previous studies have indicated that GP and non-specialists lack clinical training and experience in the genetics field, thus feeling ill-prepared to provide counseling or follow-up care. Providing resources and support to the referring physician in the way of standard tools will assist the referring HCP in decision making. Redesigning the care pathway to provide the best service with available resources (Hallowell, 2019) must be evidence-informed, and support patient safety, quality care, provider satisfaction, and system efficiencies.

**5.8 Data limitations**

The number of charts reviewed for the study was less than initially proposed. As the review and data collection moved forward, some referrals/charts were found not to be relevant for this study, despite being included in the initial round. As themes had emerged and were repeating, it was felt that adding more charts to meet the 120 charts initially identified was work and time-intensive, and would add little value to the overall outcome.

The charts selected for review were in paper format and the information reviewed included written notes and communication. The process for entering referral dates and follow-up was also manual, the risk of human error is possible. There was no noted standardized process for what client

information was required on the chart, nor was there a list of supportive documents that were required to make a clinical decision on the next steps.

Due to the variation of the available data, further investigation was required to find the needed information, making data collection and validation very onerous and fragmented. Care was taken to ensure data integrity was maintained. I was the sole data collector, and a standard data collection sheet was developed and used.

The PMGP uses an external database, Shire Genetics Database. The data entry happens at many points of the process, by several staff members. Referrals are received via email, fax, and regular mail. The referral date and date received are logged in a paper process that is set up to manually track the referral. Consistency in how this information is entered has not been established, and with a rotation of employees, a standard format had not been developed.

It was noted during the data collection and review stage, that not all practitioners charted relevant medical information the same way, therefore if the chart was passed to another practitioner for follow-up, it was not always evident at what stage of the process the referrals were, or the required next step. The process for triage was inconsistent, and the pathway for the client's referral was not always clear.

The communication between staff was often handwritten either on a clinic note or on a clinic tracking sheet, such as the need for an appointment, and suggested dates, with no indication of urgency. Although a process improvement from 2012 indicated a standard referral pathway, it was not always fully implemented and followed. A review and update of these recommendations are suggested, with implementation by the team.

The response rate for the semi-structured interviews was 42.8%, and lower than expected. A low response rate can give rise to sampling bias and must be considered. Of the responses received,

55%  were from specialists and 44.4% were from general practitioners. Irrespective of the different professional practices,  commonalities in themes were noted for both barriers and facilitators.

**5.9 Conclusion**

The research demonstrates that at the time of this study patient wait times for access to genetic services are lengthy and unpredictable. For a patient to be referred for service in 2013, and not receive the service until 44 months later is not acceptable. The process is stressful and complicated for the patient, the referring HCP, and the PMGP staff. Processes for referring patients for genetic services need to be clear, standardized, allowing for an appropriate flow of information and centered around patient safety and access. This study demonstrated that the PMGP process of triage does not ensure that the most urgent patient received care before routine patients. These findings indicate that a paper-based, multi-stakeholder model is sub-optimal, and a more efficient technology-supported system should be explored and trialed.

Service providers must assess their processes to ensure they are providing equitable access in a safe, efficient, and effective manner. Staff input is imperative, and the changes and processes must be developed with the staff who should be supported and provided with the appropriate technology and human resources with which to do this.

In the current practice, there is minimal opportunity to review cases for quality assurance with the view to continuous quality improvement. Since the start of this study in 2013, the PMGP has implemented several of the recommended processes to improve access and the patient experience. The learnings of this research may be applied to future policy development, knowledge translation, and quality improvement in the provision of genetic care. This work may also be used as a baseline for future evaluation and process improvement design.

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# Appendix A: HREB approval

**Research Ethics Office**
**Suite 200, Eastern Trust Building**
**95 Bonaventure Avenue**
**St. John's, NL**
**A1B 2X5**

August 20, 2019

6 Wilchris Place
Mount Pearl NL,Canada A1N 4L6

Dear Ms. Dooley Adams:

Researcher Portal File # 20200534
Reference # 2019.153

RE: Barriers and time to access Genetic Counselling in Newfoundland and Labrador

Your application was reviewed by a subcommittee under the direction of the HREB and the following decision was rendered:

| | |
|---|---|
| X | Approval |
| | Approval subject to changes |
| | Rejection |

Ethics approval is granted for one year effective August 20, 2019. This ethics approval will be reported to the board at the next scheduled HREB meeting.

This is to confirm that the HREB reviewed and approved or acknowledged the following documents (as indicated):

- Application, approved
- Research Proposal, approved
- Clinician invitation, Clinician questionnaire/ Interview script, approved
- Data variable chart review, Data custodian variable list, approved
- Consent form, approved

Please note the following:

- This ethics approval will lapse on August 20, 2020. It is your responsibility to ensure that the Ethics Renewal form is submitted prior to the renewal date.

- This is your ethics approval only. Organizational approval may also be required. It is your responsibility to seek the necessary organizational approvals.
- Modifications of the study are not permitted without prior approval from the HREB. Request for modification to the study must be outlined on the relevant Event Form available on the Researcher Portal website.
- Though this research has received HREB approval, you are responsible for the ethical conduct of this research.
- If you have any questions please contact info@hrea.ca or 709 777 6974.

The HREB operates according to the Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans (TCPS2), ICH Guidance E6: Good Clinical Practice Guidelines (GCP), the Health Research Ethics Authority Act (HREA Act) and applicable laws and regulations.

We wish you every success with your study.

Sincerely,

[redacted]
Health Research Ethics Board

# Appendix B: Semi structured interview guide

Clinician Questionnaire – Access to Provincial Medical Genetics Services

1. What is your professional Designation/Degree?

2. What area of the province do you practice?

3. Have you referred a patient to the Provincial Medical Genetics Department of Eastern Health in the last 5 years? Yes ____ No_____

4. Which genetic service do you generally refer your patients too?
   General Genetics_____ Cardiac genetics_____ Cancer Genetics _____

5. Are you satisfied with the current referral process? Yes ____ No_____

   Please elaborate:

6. Are your satisfied with the time to access services?
   Yes ____ No_____

   If no, please elaborate:

7. In your experience, what do you see as barriers to access and/or testing with respect to the Provincial Medical Genetics Department?

8. How can the department's current process of accessing genetic services better meet your patient's needs?
   Yes ____ No_____

9. Do you have any comments on the overall process?

# Appendix C: Semi-structured interview participant invite.

**Recruitment script template**

My name is Kim Dooley Adams, and I am a masters student with the Faculty of Medicine Department, Clinical epidemiology at Memorial University of Newfoundland. I am conducting a research project related to *Barriers to Access, and Time to Genetic testing in Newfoundland and Labrador*, for my master's degree under the supervision of Dr. Patrick Parfrey.

The purposes of the study are:

- Define the patient referral process from point of entry to provision of genetic testing;
- Determine wait times for each component of the genetic testing process in cardiac disease and in hereditary cancers, and the barriers to testing for referrals; and
- Determine the optimal configuration for the genetic testing process to inform policy recommendations.

I am contacting you to invite you to participate in an interview in which you will be asked to share your experiences with referring patients/clients to the Provincial Medical Genetics department of Eastern Health, as well as share your thoughts on the ideal referral process. Participation will require approximately 30 minutes of your time and will be held at location of your convivence.

If you are interested in participating in this study, please contact me to arrange a meeting time.

If you have any questions about me, or my project, please contact me by email at t54kaa@mun.ca or by phone at 709 689 7750

Thank-you in advance for considering my request,

Kim Dooley Adams RN BN

Student - Master of Science in Medicine
Clinical Epidemiology, Faculty of Medicine
Memorial University of Newfoundland

# Appendix D: Consent to semi-structure interview

*HREB Version March 2019*

## Consent to Take Part in Research

**TITLE**: Barriers to access, and time to Genetic testing in Newfoundland and Labrador

**RESEARCHER(S)**: Kim Dooley Adams Phone Number: 709 689 7750

**SUPERVISOR(S)**: Dr. Patrick Parfrey, Dr. Leslie Turner, Dr. Sean Connors

**SPONSOR/FUNDER**: NA

You have been invited to take part in a research study. Taking part in this study is voluntary. You may choose to take part, or you may choose not to take part in this study. You also may change your mind at any time.

This consent form has important information to help you make your choice. It is important that you have as much information as you need and that all your questions are answered. Please take as much time as you need to think about your decision to participate or not and ask questions about anything that is not clear. The researcher will tell you about the study timelines for making your decision.

**1. Why am I being asked to join this study?**

We would like to hear from you about your experience as a clinician, accessing genetics services at Eastern Heaths Provincial Medical Genetics Program. From this study we hope to learn a bit more in general about existing barriers and gain a better understanding of concerns, and potential barriers at the health-system level. The results of this study will inform the development of policy and guidelines for access to genetic testing by General Practitioner or subject matter experts

**2. How many people will take part in this study?**

This study will take place within Newfoundland and Labrador. We have invited clinicians who have referred patients to the Provincial Medical Genetics service to participate.

**3. How long will I be in the study?**

You will be asked to participate in one interview lasting about 30 – 45 min. A follow up interview may be requested if clarification of information obtained is needed.

**4. What will happen if I take part in this study?**

Interviews: If you agree to participate, we will arrange a personal interview with you to ask you some questions about your practice and experiences as well as expectation of the service. This 30-45-minute interview will take place at a location of your convenience. Alternatively, you may wish to have an interview by telephone, set at a mutually agreeable time.

Participant Initials: _____ 1

Consent Version Date: _____