# Barriers and Facilitators in Health Care: Perspectives of Persons with Lynch Syndrome

By

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## Abstract

**Background:** Lynch Syndrome (LS) is a hereditary condition that substantially increases a person's risk of cancer and makes genetic testing for LS critical for organ surveillance or preventive care. A commonly identified barrier to LS genetic testing has been the difficulty in communicating genetic status between family members. Barriers to effective organ surveillance have also been documented and a coordinated approach to LS management has been suggested. However, few studies ask persons with LS to describe their experiences in accessing care and to put forth suggestions to improve access. Although authors have listed some suggestions to improve coordination, a framework for improving access by persons with LS has not been developed.

**Methods:** Drawing on interpretive description, this qualitative study involved 17 semi-structured, in-depth interviews to understand barriers and facilitators to persons with LS accessing care for LS. Thematic analysis of the transcribed interviews entailed a general inductive approach.

**Results:** Seventeen persons with four of five possible genotypes of LS were recruited from 12 families to represent experiences of accessing care for LS. The analysis identified six themes: It's health care, not 'Russian Roulette'; Knowledge; Central force in the family; Personalized approach; Ensuring continuity of care; Need for care coordination. A framework for improving health care access by persons with LS was developed.

**Conclusions:** Implementing the framework at the health system and clinician levels could improve the uptake of genetic testing, organ surveillance, and quality of care for persons with LS.

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## List of Abbreviations

| | |
|---|---|
| CAPP2 | Colorectal/Adenoma/Carcinoma Prevention Programme 2 |
| CRC | Colorectal Cancer |
| D&C | Dilation and Curettage |
| DMPA | Depot medroxyprogesterone acetate |
| DNA | Deoxyribonucleic acid |
| et al. | *et alii*, and others |
| EGD | Esophagogastroduodenoscopy |
| FDRs | First-degree relatives |
| IHC | Immunohistochemical |
| HNPCC | Hereditary nonpolyposis colorectal cancer |
| LS | Lynch Syndrome |
| MMR | Mismatch repair |
| MSI | Microsatellite instability |
| OCPs | Oral Contraceptive Pills |
| PCR | Polymerase chain reaction |
| PEG | Polyethylene glycol |
| SDRs | Second-degree relatives |
| TDRs | Third-degree relatives |
| UK | United Kingdom |
| US | United States |

# CHAPTER 1 INTRODUCTION TO THE THESIS

## 1.1 Introduction

Genetic testing for hereditary cancer syndromes is critical to identifying persons with a high cancer risk, who benefit from specialized screening or preventive care[1,2]. From 2005 onwards, an increase has been documented in referrals and completed tests for harmful mutations in the tumour suppressor genes – BRCA1 and BRCA2 – best known to increase lifetime susceptibility to breast and ovarian cancers[3-5]. The population prevalence of BRCA1 and BRCA2 in women diagnosed with breast cancer is 3%[6]. In persons diagnosed with colon cancer, this prevalence resembles the 2-3% prevalence of Lynch Syndrome (LS)[7], an inherited disorder that increases the risk of colon and other cancers occurring. Although the prevalences of these conditions are similar, numbers of referrals and completed genetic tests for LS have stayed relatively constant over time[5]. Thus, some barriers to persons participating in genetic testing and organ surveillance appear specific to, or stronger for, LS. Among persons with LS this qualitative study explores lived experiences of barriers to accessing genetic testing and organ surveillance in the Manitoba health system of Canada. Qualitative data, gathered through audio-recorded and transcribed semi-structured interviews, have been analyzed through a general inductive approach. The Manitoba population is unique because universal LS colorectal tumour testing for those aged < 70 years was implemented in 2013. This study includes persons diagnosed before and after this program was implemented and demonstrates past and current facilitators and barriers to testing. Through this person-centred research, a framework was developed whose implementation may reduce barriers to health care for persons with LS.

## 1.2 Lynch Syndrome Overview

Lynch syndrome (LS), also known as hereditary nonpolyposis colorectal cancer (HNPCC), is a cancer predisposition condition caused by autosomal dominant inheritance[8,9]. The predisposition is caused by a germline mutation in mismatch repair (MMR) genes including MLH1, MSH2, MSH6 and PMS2 or by a deletion in the EPCAM gene, which leads to methylation of the MSH2 promoter[10-13]. The population prevalence of LS is estimated at 1 in 440[7], although this estimate may be conservative[14]. The probability and type of cancer developing in persons with LS depend on which MMR gene is affected. The risk of colorectal cancer (CRC) by age 70 for PMS2 mutation carriers is 15-20 percent[15,16] whereas for MSH6 it is 18-69 percent[17], for MSH2 it is 39-68 percent[18] and for MLH1 it is 50-65 percent[19]. Women with LS have an additional average of 43 percent lifetime risk of developing endometrial cancer[15,20-22] (Table 1.). LS also predisposes people to an increased risk of other cancers: ovarian, small intestine, stomach, upper urinary tract, and pancreas (Table 1)[23,24].

Table. 1. Lifetime cancer risk related to Lynch Syndrome genotype

| **Lynch Syndrome Genotype** | **MLH1** | | **MSH2** | | **MSH6** | | **PMS2** | |
|---|---|---|---|---|---|---|---|---|
| Cancer Site | Female % | Male % | Female % | Male % | Female % | Male % | Female % | Male % |
| Colorectal | 45-53 [21,25,26] | 46-65 [21,25,26] | 33-68 [21,25,26] | 37-63 [21,25,26] | 26-30 [17,20,21] | 14-69 [17,20,21] | 15 [15] | 20 [15] |
| Endometrial | 34-66 [21,22] | - | 21-51 [21,22] | - | 16-71 [20,21] | - | 13-24 [15] | - |
| Ovarian | 12-20 [21,22] | - | 15-24 [21,22] | - | 0-1 [20,21] | N/A | - | - |
| Upper urologic tract | 0.4 [21,22] | 2.1 [21,22] | 9-19 [21,22] | 19-20 | 9* [21] | | - | - |
| Gastric | 6-17* [21,22] | | 2-6* [21,22] | | 2-4 [21,22] | 2-6 [21,22] | - | - |
| Small bowel | 6-17 [21,22] | 3-17 [21,22] | 6 [21,22] | 3-6 [21,22] | 2 [21] | 2 [21] | - | - |
| Pancreatic/biliary | 4-17 [21,22] | | 6* [21,22] | | 2 [21] | 2 [21] | - | - |
| Brain tumors (gliomas) | 1.7* [22] | | 2.5* [22] | | - | - | - | - |
| Sebaceous gland tumours | 42ǂ [27] | | 44ǂ [27] | | 0ǂ [27] | | - | |

NOTE. *Reported values not separated by sex, ǂ of families.

Medical management of LS depends on genetic testing and involves multiple organ surveillance[28]. A diagnosis of LS substantially impacts an individual's life, as knowledge of an elevated risk of developing cancer can distress the person with LS and their family. A positive test result presents challenges of deciding whether to communicate familial genetic status, to whom, when and how[29,30]. Annual or biannual colonoscopies with polypectomy decrease the incidence of CRC[31,32]. After childbearing is completed, prophylactic hysterectomy and bilateral salpingo-oophorectomy are recommended to eliminate the risk of endometrial and ovarian cancer occurring in women[33].

### 1.2.1 Muir-Torre Syndrome

Muir-Torre Syndrome is a variant of the autosomal dominant LS and is found in about 28% of the families with LS[27]. Muir-Torre is diagnosed when there is both an internal organ cancer related to LS that tested positive for an MMR mutation and sebaceous neoplasms[34]. The recommended organ surveillance for persons with Muir-Torre Syndrome is based on the recommendations for persons with LS and: annual skin evaluation for lesions suspicious of sebaceous carcinoma or keratoacanthorna, complete blood cell count, and measurement of tumour markers[35].

## 1.3 Genetic Testing for Lynch Syndrome

Genetic testing for LS occurs primarily through tumour samples. Since multiple genotypes can account for LS it is vital to identify in the tumour the exact one responsible before taking a blood sample to confirm the presence of LS. Thus the person with the cancer diagnosis in the family whose tumour and then blood have tested positive for LS is responsible for sharing or not

the possibility of genetic risk with family members. This person is referred to in the genetics literature as a 'proband' or 'index patient'.

### 1.3.1 Specifies of Genetic Testing: Microsatellite Instability

Genetic testing for LS relies on detecting a DNA mismatch repair (MMR) protein deficiency in the tumour sample[36]. This testing is based on microsatellite instability. Microsatellites are regions in the genome that repeat sequences between 1 and 6 base pairs and there are loops of inaccurately copied repetitive sequences. In a properly functioning MMR system, proteins bind to these nucleotide base mismatches and target them for repair[37]. The gene codes of MLH1, MSH2, MSH6 and PMS2 are for proteins that are essential for MMR to maintain genomic conformity. MMR corrects the nucleotide mismatches that have circumvented the usual editing function of DNA polymerase. Mutations in the above gene codes are associated with LS. In 2009, the EPCAM gene was found in families with LS-like cancers. This gene featured loss of MSH2 protein expression without a detectable MSH2 mutation[38,39]

### 1.3.2. Specifics of Genetics of Testing: Molecular Tumour Testing

Two types of microsatellite instability (MSI) detection methods have been established to detect MMR: MSI polymerase chain reaction (PCR) based detection and immunohistochemical based approach (IHC). PCR based analysis of microsatellite markers compares the increase of microsatellites (contraction or expansion in microsatellite DNA as a result of mutations in DNA MMR proteins) in a tumour to normal tissue[40]. This comparison test uses a panel of 5-10 microsatellite markers where tumours are MSI-high when 30 percent or more microsatellite sequences are mutated. Sporadic cases of CRC may display MSI-H phenotype and IHC testing should also be considered to evaluate for LS[41].

Despite screening reliably for mutations in tumours that result in protein degradation, IHC cannot detect mutant proteins (missense mutations) and wild-type polypeptides[42]. In the MLH1, MMR gene, one-third of mutations are missense mutations which cause mutant proteins and a false-normal staining pattern for MMR-IHC testing[43]. MSI –PCR testing is costlier than IHC testing in laboratory fees, with IHC costing 25 percent of MSI-PCR testing[44,45]. There is also the possibility of using an IHC test and adding PMS2 and MSH6 to the antibodies to obtain a more accurate staining pattern[43].

Once an IHC or MSI test is positive further genetic testing will be completed. Some people have these gene mutations only in their cancer cells whereas people with LS have the mutations in all their cells. A person with suspected LS would have a blood test to look for specific mutations in the tumour and subsequently in the blood. It is easier to identify these mutations if the individual or family member has had a tumour sample that detected the genotype affected.

### 1.3.3 LS Diagnosis Without a Tumour Sample

Clinicians use two diagnostic guidelines – Amsterdam II criteria and the revised Bethesda guidelines – to identify persons who may be at a high risk for LS and should be referred for genetic testing[46]. Rooted in persons having a detailed family medical history, these guidelines are for use only when a tumour sample is not available for testing. For a person to be tested for LS, the Amsterdam II criteria require three or more relatives with a LS associated cancer (e.g. colorectal cancer or endometrial cancer, small bowel cancer, or ureter cancer). In addition to the above criteria, the family history is required to exhibit: a cancer diagnosis in at least one relative before the age of 55, a cancer diagnosis in two or more successive generations, and the

possibility of familial adenomatous polyposis must be excluded. [46]. If the Amsterdam II criteria are met it is recommended that the patient be referred for LS genetic testing[46].

The revised Bethesda guidelines list multiple criteria but only one criterion must be met to refer the patient for genetic testing for LS. The criteria for the Bethesda Guidelines are described in Table 2. Several studies have found the revised Bethesda guidelines to be more sensitive than the Amsterdam II criteria in identifying persons with LS[47].

Table 2. Revised Bethesda Guidelines (2003)[46]

| One of the following criteria must be met to refer patient to genetic counselling |
|---|
| 1. Diagnosed with colorectal cancer before the age of 50 years |
| 2. Other LS related tumours (include stomach, bladder, ureter, renal pelvis, biliary tract, brain (glioblastoma), sebaceous gland adenomas, keratoacanthmoas and carcinoma of the small bowel), regardless of age. |
| 3. CRC with high-microsatellite-instability morphology that was diagnosed before the age of 60 years. |
| 4. CRC with one or more first-degree relatives with CRC or other LS related tumours. One of the of the cancers must have been diagnosed before the age of 50 years (this includes adenoma, which must have been diagnosed before the age of 40). |
| 5. CRC with two or more relatives with CRC or other tumours regardless of age. |

### 1.4. Genetic Testing in Manitoba

Before 2013, persons with a clinical suspicion of LS were identified through family history and referred to a medical geneticist but most did not qualify for genetic testing[48]. Prior to 2013, there was confusion around the criteria for genetic testing at the public level and the level of the Provincial Minister of Health (Appendix A). On March 1, 2011 the Minister of Health, Theresa Oswald replied to a concerned Manitoban, "Diagnostic Services of Manitoba Inc. does testing for Lynch syndrome in Manitoba" (Appendix A). The Heath Minister went on to write, "Contrary to media reports, should a Manitoban of any cultural heritage require testing for Lynch

syndrome, they can receive that testing" (Appendix A). A follow-up letter by the Health Minister on May 11, 2011 states, "…regrettably [I] have learned that inaccurate information was sent to you in my March 1, 2011, letter, regarding Lynch syndrome" (Appendix A). The Health Minister then outlined the correct criteria for genetic testing in Manitoba, "Only Manitobans where the proband has been tested elsewhere can have testing for a family-specific mutation" (Appendix A).

In 2007, two persons diagnosed with colorectal cancer, Kai Arnot and Sid Chapnick, came together to create a walk/run to raise funds for LS testing in Manitoba[49]. The younger than average age of Sid Chapnick's diagnosis had clinicians wondering about the possibility of LS but Manitoba was unable to test his tumour as he was the proband; he was concerned for the future health of his family members and other people in a similar situation (Chapnick, S, personal communication, August 24, 2017). The name of this event is the *Denny's Kick Butt Run/Walk* and throughout the 11-year history $334,856.55 have been raised for LS testing and research in Manitoba[49]. In 2013, Manitoba purchased genetic testing equipment for LS, in part due to the significant financial contributions of Denny's Kick Butt Run along with a dedicated group of medical professionals and patient advocacy[48].

Documentation from the Canadian Agency for Drugs and Technologies in Health demonstrates that Manitoba was the last province in Canada to implement LS testing[50]. The results of my study should be viewed in this context, as persons and families living in Manitoba were disadvantaged regarding prevention and treatment of LS cancers. In December 2016, Diagnostics Services of Manitoba expanded to include gynecologic (endometrial carcinomas) testing and they were planning on reducing the volume of MSI tests to move towards solely IHC testing[48].

### 1.5. Recommended Organ Surveillance

When a genetic test confirms at the molecular level a LS diagnosis this finding justifies the surveillance of multiple organs and possible surgical and chemoprevention management[51]. Persons with LS are at increased risk of developing colorectal and extracolonic cancer (Table 1).

#### 1.5.1 Recommended Organ Surveillance: Colorectal Cancer

The highest risk for both sexes is CRC, and thus distinctive characteristics of LS malignancies guide prevention. These features include younger age of presentation, predominance in the right colon, and faster polyp growth with a rapid adenoma-carcinoma sequence[51]. In several studies[52-55], frequent colonoscopy surveillance $\leq 2$ years has been linked to increased CRC detection at earlier stages and fewer CRC diagnoses. The United States (US) Multi-Society Task Force on Colorectal Cancer recommends CRC screening by colonoscopy for persons at risk (first degree relatives of those with LS) or persons with LS, every 1-2 years, beginning at 20-25 years or younger if family members' CRC diagnosis occurred before the age of 25[51].

#### 1.5.2 Recommended Organ Surveillance: Endometrial Cancer

Endometrial cancer is the second most common cancer occurring in persons with LS[51]. Cumulative lifetime risk of endometrial cancer ranges from 21 to 60 percent depending on the specific gene mutation (Table 1). Several annual screening options have been proposed: pelvic examinations, transvaginal ultrasound, endometrial sampling, and CA-125 testing. Studies of these interventions have not demonstrated a decrease in mortality, since 75% of LS females with endometrial cancer present with stage I disease and have a high survival rate[51]. Endometrial sampling seems useful in identifying asymptomatic women with endometrial cancer[56]. The US Multi-Society Task Force on Colorectal Cancer recommends offering endometrial cancer

screening annually to women with LS, through pelvic examinations and endometrial sampling from ages 30-35 years[51].

### 1.5.3. Recommended Organ Surveillance: Ovarian Cancer

In women with LS the estimated cumulative lifetime risk of ovarian cancer ranges from 0.3 percent to 20 percent (Table 1). Although no studies to date have been completed on the effectiveness of ovarian cancer screening for women with LS[51], the US Multi-Society Task Force on Colorectal Cancer recommends annual transvaginal ultrasound starting at ages 30-35 years[51].

### 1.5.4 Recommended Organ Surveillance: Gastric Cancer

The average lifetime risk for gastric cancer can be as high as 13 percent for persons with LS[51]. A time trend study of gastric cancer found an 8 percent risk for males and a 5.3 percent lifetime risk with a lack of familial clustering[57]. Gastric cancers in persons with LS appear to be the intestinal type[57,58] and could be potentially screened through endoscopy. The US Multi-Society Task Force on Colorectal Cancer recommends initial screening by esophagogastroduodenoscopy (EGD) with a gastric biopsy of the antrum between the ages of 30-35 years along with treatment of *H pylori* infection when found[51].

### 1.5.5 Recommended Organ Surveillance: Renal System Cancer

A Danish study[59] of 288 families with LS found: 54 urinary bladder cancers, 34 cancers of the renal pelvis, and 48 cancers of the ureter. Ninety percent of the tumours displayed loss of MMR protein expression. Persons with mutations in MSH2 were overrepresented (73 percent) and had an increased risk of developing urinary tract cancer when compared to mutations in MLH1 or MSH6. Joost *et al.*[59] recommended surveillance of urinary tract cancers for persons

with MSH2 mutations, yet there is a lack of research evaluating screening for urinary cancer in persons with LS[51]. The consensus is that the benefit of ultrasound screening is unknown, and urinalysis, which is inexpensive and noninvasive, is recommended[51].

### 1.6 Prophylactic Surgery

This section describes two prophylactic surgeries, which can be recommended for persons with LS. Prophylactic surgery is the surgical removal of an organ or gland showing no sign of cancer to prevent cancer occurring in the future[60]. This procedure is more common among persons with a high risk of developing cancer[41], such as those with LS.

#### 1.6.1 Prophylactic Surgery: Subtotal Colectomy

Subtotal colectomy has been offered to persons with LS after a CRC diagnosis and occasionally before a CRC diagnosis as a preventive measure[61]. The rate of colonic polyp progression to carcinoma appears more rapid in people with LS than in the general population, among whom progression is slow with an estimated timespan of 10 to 15 years[62]. Lanspu *et al*.[63] studied 225 individuals from LS families, of whom 10.2 percent had colorectal cancer diagnosed within five years of colonoscopy or colon resection. Various hereditary cancer institutions around the world recommend subtotal colectomy in persons with LS and CRC due to increased risk of metachronous CRC and the associated high risk of mortality[64]. A retrospective cohort study[64] found that patients who underwent limited resection rather than a subtotal colectomy, had a statistically significant (20 percent) increased risk of developing a second CRC. Patients with LS who underwent subtotal colectomy had fewer overall abdominal surgeries than those with only limited resection[64]. Even with subtotal colectomy, persons with LS require lifelong evaluation through endoscopy of the rectal segement[61]. Note that subtotal colectomy can have an impact on a person's quality of life and this variable should be considered in decision-making.

### 1.6.2 Prophylactic Surgery: Prophylactic Hysterectomy and Oophorectomy

Women with LS have a substantial risk of endometrial and ovarian cancer (Table 1). A study[33] retrospectively analyzed data on 315 women with LS, of whom some had undergone prophylactic gynecologic surgery. The results indicated that the women who underwent gynecologic surgery had zero cancers whereas in the nonsurgical group 33 percent developed endometrial cancer and 5 percent developed ovarian cancer[33]. The US Multi-Society Task Force on Colorectal Cancer recommends that hysterectomy and bilateral salpingoo-ophorectomy be advocated to women after childbearing is completed or at the age of 40 years.

## 1.7. Chemoprevention

Chemoprevention is defined as, "use of pharmacologic or natural agents that inhibit the development of invasive cancer either by blocking the DNA damage that initiates carcinogenesis or by arresting or reversing the progression of premalignant cells in which such damage has already occurred" (p.743)[65]. There is growing evidence that aspirin may reduce CRC in persons with LS[66]. Some research indicates that oral contraceptive pills (OCPs) could also be used as a chemopreventive option in women with LS but this option must be weighed against a possible, small increase in breast cancer[67].

### 1.7.1 Chemoprevention: Aspirin

The Colorectal/Adenoma/Carcinoma Prevention Programme 2 (CAPP2) was a randomized placebo controlled trial that investigated the effect of Novelose (resistant starch, 30 g) and aspirin (600 mg) over four years on the development of adenomas and cancer in persons with LS[68]. The intention-to-treat analysis did not show a reduction in CRC incidence but secondary analysis reached statistical significance. Participants who completed two years of treatment with aspirin experienced a 50 percent reduction in CRC[68]. Currently, no North American guidelines

exist on the clinical management of LS patients in regards to chemoprevention, while European Guidelines recommend offering aspirin (≤ 100 mg) to prevent CRC[32]. The optimal dose of aspirin has not yet been found and the CAPP2 trial has been expanded into CAPP3, a non-inferiority trial comparing doses of aspirin in 100 mg, 300 mg and 600 mg, among 2000 people with LS in the United Kingdom (UK). Results from CAPP3 trial are expected in 2020.

### 1.7.2 Chemoprevention: Oral Contraceptive Pills (OCPs)

The use of OCPs has been associated with statistically significant reductions in the following malignancies: ovarian, endometrial and colorectal cancers[69,70]. A detailed meta-analysis published in 1995[71] and the updated version published in 2013[69] reported risk reduction for endometrial cancer of 0.57 (95 percent Confidence Interval 0.43-0.76) with the use of OCP, and risk reduction increased the longer that OCPs were used. Limited research has been conducted regarding the use of OCPs in women with LS. Lu *et al*.[72] reported the effects of OCPs on biomarkers of endometrial cancer in women with LS. This multicentre randomized controlled trial in women diagnosed with LS compared medroxyprogesterone acetate (DMPA) to OCPs. The results provided mechanistic support that OCPs could facilitate chemoprevention in women with LS[67,73]. The recommendation of OCPs for women with LS should be considered in the context of age, sexual relationship characteristics, and preference to have prophylactic surgery[67].

## 1.8 Summary of Background Information

Providing background information to understand LS, this section has demonstrated many complexities in both receiving the genetic diagnosis and preventive care management. First, the five possible genotypes of MMRs necessitate initial genetic testing with a tumour sample. Also each mutation has a different lifetime risk of cancer, which requires a personalized organ surveillance approach. The second complexity demonstrated is that multiple organs have an

increased risk of developing cancer, particularly in women, which requires care from clinicians with specialist knowledge of LS. Thirdly, prophylactic surgery and chemoprevention add other levels of coordination and knowledge to screening procedures. It is necessary to understand through this background review of LS, all the complexities within genetic testing for LS and the medical management involved for LS care, so that a realization of the extent of possible barriers and facilitators can be conceptualized. The complexities shared in the background review add credence to the value of my research. The research identified in this thesis will demonstrate what barriers and facilitators a sample of persons with LS experience in receiving care. This research will provide a framework of what could be implemented for persons with LS to balance the genetic testing cost benefit ratio by ensuring access to care[74].

### 1.9 Goals and Rationale for the Study

#### Goals

**1.** The first goal for my study is to determine the barriers and facilitators to genetic testing and organ surveillance, as identified by persons with LS, in the same health system.

**2.** The second goal for my study is to develop a framework to improve access to health care for persons with LS.

#### Rationale

A few studies have investigated barriers to multiple organ screening[75,76], colon screening[77], and gynecological screening[78] for persons with LS. A Canadian study[75] indicated barriers associated with clinicians' lack knowledge of LS and the uncoordinated publicly funded health system. No studies have explored perceived barriers to genetic testing and organ surveillance in the same health system for persons with LS. The focus of many studies has been on identifying

barriers to communicating a diagnosis of LS to family members[79-82]. Personal theories of hereditary conditions and perceived cancer risk in the communication of LS status have also been studied[83,84]. From my knowledge of the literature there seems to be a gap in the literature where persons with LS share their experiences in accessing health care as well as their suggestions to improve access to health care. In addition, exploring barriers to genetic testing and organ surveillance in the same health system may identify whether persons with LS receive sufficient health care for the genetic testing cost benefit ratio to balance[74].

It has been identified that health care needs a research approach that is pluralistic and takes into account multiple perspectives and complex dynamics[85]. The approach that is chosen to contemplate the multiple perspectives impacts what is valued, and thus shared. Through utilizing the definition of person-centred health care by Buetow (2016)[86], my study encompasses a values-based ethic of virtue, which respects the personhood of participants, and thus their perspectives.

Using the ideas shared by persons with LS in combination with the literature, I will seek to achieve the second goal of my study: the development of a framework to improve access to health care for LS. The implementation of the framework at the health system and clinician level could steer resources to improve the uptake of genetic testing, adequate organ surveillance, and provide high quality care for persons with LS.

# CHAPTER 2 LITERATURE REVIEW

## 2.1. Introduction

This section first reviews literature about the frequency and predictors of uptake of genetic testing among relatives of the proband. Studies exploring the communication of genetic status

between family members are also included. The second portion of the review focuses on experiences of organ surveillance among persons with LS. The third section of the review focuses on literature to connect my study to disability studies.

### 2.2 Literature about the Uptake of Genetic Testing in Families with LS

It has been recommended that all newly diagnosed persons with CRC receive molecular tumour testing for LS[87]. The number of first degree relatives (FDRs) tested per proband has been identified as a key determinant of the cost effectiveness of this approach[74]. A minimum of three family members need to be tested to create a cost effective system[74]. Thus, it is important to understand and address the reasons for the limited uptake of genetic testing in some families with a LS diagnosis. In relation to my study, it is important to remain cognizant of the barriers described by the literature that limit uptake to genetic testing, as this is the first step in achieving health care for LS.

A systematic narrative review was published in 2013 on the frequency of, and factors associated with, the uptake of genetic testing of FDRs of a family member newly diagnosed with LS[88]. The review reported four main conclusions. First, a substantial proportion of the FDRs did not undergo genetic testing. Four of the eight studies[79-82] included in the review reported the number of notified family members that received genetic testing in a specialized clinical setting and the rate of genetic testing was 52 percent or less. Since these studies were carried out in well-established cancer registers that would be expected to facilitate the contact of relatives, the authors of the review suggested that uptake in community settings would be lower[88]. The implication of the limited uptake of genetic testing is that persons who could benefit from organ surveillance are not receiving this opportunity and their likelihood of a preventable cancer

diagnosis is increased[88]. Secondly, fewer than three FDRs were tested in three of the same four studies[79-82]. Thirdly, health system concerns about insurance, privacy, discrimination and cost may affect the uptake of genetic testing. Two studies[79,89] in the US raised concerns about how a diagnosis of LS would affect insurance coverage and employer discrimination. The fourth conclusion related to personal characteristics that influenced the uptake of genetic testing. People were more likely to participate in genetic testing if they were: ≤ 50 years of age, female, a parent, college educated, did not have depressive symptoms, and had multiple family members diagnosed with cancer.

### 2.3 Literature about Familial Communication of LS Diagnosis

Several factors facilitate communication of familial genetic status: well-functioning relationships, positive attitudes, and support from partners[90,91]. Suboptimal communication has been connected to distant family relationships or lack of relationship with family members, and decisions to restrict disclosure of information. This section of the literature review provides context, as the results of my study and framework indicate a need for an enhanced communication technique to share a LS diagnosis with blood relatives and I suggest a method to do so in the framework.

Some studies have identified factors associated with communicating genetic test results to close and distant family members. Stoffel *et al.* (2008)[91] surveyed 162 persons who had been tested for LS to see how many disclosed their genetic test result to FDRs, second degree relatives (SDRs) and third degree relatives (TDRs). Almost all (98 percent) respondents reported sharing their test result with one or more FDRs and 67 percent reported sharing the result with SDRs or TDRs. The most cited reasons for sharing a genetic test result with family members were: (1) to

inform them of their risk, (2) to encourage testing, and (3) to obtain emotional support[91]. The most cited reasons for not sharing a genetic test result were: (1) not being close to family members, (2) concern that family members would not understand the test result, and (3) not wanting relatives to worry[91]. When the genetic test for LS was positive, 75 percent of respondents shared this result with one or more SDRs or TDRs. In the case of an 'indeterminate' genetic test result (which occurs when the test result is not clearly negative or positive), the disclosure to SDRs or TDRs was 57 percent. These two results may indicate that persons with LS encounter barriers to communicating genetic status to distant relatives[91].

Bartuma *et al.* (2012)[84] conducted in-depth qualitative interviews with 27 people from nine LS families to assess how their perspectives of hereditary cancer affected their ability to manage communication of an increased cancer risk and genetic testing. In families where cancer was diagnosed at a relatively young age, participants were more likely to undergo genetic testing and organ surveillance at an early age. In contrast, young people in LS families where cancer developed at an older age postponed or did not participate in genetic testing or organ surveillance[84]. The key communication difficulties reported were: (1) grasping the complexity of information during genetic counselling, (2) uncertainty around how to bring up the issue with adult children, and (3) dysfunctional relationships[84]. These difficulties put a large burden on families and especially on female family members who were found to be responsible for coordinating information about LS and providing support. The authors recommended providing updated information and education to families with LS, specifically where cancer was less common or occurred at an older age[84]. My study is in agreement with the authors, as the framework I created suggests that persons with LS be provided with up to date guidelines.

Palmquist *et al*.[83] produced a qualitative study of communication networks in regards to genetic test results and identified that the possibility of creating fear and worry in other family members was a reason for not discussing genetic information with them[83]. This finding has significance to my study, as when participants made the choice to share results with family members, it demonstrated their commitment to justice was tantamount to producing fear. When communicated to family members, a positive genetic test could meet with denial, limiting uptake of genetic testing and organ surveillence[83]. Families who shared genetic test results and participated in organ screening perceived a high risk of cancer. Other studies[83,84,91] have shown that a genetic counsellor advising people to inform family members of a positive result for LS is insufficient for this action to occur, which demonstrates a need for enhanced communication. The framework I created in my study suggests a method for an enhanced communication technique as identified by the participants.

### 2.4 The Experience of Organ Surveillance in Persons with LS

Even though organ surveillance reduces mortality from CRC[31,92], there is suboptimal participation in organ surveillance for persons at high risk of developing CRC[93-95]. For persons with LS, participation in colonoscopy ranges from 53 to 100 percent[95-100], transvaginal ultrasound ranges from 69 to 86 percent[96,100,101] and endometrial biopsies hover around 54 percent[101]. Reasons for these levels of participation include deficits in clinician knowledge and screening for LS[93,102]. Limited continuity, poor coordination of care, and insufficient access to screening services can also hinder organ surveillance[103]. However, to my knowledge there remains a gap in the literature, where the suggestions of persons with LS are utilized to improve access to health care.

Watkins *et al.* (2011)[75] conducted semi-structured interviews with 23 persons with LS in Newfoundland, Canada. The participants were asked questions about, "cancer in the family (first awareness of hereditary link, perceived personal risk, screening motivation) and genetic testing (decision-making, counseling experiences, reaction to status, understanding implications, impact on family)" (p.3) [75]. Organ screening experiences were asked about in a follow up interview. Emotional forces were found to impact a person's willingness to participate fully in organ surveillance. Scheduling appointments and waiting for results were time-consuming and draining. In terms of barriers encountered in health care an unstated number of participants perceived physicians to have little knowledge of LS and some felt that endoscopists were unaware of the current flat polyp literature, a barrier also noted by participants in my study. A lack of communication between specialists was also identified. All participants supported a coordinated approach to LS management[75]. The authors concluded that, "the current uncoordinated, physician dependent organization of screening for individuals with Lynch syndrome in Canada is inadequate" (p. 8)[75]. Unfortunately, the authors did not propose a framework to improve access to health care for persons with LS, so this remains a gap in the literature.

Burton-Chase *et al.* (2014) conducted interviews to elicit information about how women with LS interacted with the health system in regards to gynecologic cancer surveillance[78]. The 74 participants were part of the Texas MD Anderson Cancer Centre LS registry. Half of the participants reported that their primary care provider and specialists did not inform them or implement high-risk gynecologic cancer screening. For example, gastroenterologists performing colonoscopy screening were identified as not knowing the risk of gynecologic cancer for women

with LS. Primary care providers often had not heard of LS before meeting the patient with LS. A minority of women with LS were referred for gynecologic screening. Yet, most participants did not express concerns about the overall care received from providers, which may indicate that women with LS do not fully understand their risk for gynecologic cancer[78]. Also, when communicating with providers some participants did not use the term 'lynch syndrome' but rather explained themselves in terms of family history of CRC even though they had a positive genetic test indicating LS. Without an integrated health system and universal access to medical records, the authors conclude, "it is mainly the responsibility of patients to effectively communicate or inform providers of their familial health risks" (p. 189)[78]. The study described above highlighted the need for me to implement tailored questions for women with LS in my study's interview guide.

### 2.5 Literature about Persons Who Decline Genetic Testing for LS

Keogh *et al.* (2017)[104] studied persons who had been offered genetic testing for LS but chose to decline. This study occurred in Australia where tumor testing for LS is routine. A previous study by the same authors[105] identified that the participation by family members in genetic testing for LS ranged from 56 to 86 percent. Semi-structured interviews in the 2017 study were conducted with 31 persons who declined genetic testing for LS after a family member tested positive. Four participants were uninformed that genetic testing was available for hereditary colon cancer, although the cancer registry sent letters to their last known address. Participants receiving a letter either did not understand it or believed that cancer could not be hereditary and depends on lifestyle. Nine participants reported a 'weak intention' to participate in genetic testing. One participant was concerned about how health insurance would be affected, while two participants did not think a genetic test would change anything, and six participants did not think

genetic testing was of enough value to spend time scheduling, traveling and participating in the test. Eleven participants identified as definitive decliners with no interest in genetic testing because of insurance concerns (*n*=5) and their belief that results would cause anxiety (*n*=4) or not change anything (*n*=2). This study by Keogh *et al.* (2017)[104] identified that a lack of understanding of how LS testing could positively impact the future of the participants was a major reason for the decline. This deficit in understanding may indicate there is a lack of familiarity of what LS is and how it can be managed, in the public sphere.

### 2.6 Literature Summary of Relevant Lynch Syndrome Studies

This literature review has reported the small number of qualitative studies that have explored barriers to genetic testing and organ surveillance for persons with LS. However, there is a paucity of literature connecting perceived ease of access to genetic testing and ease of coordination for organ surveillance in the same health system. Understanding this connection is vital because if persons with LS identify barriers to genetic testing and organ surveillance in a health system regularly testing colonic tumours for LS, the testing may be redundant since persons cannot adequately participate in disease management following diagnosis. Studies exploring familial communication focus on how or if the proband notifies family members about genetic testing. No published qualitative study asks persons with LS what they believe prevented family members from going for genetic testing in terms of the health system. Identifying whether the barrier is predominately the method of communication, health system barriers or a conscious rejection of genetic testing could steer resources into improved uptake of genetic testing. Based upon this literature review my study's interview guide focused on communication between

family members, health care for women with LS, and barriers and facilitators in receiving LS genetic testing and organ surveillance at the health system level.

### 2.7 Literature to Connect the Study to Disability Studies

Persons with LS who participated in my study did not use the term 'disability' or 'chronic disease' in relation to LS or themselves and I am uncomfortable giving persons labels that they themselves have not used. Furthermore, literature focusing on LS does not mention the term 'disability' or 'chronic disease'. However, this finding does not preclude translating the concepts developed in disability studies into an approach that disrupts traditional research norms that view persons not as persons, but rather in terms of the disability or health condition. My study utilizes person-centered health care as the main approach in addition to concepts from the social model of disability to respect and dignify the personhood of the participants. This section of the literature review presents definitions of disability and chronic disease, models of disability, and links concepts in disability studies scholarship to person-centered health care.

#### 2.7.1 Definitions of Disability and Chronic Disease

The definitions associated with 'disability' and 'chronic disease' may assist in explaining why these terms have not been used in relation to LS. The World Health Organization in the World Report on Disability[106] states, "disability is an umbrella term, for impairments, activity limitations, and participation restrictions" (p.7). A possible linkage to LS could be made regarding 'participation restrictions', however the restrictions that persons with LS experience are in the form of specialized health care and not basic health care or employment restrictions as experienced by many persons with disabilities. The lack of societal participation restrictions experienced by persons with LS prevents a direct parallel being formed with persons with

disabilities, as the magnitude of societal restrictions in daily living experienced by many persons with disabilities is often unfortunately, more impactful. However, there are similarities between some experiences of persons with LS accessing health care and persons with disabilities accessing this care, which the concluding chapter explores.

There are many definitions of chronic disease, but for consistency I returned to The World Health Organization, which states that,

" noncommunicable diseases also known as chronic diseases, tend to be of long duration and are the result of a combination of genetic, physiological, environmental and behaviour factors. The main types of noncommunicable diseases are cardiovascular diseases (like heart attacks and stroke), cancers, chronic respiratory diseases (such as chronic obstructive pulmonary disease and asthma) and diabetes".

The World Health Organization lists cancers as a chronic condition. In relation to my study, some persons with LS develop cancer and some persons with LS do not. Cancer often requires treatment in the form of surgery, medication, chemotherapy, or radiation; whereas, health care specific to LS requires organ surveillance and possible surgeries to prevent cancer. The question that comes to my mind is, " does being predisposed to the possibility of cancer warrant the classification of chronic disease?". Unfortunately, it is beyond the scope of my study to determine whether persons with LS identify LS as a chronic disease. However, connections can be made between some of the experiences of those with chronic disease who also identify as having disabilities and the experiences of persons with LS, which the concluding chapter explores.

### 2.7.2 Models of Disability

The medical model of disability is reinforced by the "personal tragedy theory of disability" and the medicalization of disability[107]. The medical model views the individual with disability as having a "problem" that needs to be fixed [108]. In contrast, the social model refers to the concept expressed by Paul Hunt of the British organization Union of the Physically Impaired against Segregation in the early 1970s which Mike Oliver further refined in 1990[109]. The social model of disability views disability as a social concept caused by barriers in society, not based on an anatomical or physiological state of a person's body[110]. The social model strongly argues that disability is mostly constructed by society; people are disabled by societal structures and attitudes not by the physiological or anatomical impairments[111]. Shakespeare and Watson (2001)[112] furthered the original social model by clarifying that societal barriers must change to enable people living with impairments to participate on an equal basis in society. There has been criticism of the social model, particularly in terms of illness, as explored in the next subsection.

### 2.7.3 Criticism of the Social Model

The social model of disability relies on a sharp distinction between impairment and disability, in order to separate the medical model from the social model. The dichotomy between the two models was historically positive as it reduced the medicalization of disability, where disability is regarded as unfortunate. Yet, as time progresses, ideas and models progress as well; for example in 1996, Liz Crow, stated, "our criticism[s] of the medical model of disability have made us wary of acknowledging our experiences of impairment" (p.209)[113]. Crow emphasized the need to focus on disability and impairment, as "impairment *in itself* can be a negative, painful experience" (p.219)[113]. In connection to Crow (1996), my study provides the opportunity for persons with LS to share experiences that happen outside and within a clinical setting that may

have been painful or negative. As a researcher I do not shy away from negative experiences that participants share, as these are part of their personhood, and the personhood of the person is paramount.

Susan Wendell wrote in her book, *The Rejected Body*, "I want to have more energy and less pain, and to have a more predictable body…I would joyfully accept a cure, but I do not need one" (p.83-84)[114]. This preference is expressed in relation to Wendell's own chronic illness, which is of value as it promotes, "different ways of understanding and appreciating the world, of conceiving the self, of questioning the foundations of "normal" (p.30)[115]. My study incorporates the same sentiment as it respects the personhood of the participants. Hence, concerns and narratives shared by persons with LS, were not only grouped into themes but further valued by incorporating their experiences into a framework whose implementation at the health system and clinician levels could improve the uptake of genetic testing, organ surveillance, and quality of care for persons with LS. My study was conceptualized to remove the possibility of tokenism towards persons with LS, as the framework whose foundation was built upon participants' experiences could be pragmatically implemented. The participants' narratives are so valued that I consider those experiences to be a catalyst for change.

### 2.7.4 Perspectives of the Social Model and Person-Centred Health Care

Authors have articulated the importance of integrating of the social model of disability into medical practice[116,117]. Within disability studies scholarship, Goering (2015) writes that through the social model, "we must ensure that medical professionals talk candidly about negative impairment effects while maintaining full respect for individuals with disabilities" (p.135)[118]. However, person-centred health care considers respect and dignity for a person as a value-based ethic of virtue; caring in health care is a moral value and ethical practice, central to

the concerned attention to the concrete needs of others and oneself [86]. It is my understanding that the moral value and ethical practice of clinicians will not be transformed by the social model as Goering argues, as this model does not engage with the fundamentals of ethics, which create the foundation of a health system; rather the social model could be used as a tool. The question that this possibility raises is what can revolutionize the ethics of the health system? Answering this question is beyond the scope of my study but it is a complex question which is worth investigating by others. Goering goes on to articulate that the social model can be used to eradicate the view from the non-disabled clinician that the impairment itself is negative, and transition to a new perspective that the, "main and sometimes only disadvantage of the impairment is not physiological, but social; the ugly and unwelcoming attitudes …and the physical impediments to access all make people suffer" (p.137)[118]. In this respect, the social model is powerful in identifying the ways that aspects of society can be disabling.

At the root of negative attitudes towards disability is the view that disability precludes a person from achieving social equality. Equality consists of the fundamental idea of equal respect and the equal dignity of all persons[119]. For my study to honour a commitment to equality, I am utilizing a person-centred approach, described in the next subsection, whose moral respect for the inviolable dignity of persons is consistent with the social model.

### 2.7.5 Person-centredness in Health Care

There are many definitions of person-centredness[120].The definition that I identify with is from Buetow (2016)[86], "person-centred health care socially valorizes the person as being above divisions of role, function and social and health status"(p.126). This definition constructs persons as whole beings who happen to perform social roles such as a patient and a clinician. These roles

do not define them as persons, whose lives and relationships are at the heart of health care. In contrast, patient centred health care reduces the person to a role, such as a patient or clinician[86]. It also emphasizes the patient as a person even though not all persons are patients.

Fundamental to person-centred health care is the observation that the term patient reduces a person to their social role and potentially to a disease and is not related explicitly with their personhood. 'People-*first*-language' as evident in disability studies, utilizes the term 'people with disabilities', rather than 'disabled people', which puts, "people's personhood ahead of their disabilities and describes what they have, not who they are" (p.30)[86]. The recalibration through language helps to disrupt the singular social role of the patient in health care. My study commits to the personhood of the participants by using the term 'persons with LS', and I maintain this perspective throughout, for example in conceptualizing the study, interviewing, analysis, and writing, so that each person is recognized as whole within their complex life project.

My person-centred study of LS recognizes the unique dignity of persons who are patients without this social role limiting their personhood. Hence, the interview questions focused on participants' experience in the health system but also their interaction with others such as family members.

A truly person-centred approach to LS care would also explore the experiences of clinicians as persons, also at the centre of health care, whose welfare is closely integrated to the welfare of the patient[86]. However, it was beyond the scope of this small study to explore the experiences of all persons participating in the LS journey. My study explores solely the experiences of the persons with LS and not the clinician and others caring for them.

# CHAPTER 3 METHODOLOGY

## 3.1 Philosophical Orientation

The selection and investigation of a research topic reflect basic assumptions by the researcher about the philosophy of knowledge and how knowledge is attained. Describing the influence of the researcher is vital to for credibility and trustworthiness in the research[121]. I assume that human knowledge captures a portion of reality, as the existence of a world without a human mind is conceivable, reality is not solely constructed through and within human knowledge. I assume that persons with LS can only know what is probably true from their lived experience of facilitators and barriers to genetic testing and organ surveillance can yield context-dependent generalizations.

## 3.2 Study Design: Interpretive Description

Interpretive description as outlined by Thorne (2016)[122] was selected as a study design owing to its potential for tentatively developing an interpretive account of behaviour from description. This design offered me an opportunity to answer the lines of research inquiry and generate knowledge that might improve health care for persons with LS. It extends the use of qualitative description as described by Sandelowski[123] to ascertain the "who, what, where" of events, by enabling the researcher also to consider the "why". In addition, interpretive description values clinical interpretation as foundational knowledge, supports purposeful sampling, does not rely on direct observation, encourages comparisons of differences in experience, and generates health care practice knowledge as its goal.

## 3.3 Lived Experiences as a Study Formation Tool

Before the study commenced, I attended the monthly meetings of Lynch Syndrome

Canada to gain an understanding of what persons with LS thought the focus of my research should be. My involvement at the Lynch Syndrome Canada meetings was limited and within the bounds set by the Research Ethics Board. I was only an observer and listener to the ideas and concerns raised at the meetings, ensuring that no one felt obligated to participate in my study. The meetings were centered around planning for the annual LS day on March 22 and fund-raising opportunities. I did consult with the President of Lynch Syndrome Canada and four other members who identified that the most pressing issues were: access to genetic testing, and organ surveillance in the Manitoba health system not adhering to the National Comprehensive Cancer Network® Guidelines in the absence of Canadian guidelines. Other key issues, in their experience, included family physicians lacking knowledge of LS. Based on this consultation and my comprehensive literature review it was decided to focus the interview questions on experiences of genetic testing and organ surveillance. Following interviewing the members of Lynch Syndrome Canada, I continued to attend meetings and became more involved as there was no longer the possibility of my involvement causing coercion. I became one of the lead organizers for the 2018 March 22 Lynch Syndrome Day and sought guest speakers and partnered with the University of Manitoba to host the educational evening.

### 3.4 Participant Inclusion and Exclusion Criteria

The criteria for inclusion in the research are as follows:

1. Persons who have been diagnosed with LS and know which MMR gene is associated with the mutation.

2. Persons who have received a genetic test result for LS in Manitoba; OR organ surveillance for LS in Manitoba; OR have received a cancer diagnosis in Manitoba which was later found to be from LS.
3. Persons who can speak and understand English.
4. Persons with the cognitive ability to consent to participate in this study and aged > 18 years.

The exclusion criteria for the research study are:

1. Persons who do not meet the above criteria.
2. Persons who have moved to Manitoba within the previous 12 months but have not interacted with the health system regarding organ surveillance or genetic testing for LS.

### 3.5 Sampling Strategy and Recruitment

#### 3.5.1 Purposeful Sampling

This study uses purposeful sampling[124] to select the participants most relevant to addressing the study question. The participant inclusion criteria ensured all enrolled participants had experienced a minimum of one organ surveillance procedure in Manitoba. The participants represent the study phenomenon but not the population from which they were selected. The logic and power of such sampling lies "in selecting information-rich cases for study in depth...[of] those [cases] from which one can learn a great deal about issues of central importance to the purpose of the inquiry"[125] (p. 169). In-depth analysis can then give readers sufficient information to decide whether the study findings are transferrable to their setting. Transferability depends therefore on logical inference rather than statistical inference.

Although I had planned on selecting equal numbers of males and females, I did not know the gender of participants at the outset and most of the persons who expressed interest in taking part

in the study were female. I used this selection bias as an opportunity to explore the experience of women in relation to the research question[125].

### 3.5.2 Recruitment

Recruitment of participants began with a letter (Appendix B) that introduced the researcher and explained the study. The letter was sent electronically via email from the President of Lynch Syndrome Canada to all members and/or attendees of the organization's events who requested to be on the email listserv. It was then at the discretion of these anonymized contacts to contact me directly or not. The second method of recruitment involved providing the same letter in a physical format in-person to the attendees of the 2018 Lynch Syndrome Day Event. Thus, the sample was drawn from a population that had at minimum an interest in the latest information about LS. The results should be viewed with this constraint in mind because advocacy by knowledgeable persons with LS for their organ surveillance raises concern regarding how other persons with LS fare in having organ surveillance performed.

## 3.6 Data Collection

Consistent with the research design, the data for this study were collected through semi-structured, in-depth interviews in which participants answered open-ended questions about their LS diagnosis and interactions with the health care system in regard to LS. The interviews were personal encounters in which, "open, direct, verbal questions are used to elicit detailed narratives and stories" (p. 316)[126]. All interviews occurred face-to-face either in a small seminar style room at the University of Manitoba or in the interviewee's home.

Each interview covered the same core topics around experiences in receiving health care specific to LS. However, the wording and order of individual questions varied across interviews

since qualitative inquiry aspires to be naturalistic and emergent[125]. The approach allowed flexibility in relating the interview to each participant's circumstances and avoided the risk of standardized questions constraining the participant's responses[125].

Each audio-recorded interview lasted approximately 30 to 60 minutes, as recommended by Kvale [127]. The interview guide was crafted to introduce the interviewer and interviewee to each other and create rapport. The questions asked were subject to skeptical peer review by my committee members to check that the questions were respectful and culturally sensitive. Although interviews as a source of data demonstrate what people say rather than necessarily do[128], interviews elicit information about experiences not obtainable through other means such as observation or a survey. Demographic information such as gender, age, relationship status, year of diagnosis, sharing of LS status, cancer diagnosis, current organ surveillance, and chemoprevention was also collected from each participant (Appendix C).

### 3.6.1 Pilot Interviews

The interviews were audio-recorded with participants' consent. A pilot interview[127] was conducted with one participant who was interested in the study and had experienced a complex trajectory in obtaining genetic testing for LS. This complexity was rooted in receiving a cancer diagnosis before the identification of LS and being female, which increases one's lifetime cancer risk (Table 1). The pilot interview allowed for a conversation about the clarity and acceptability of the questions, the burden they put on participants, and how to improve the questions.

### 3.6.2 Interview Questions

The interview guide (Appendix D) has five parts. The first part is a common introduction that I read to each participant prior to asking questions. It provided an overview of the study and indicated that there were no right or wrong answers. Secondly, a general question asked about how the participant first gained knowledge about LS. This question was intended to help them feel comfortable speaking about LS. The third part consisted of questions about genetic testing. The fourth section was about organ screening and health care recommendations for LS. The fifth part consisted of closing remarks to end the interview.

Rapport was easily established and at the end of the interview, each participant was invited to contact me directly if they had any concerns about their interview. Several participants contacted me following the interview for other reasons, such as expressing gratitude for the positive experience they had in speaking with me about their familial condition, providing updates on previously shared health conditions, or asking about the progress of the thesis. The ease of communication implied by these interactions adds credence to the interview data, given that the better the rapport established, the better the data are likely to be[129]. In addition, each participant agreed to be recontacted to clarify anything stated in the interview as needed and assist with preliminary analysis.

## 3.7 Data Analysis Overview

I transcribed the audio-recordings of the interviews verbatim. I have transcribed interviews professionally before for qualitative health research conducted within the Department of Psychiatry, McGill University and I currently transcribe professionally in my evaluation position for Research Manitoba. In producing and reading the transcripts of the interviews for this study on LS, I became deeply familiar with the data and generated a preliminary thematic coding

scheme through a general inductive approach. [130] A committee member (SB) was consulted during this process and read some transcripts to enhance the trustworthiness of their thematic interpretation.

### 3.7.1 General Inductive Approach

The thematic coding scheme was developed through a general inductive analysis.[130] This approach allows "research findings to emerge from the frequent, dominant, or significant themes inherent in raw data"[130]( p. 238) rather than from a specific hypothesis, theory or model postulated *a priori*. The general inductive approach suits studies like mine that are interested in identifying what has worked well in a program and what could be improved[130] by understanding and managing barriers and facilitators in the health system for persons with LS. Themes were developed by indexing and grouping text that I had coded to label interactions with the health system for LS genetic testing and organ surveillance, and experiences with clinicians. Interpretation enriched understanding of the themes and their relationships with the research objective. The latest version of the software package Nvivo 12 was used to aid data management.

## 3.8 Quality Considerations

In traditional or rationalistic research approaches, validity, reliability and generalizability have reached the status of a "scientific holy trinity"[127] (p. 229) that distinguishes quantitative research from qualitative research. To maintain research quality, 'The Consolidated Criteria for Reporting Qualitative Research' guided reporting of the findings and critical apprasial[131].

### 3.8.1 Credibility and Trustworthiness

Credibility, according to Guba & Lincoln (1989)[121], refers to the idea of "isomorphism between constructed realities of respondents and the reconstructions attributed to them" [121](p. 236). In this study, credibility and trustworthiness were increased through:

1. **Reflexivity.** I completed a journal to reflect self-critically on how 'being me' influenced this research and changed me. I considered how I see myself and imagined how others see me.
2. **Prolonged Engagement**. To contribute to "substantial involvement at the site of the inquiry" [121](p. 237) the research involved 17 participants over a period of seven months.
3. **Triangulation**: Different strategies (e.g. interviews and observation; different team members from disciplines) were combined to look for differences and similarities between the study findings.
4. **Audit trail**. I produced a written record (example in Appendix D) that clearly documents the research process to hold it accountable to critical scrutiny. This record includes my note taking from interviews and memos. Use of participant quotations further increased transparency around how the research took place.
5. **Peer debriefing and skeptical peer review**. These processes allowed me to discuss emerging themes with SB. Guba and Lincoln (1989)[121] describe the importance of the peer debriefing process in that it affords the researcher an opportunity to discuss "one's findings, conclusions, tentative analyses" (p. 237) and test out working hypotheses.
6. **Participant checking**. Guba and Lincoln (1989) describe participant checking under the heading of 'member checking' and write that these checks are, "the most crucial techniques for establishing creditability" (p. 314) in a study. Participant checking consists of taking the data

and interpretation back to participants so they have an opportunity to comment and check results[132].

Trustworthiness was achieved through transferability and dependability. According to Lincoln and Guba[133] (p. 299) dependability "seeks means for taking into account both factors of instability and factors of phenomenal or design induced changes" which means that the degree to which the data change through the interpretation phase is recorded and justified. Transferability refers to, the extent to which readers determine that the findings can be transferred to their setting or group[134]. A detailed description of the sample size was provided to aid transferability to other similar situations[135]. To facilitate transferability in the results section, a clear description of the culture in Manitoba health system has been provided as well as characteristics of the participants.

### 3.8.2 Reflexivity

A researcher's background influences the study design, questions, findings, and analysis. My background is multidisciplinary; ranging from physical geography, interdisciplinary disability studies, and internal medicine. My training has provided me a grounding in the scientific method, empirical evidence, and the value of lived experiences. My role as a research coordinator and later published researcher in gastroenterology created a depth of knowledge in all aspects involved in colonoscopy, which in turn influenced the questions in the interviews. In addition, I am a female and this has influenced both the questions I asked regarding gynecological surveillance, prophylactic hysterectomy, and bilateral oophorectomy; and likely the comfort level of female participants in sharing their experiences with me. The study design

and analysis were rooted in my view that personal narratives are so valued that they can be a catalyst for change.

The researcher is responsible for reflecting on these processes through reflexivity. The root of the word 'reflexive' means to 'bend back upon oneself' which in research means self-awareness of the dynamics between the researcher and the particpants[136]. I kept a reflexivity journal throughout the research to help me think carefully about what I was doing, why, and how it impacted my participants, the study and me. An excerpt of the reflexivity journal is in Appendix E.

### 3.9 Ethics

This study, like all research involving human beings, had to ensure the safety of my participants and me. Ethical approval was obtained from the Joint-Faculty Research Ethics Board at the University of Manitoba (Appendix F). The research required the disclosure of a hereditary genetic condition and had the potential to resurface painful emotions regarding the loss of participant's family members whose deaths were attributed to LS cancers. Whenever this occurred, I provided emotional support and gave the participant time and space to express their feelings. I reiterated to the participant their right to terminate their participation at any time and provided options for cost-free support(e.g., professional counselling services), as listed on the consent form.

#### 3.9.1 Informed Consent

Informed consent mandates that research participants have the right to be informed about the nature and consequences of the research they agree to participate in[137]. With respect to this study, I adhered to the informed consent criteria by informing participants about the content of the research and their participation through an information sheet and informed consent letter.

Each of the informed consent letters (Appendix G) indicated that participation was entirely voluntary and that participants could withdraw their participation at any time. The data were collected from audio-recordings of interviews and the letters of consent included a statement indicating this data collection method. The participants were assured in the letters that their audio-recording would only be used to transcribe their conversation and only I would have access to the audio-recording, as it revealed their identity. During transcription all identifiers were removed from the transcript, so committee members could not access participants' personal information.

### 3.9.2 Confidentiality and Ethical Treatment of Data

I ensured the anonymity of the participants and the privacy of their data. Immediately following the interview every participant received a number to identify their audio-recording and descriptive statistics survey. The interview data were kept confidential by using numbers and the data were kept in a secure area during the research, which they will remain for three years. The audio-recorded interviews and transcriptions are on my password-protected computer hard drive.

### 3.9.3 Member Checking

To help ensure that the results were faithful to the voices of participants, preliminary results were emailed to all the participants. All participants responded by confirming their written agreement with the preliminary themes and 15 of 17 participants indicated what they would like me further to emphasize or add. The final dissemination of the data will include a public lecture and a full summary will be emailed to research participants.

# CHAPTER 4: FINDINGS

Both the quantitative and qualitative findings are included in this chapter. Although the qualitative part of the study is emphasized, the secondary, quantitative strand of the study is described and presented first in the findings. Although not generalizable to the larger population, the quantitative description will situate the context of the qualitative findings.

## 4.1 Quantitative Findings: Participants

Seventeen participants completed the study between October 2017 and April 2018. All interviews and surveys were conducted in person. The age of participants varied from 25 to 90 years, the mean age being 51.5 years (Table 3). Most participants were female (Table 3.) and either married or living with a common law partner. Almost 65 percent of the participants held a university degree (Table 3.).

Table 3. Participant demographic characteristics (*N*=17)

| **Characteristic** | **% (n)** |
|---|---|
| Mean age (SD, range), years | 51.5 (18.3, 25-90) |
| Females | 64.7 (11) |
| Males | 35.3 (6) |
| Married or common law | 76.5 (13) |
| Highest education, grade 12 | 11.8 (2) |
| Highest education, college diploma | 23.5 (4) |
| Highest education, Bachelors degree | 47.1 (8) |
| Highest education, Masters degree | 17.6 (3) |

MLH1 was the most common genotype affected by LS. Eight participants had this genotype, two carrying the MLH1 affected variant with Muir-Torre syndrome (Table 4). One participant had the EPCAM genotype affected as revealed by genetic testing privately funded by the participant's family at the Mayo Clinic in Rochester, Minnesota because Manitoba does not have the infrastructure to test for that variant.

Sixty-six percent of the male participants had been diagnosed with colorectal cancer whereas 18 percent of female participants had a previous colorectal cancer diagnosis. Forty-five percent of female participants had received an endometrial cancer diagnosis. One participant discovered she had endometrial cancer after her prophylactic hysterectomy and bilateral oophorectomy. Six of 11 female participants had not undergone prophylactic surgery – Hysterectomy and bilateral oophorectomy: four participants already had an endometrial cancer diagnosis prior to surgery, and the remaining two females were considering prophylactic surgery; their concerns regarding the surgery are described in the qualitative strand of this study.

Table 4. Participant medical characteristics (*N*=17)

| **Characteristic** | **% (n)** |
|---|---|
| Genotype | |
| MLH1 | 35.3 (6) |
| MLH1-Muir Torre syndrome | 11.8 (2) |
| MSH6 | 17.7 (3) |
| MSH2 | 29.4 (5) |
| EPCAM | 5.9 (1) |
| Prior bladder cancer diagnosis | 5.9 (1) |
| Prior colorectal cancer diagnosis | 35.3 (6) |
| Prior endometrial cancer diagnosis (% females) | 45.5 (5) |
| Prophylactic surgery-hysterectomy bilateral oophorectomy (% females) | 45.5 (5) |

Twelve different families, where family was defined as having a different and unrelated proband, were involved in the study. Sixty-six percent of probands in the 12 different families were tested outside of Manitoba (Table 5). This result aligns with the history of genetic testing for LS in Manitoba, as no proband testing on CRC tumours was permitted in Manitoba until late 2013. As noted previously (Section 1.4), Manitoba was the last province to initiate proband testing[50]. The qualitative strand highlights the barriers and personal financial burden that Manitoba Health's decision to exclude LS proband testing had on families.

Table 5. Participant characteristics regarding proband status and location of genetic testing

| **Proband and participant testing characteristics** | **% (n)** |
|---|---|

| | |
|---|---|
| Participants who are the proband | 23.5 (4) |
| Proband participants tested in Manitoba | 50.0 (2) |
| Testing location of probands of the 12 different families involved in the study | |
| Manitoba | 33.3 (4) |
| Mayo Clinic, Rochester, Minnesota | 16.7 (2) |
| Myriad Genetics, Salt Lake City, Utah | 16.7 (2) |
| Zane Cohen Centre, Toronto, Ontario | 16.7 (2) |
| British Columbia | 8.3 (1) |
| Quebec | 8.3 (1) |

Almost 65 percent of participants underwent annual colonoscopies (Table 6) as recommended by the National Comprehensive Cancer Network®[138]. Some participants had clinicians recommend this time period and other participants advocated for this time period. Barriers and facilitators regarding colonoscopy spacing are described in the qualitative strand. Thirty-three percent of participants had colonoscopies bi-annually and one participant had colonoscopies spaced every 18 months (Table 6). Most gastroscopies were performed on an annual bias. Three participants had never had a gastroscopy performed. It is recommended that the gastroscopy spacing be at least 3-5 years depending on the family history of gastric cancer[138]. The 90 year-old participant had never had organ surveillance discussed or recommended and thus had never had it performed. Urine cytology is recommended for all persons with LS and only two participants had it completed. Just over a quarter of the female participants had undergone endometrial sampling and under 12 percent had a pelvic ultrasound (Table 6). The current guidelines for female organ surveillance highlight that endometrial cancer screening does not have proven benefit for women with LS, as even without surveillance endometrial cancer is often caught early[138]. An annual CA 125 blood test is recommended for persons previously diagnosed with cancer[138]. The participant who undergoes annual computed tomography had been diagnosed with multiple different cancers and it was shared in the qualitative strand of the study

that the gastroenterologist had enacted a personalized approach to reduce the risk of future cancers.

Table 6. Participant organ surveillance characteristics.

| **Organ surveillance** | **% (n)** |
|---|---|
| Annual colonoscopy | 64.7 (11) |
| Annual gastroscopy | 47.1 (8) |
| 1.5 year spacing colonoscopy | 5.9 (1) |
| 2 year spacing colonoscopy | 33.3 (4) |
| 2 year spacing gastroscopy | 33.3 (4) |
| Sporadic gastroscopy | 5.9 (1) |
| Endometrial sampling either currently or before prophylactic surgery (n of females) | 27.3 (3) |
| Pelvic ultrasound (n of females) | 27.3 (3) |
| Urine cytology | 11.8 (2) |
| CA 125 blood test | 11.8 (2) |
| Annual computed tomography scan | 5.9 (1) |

All participants in the study indicated that they had shared or would share their LS diagnosis with their children and discuss with them the risks and benefits of genetic testing (Table 7). Two participants had children who died from a LS cancer identified at a late stage and before LS testing was implemented in Manitoba. Forty-one percent of participants had a parent die of a LS cancer (Table 7). Thirty-three percent of participants had a sibling die of a LS cancer (Table 7.). Forty-seven percent of participants had a grandparent die of a LS cancer (Table 7.).

Table 7. Participant characteristics regarding familial communication and family history

| **Communication and family history** | **% (n)** |
|---|---|
| Parents or potential parents that would or have share(ed) their diagnosis with their children and discuss(ed) the risks and benefits of genetic testing | 100 (17) |
| Number of participants who had a child die from a LS cancer | 11.8 (2) |
| Number of participants who had a parent die from a LS cancer | 41.2 (7) |

| | |
|---|---|
| Number of participants who had a sibling die from a LS cancer | 33.3 (4) |
| Number of participants who had a grandparent die from a LS cancer | 47.1 (8) |

A diagnosis of LS can cause a person to change their lifestyle and take daily aspirin as a form of chemoprevention. Fifty-three percent of participants took daily aspirin for this reason (Table 8). Forty-seven percent of participants increased or began an exercise regimen following diagnosis and 47 percent also increased their fruit and vegetable intake (Table 8). Forty-one percent of participants lowered their red meat consumption (Table 8). Once a person makes lifestyle changes to try prevent the development of cancer it demonstrates the impact a diagnosis can have on someone's life.

Table 8. Lifestyle changes and chemoprevention following diagnosis

| **Lifestyle change or chemoprevention** | **% (n)** |
|---|---|
| Lowered red meat intake | 41.2% (7) |
| Increased fruit and vegetable intake | 47.1% (8) |
| Lowered sugar intake | 29.4% (5) |
| Began or increased exercise regimen | 47.1% (8) |
| Began taking daily aspirin | 53.0% (9) |

## 4.2 Qualitative Findings

Although data collection consisted of a private interview with each participant and each person's story is fascinating in its own right, the collective account is the central focus of this research. The voice of the individual is the medium of communicating the larger account. Most participants described a combination of themes and all participants experienced barriers in receiving care. Themes (It's health care, not 'Russian Roulette'; Knowledge; Central force in the family; Personalized approach; Ensuring continuity of care; Need for care coordination) are

presented in the order in which persons with LS described their trajectory within the health care system, beginning with genetic testing and diagnosis. Figure 1 seeks to elucidate how the themes were organized.

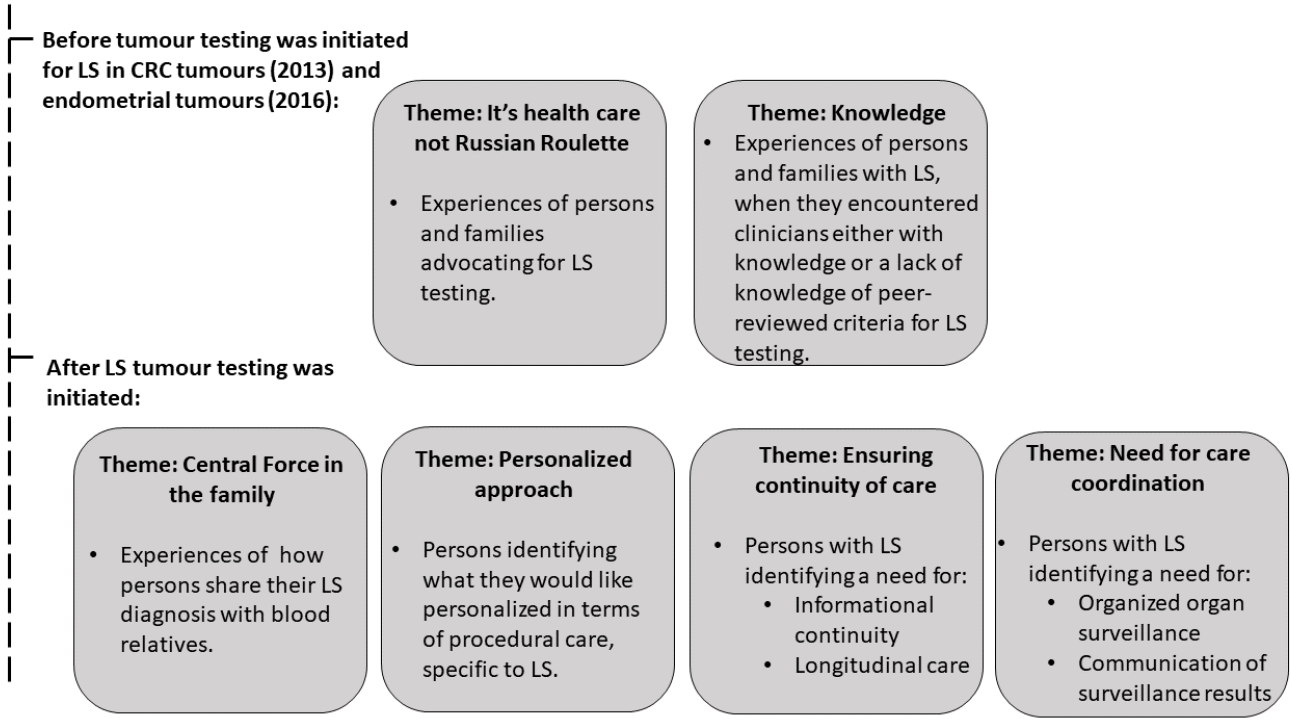


Figure 1. Chart of themes and their corresponding description.

The research required the disclosure of a hereditary genetic condition and the Research Ethics Board expressed concern that readers might identify at least some of the participants based upon the information shared. I do not want inadvertently to compromise any participant's anonymity and thus was unable to link basic information about each participant to their quotes.

### 4.2.1Theme: *It's health care, not 'Russian Roulette'*

The first theme, *It's health care, not 'Russian Roulette'* is historical in nature and although not relevant in Manitoba today, five families in my study were denied genetic testing (unless they went to the Press[139]) until Manitoba approved LS tumour testing in colorectal

tumours in late 2013[48]. If the mutation presented itself in a family as endometrial cancer, which is common with the MSH6 genotype, tumour testing was denied until 2017. It is likely that other jurisdictions globally, and possibly the Territories in Canada, do not provide LS genetic testing, and this theme represents the experience of those persons or families that were either denied or denied at some point.

The term, 'Russian Roulette' for this theme comes directly from the quote below where the participant describes their advocacy for LS testing in light of being denied.

Interviewer: So how did you advocate to have genetic testing when it was denied at first?

Participant 7: So we wrote letters that were fairly – they were poignant. I definitely called out the fact that I felt that they [health care system] were playing 'Russian roulette' with my life and that I could have a better quality of life if I did know that this [LS] was something I had, and that I did not want to repeat what my father's experiences had been.

> So yeah, some pretty strongly-worded letters were sent. It took a few months to get results, a response from them. The response that was sent was from the province [Government of Manitoba] to Cancer Care asking them to fund my testing.

This participant's father died at a young age from colorectal cancer and once the participant had their own colorectal cancer diagnosis as a young adult, they were dedicated to finding out whether they had LS.

Participant 7 used the term "playing 'Russian Roulette' with my life" when explaining how it felt to have genetic testing denied by the health system. Some participants identified that being tested for LS was more for their family's future than for themselves. In one instance it was the mother who after her son's death from cancer discovered that she and the family had LS. The participant explained why her son wanted the genetic testing completed:

Participant 14: ____ [name of son] made sure the doctor had his tumour sample saved and took a vial of his blood so he could be tested once the test became available in the province. He wanted it more for his family than for himself. His family's health was very important to him. If they got cancer then they would be able to have informed decisions for treatment.

Following the extensive family history of cancer, another participant's surgeon took an active role in outsourcing as much testing of the patient's colorectal tumour as Manitoba would permit. The step from MSI testing to LS testing though was not completed until Manitoba permitted LS testing on probands with colorectal cancer in late 2013. The dedication of the surgeon to finding ways to support the case for Manitoba to fund LS testing demonstrates that some clinicians felt that a diagnosis was important for their patient's health as well as the health of their family . The participant states at the end of this description how LS testing was important for their family:

Participant 9: Yeah. So my grandpa had colon cancer before I ever knew him. I wasn't born yet. And then my Mom was endometrial, and my Auntie was actually brain cancer. My Auntie got it, brain cancer. She kind of got better, but then at the same time, you know, we were young, too. I hope my Mom doesn't get it. Then my Mom got cancer. She passed away. Then Auntie got it back and she passed away.

Interviewer: And based on your cancer diagnoses, there was no interested health care professional besides the surgeon who sent the sample out to British Columbia for MSI testing?

Participant 9: No, because even at that point Lynch syndrome wasn't even a - wasn't even on – wasn't even mentioned. The surgeon just did it on his own time. I guess he might do it all the time. He gave me the results and said you probably could have Lynch syndrome. But we had to wait a long-time for the people at Genetics to approve Lynch Syndrome testing.

The idea of knowing whether you have LS is important for families as the participant states, "for anybody that has a kid, he'll, it seems like he'll probably want to know". This demonstrates that LS testing is not a test that affects one person in the health system but rather has impact on all blood relatives.

The theme of *It's health care, not 'Russian Roulette'* also relates to the concept of worry and fear when LS genetic testing is denied. The unknown fear may be stronger for women as they have additional organs to be concerned about. Women with LS contemplate whether it would be

best to have their uterus, ovaries and fallopian tubes removed prophylactically. The quote below demonstrates this concern in female family members diagnosed with endometrial cancer:

Participant 3: And that, I guess, came about because first of all my sister in 2007 was diagnosed with endometrial cancer. She had the same kind that I would have laterally, it's clear cell carcinoma, and it's a very aggressive kind of cancer. And they thought that was rather strange as my Aunt had that.

But she [Mother] was over 70 also got endometrial cancer so they wouldn't do any Lynch syndrome testing on her because of her age which we didn't really agree with because then we were starting to get worried that how come, like, this is the third person in our family that has it and then my sister was talking to Cancer Care Manitoba saying could this be Lynch syndrome? And then she went to the genetics department here and they're like no way like that'd be Lynch syndrome. There's no way. You don't have a history of colon cancer so doesn't make sense.

So anyway, so then my younger sister started to get really worried because she's got three little kids and she was going oh my gosh, like am I next? Like, so she was getting really stressed out.

Chemotherapy for cancer treatment has associated risks, and often impacts negatively on a person's immediate and future quality of life[140]. It has been known since 2007, that current chemotherapy options for colorectal cancer caused by LS do not provide benefit[32]. When Participant 7 was diagnosed with colorectal cancer in 2012, her experience with her oncologist was as follows:

Participant 7: So following the surgery for my cancer, he did not like what he saw. He did find that there were lymph nodes affected and proceeded with a subtotal colectomy, so removing 80% of my colon. I recovered for six weeks, and right at the end of my six-week recovery I began my first chemotherapy treatments on FOLFOX-4. I did the full – I was in treatment every second week until March. I think it was 12 treatments; my math is off and I'm glad to have done that.

When I asked Participant 7 about what they would like to see change in their initially rejected and later delayed process to genetic testing the first comment made was in regard to the

chemotherapy provided and how at the time they were unable to evaluate whether it was the best option because they did not know if they had LS:

Interviewer: So would you change anything in the process to receive genetic testing?

Participant 7: Yeah, quite frankly just based on the few little hits that I had, the things I had identified with my surgeon, if he was able to pick up on the potential link, I can't help but think that I should've been sent for genetic testing prior to even beginning my chemotherapy treatment, especially knowing that a little bit more now about Lynch syndrome. I might have been more hesitant to undertake chemotherapy with what I know now, and I just don't feel like I had a full knowledge set to go into it with.

And I kind of feel like the medical profession played off of my fear a little bit, that I was concerned obviously about a recurrence and making sure that I didn't still have cancer and there wasn't any evidence of it. But I also didn't have all the information to be basing that on. I was pretty unhappy about chemotherapy.

Russian roulette is often viewed as a lethal game of chance. The probability is related to the number of rounds in a revolver. The number of rounds is usually known so a probability can be estimated. This is not the case when LS testing is denied, as persons and family members do not know the likelihood of receiving a diagnosis. Thus, being denied LS genetic testing and necessary health care in general often also warrants the investigation of knowledge.

### 4.2.2 Theme: *Knowledge*

This theme centers around participants' narratives where knowledge and lack of knowledge of the health system were shared. According to various dictionaries, knowledge in the form of a noun is "familiarity with facts, truths, and principles from study"[141]. In contrast ignorance is defined as the "want or lack of knowledge"[141]. These definitions treat knowledge and ignorance as polarities: as exclusive opposites. In our everyday experience, "knowing" and

"not knowing" intermingle, for it takes knowledge to acknowledge ignorance. As persons our abilities to understand are limited, since we cannot know what we cannot know.

The Canadian Medical Association Code of Ethics[142] states, under *Virtues of an Ethical Physician*:

> "A humble physician acknowledges and is cautious not to overstep the limits of their knowledge and skills or the limits of medicine, seeks advice and support from colleagues in challenging circumstances, and recognizes the patient's knowledge of their own circumstances.
>
> A prudent physician uses clinical and moral reasoning and judgement, considers all relevant knowledge and circumstances, and makes decisions carefully, in good conscience, and with due regard for principles of exemplary medical care" (p.1).

This Code of Ethics joins the statement, "cautious not to overstep the limits of their knowledge" with "considers all relevant knowledge", I interpret this as meaning a physician does their best to seek knowledge.

Many of the participants who had LS genetic testing initially denied by clinicians had a misunderstanding of why testing was denied. In addition, some participants experienced outright denial of the possibility of LS, however at the time they met either the Amsterdam II criteria or Bethesda guidelines. The theme, knowledge describes when clinicians appeared to lack knowledge about LS diagnosis criteria and also the instances where clinicians were knowledgeable about LS diagnosis criteria.

This portion of the theme *knowledge* describes the participant's experiences of being told the true nature about the investigation regarding the possibility of LS in Manitoba and elsewhere. The first set of quotes demonstrates how the action of surgeons sharing knowledge about the possibility of LS empowered participants to advocate for and/or undergo LS testing.

Participant 7's surgeon suggested the possibility of LS even though it was known at the time that Manitoba did not fund proband LS testing. The surgeon also proposed to complete a subtotal colectomy, a standard procedure for persons with LS. It was performed without a diagnosis of LS because the surgeon felt that the strong family history and cancer type warranted precautions. The surgeon was transparent with Participant 7 about his concern. If it were not for the surgeon's truth telling regarding a possibility of LS the participant would never have heard of LS:

Participant 7: My first exposure to Lynch syndrome was through the surgeon that would eventually do my surgery. Because it was a cancer diagnosis I was speaking with him about the connection with my dad and his cancer, and I guess that was enough to clue him in that there might be more to it than just a rare advanced-stage cancer. And so he said the words Lynch syndrome to me and was also the one to propose a subtotal colectomy, if when he got in there the tumour looked like it may be Lynch-related and hereditary.

The surgeon in care of Participant 14's son also shared their knowledge regarding the possibility of LS in 2010 even though Manitoba was not offering LS proband testing at the time:

Interviewer: So when did there term 'Lynch Syndrome' start being used?

Participant 14: Ummm immediately. The surgeon told his wife, my-daughter-in-law ___, and him that he was 90 or 99% sure it was Lynch Syndrome based on the family history. That is when we got referred to Medical Genetics by the surgeon. It took a long time to get in to see them.

The experiences of these two participants demonstrate that even when a health system may not offer a specific test, the knowledge of a physician regarding the possibility of a condition does not need to be withheld from their patient. Beauchamp and Childress (2001) propose using four principles when making ethical decisions in health care: respect for autonomy, beneficence, non-maleficence and justice[143] as explored in the discussion chapter.

One participant received treatment for colon cancer in Thunder Bay, Ontario in the late ’90s and experienced the following in a health system that permitted LS proband testing:

Interviewer: So the first question is general: how did you first hear about Lynch Syndrome?

Participant 15: After I had the colon cancer because I had the colon cancer when I was 22. Ahh because of my age the Doctors, well they figured there might be a connection with Lynch Syndrome. I am not sure if it was the cancer centre in Thunder Bay or if it was my Mother that contacted the Zane Cohen Centre in Toronto and they carried through with the genetic testing for the family and stuff and so.

Interviewer: So was it your surgeon that connected the dots?

Participant 15: Ahh yeah. It would have been because my Grandfather died of colon cancer in 40s no the 1950s. Ahh then what ended up happening is because I did have it so young that there must be something. So then of course they connected back with him because they said ‘oh your Grandpa he died of that’.

Surgeons were not the only clinicians to notice the possibility of a hereditary condition. In one instance a participant’s family lived in rural Manitoba where the practicing physician noticed the pattern of cancer diagnoses among family members:

Participant 6: So one of them had - I believe she had endometrial cancer as well, so she had a hysterectomy and because all the doctors out there, they go to the same doctor realized hey, this is the same family, we should check into this, so that's when the genetic testing started. And then of course, because my mom's had cancer twice, so figured, I wanted to get checked.

Another participant shared that during the time period when Manitoba did not permit LS testing their gastroenterologist began performing annual colonoscopies due to the participant’s family history:

Participant 12: So, my GI doc said we don't have evidence of Lynch because we can't get you tested, but because your dad had colon cancer and we found these polyps, we're going to keep surveilling you every year anyway.

Interviewer: Oh, okay. So, he or she made that executive decision?

Participant 12: Right.

In response to Manitoba's denial of LS proband testing one participant reached out to Dr. H.T. Lynch in Nebraska who discovered LS, in order to receive medical information:

Interviewer: Did you do outside research before about Lynch syndrome?

Participant 4: I did. Once my friend had told me about Lynch syndrome I did. I did a lot of research about it. I wrote to Dr. Lynch who responded right away and told me all the steps that we needed to take and what we should be doing and the testing that we should have.

The above quote demonstrates that this participant felt the need to go outside the Manitoba health system to obtain accurate and trustworthy information.

**Lack of knowledge**

Within the scope of knowledge is lack of knowledge. The idea of lack of knowledge begins with exploring the interaction that participants had with the Manitoba health system. The first experience described is that of a physician who failed to communicate important information to a patient that was not immediately related to treatment decisions but would have had a potential impact on family members.

Participant 1: So my two sisters were fighting cancer; my younger sister at the time was 44 and my older sister at the time fighting was 50. And a friend of the family spoke to us and asked if we ever tested for Lynch because she tested for Lynch out in California. We had never heard of it before; we started doing some research and

> then my 50 year old sister who was fighting endometrial cancer, really advocated with Cancer Care Manitoba to have this test done. So she had a meeting with her doctor and they asked her many, many, many questions and she answered all of them as if she should have the Lynch testing and then the last question was are you Mennonite background and she answered no because she's Jewish, and there's a very strong correlation to Mennonite backgrounds and I think they're finding now a correlation to Ashkenazi Jewish backgrounds. So she answered no and right at that spot they said sorry you cannot have this test done.
>
> So being the sister she was - very fiery and a strong advocate for herself - she didn't want no for an answer, she was convinced that she had Lynch. So after a big, big fuss with Cancer Care, they agreed to send her to Mayo Clinic.

This quote demonstrates the impact of a physician's lack of knowledge The physician used the criterion of a Mennonite background, as some Mennonite families in Manitoba had probands tested elsewhere for the MLH1 mutation[144]. Thus, the criterion was not truly based on ethnicity but rather on a family's proband living in a province that at the time funded LS testing. The participant's sister did not only ask that LS testing be performed outside Manitoba, as she additionally asked that Manitoba Health fund an appointment with a specialist at the Mayo Clinic in Rochester, Minnesota. This request likely demonstrates that the physician's lack of knowledge with the participant's sister caused the sister to lose some trust in physicians in Manitoba, as she requested that the genetic testing be investigated outside Manitoba. The sister of the participant died a few months after receiving her LS diagnosis at the Mayo Clinic, yet her advocacy of LS testing as the proband in the family, permitted her blood relatives to receive LS testing in Manitoba.

The terms 'truth' and 'trust' are embedded when one shares knowledge and they are concepts often associated with the health care professions. For physicians, embedded within these terms are the conditions of respect for the autonomy of persons, refraining from harming persons, but also contributing to a person's welfare[143]. Some persons in Manitoba were made aware of LS through a family member. When this finding was brought to the attention of the physicians providing care, the possibility of LS was denied:

Interviewer: Okay. So first to start out with a general question. How did you first hear about Lynch syndrome?

Participant 3: I had heard about it vaguely through my sister who's a ____ [health professional] and she was wondering if our family had Lynch syndrome because we had a family history of endometrial cancer. My aunt had it and then my sister had it and then my mom had it and then she started talking about it and then we had asked more questions about it and the Cancer Care that was dealing with my mom at the time said no, we don't think that it's Lynch syndrome and they didn't want to really do any testing.

Participant 3: And what year would that have been for your family?

Respondent: My mom … Let's see. Mine was in 2015 and my mom's was maybe a year prior, like maybe 2014, 2013. I can't remember the exact dates.

The participant goes on to explain later in the interview that the explanation given by the Metabolism and Genetics department regarding their impossibility of having LS was the lack of colorectal cancer in the family:

Participant 3: And then she [Mother] went to the genetics department here and they're like no way like that'd be Lynch syndrome. There's no way. You don't have a history of colon cancer so doesn't make sense.

A database search of MEDLINE (Ovid) revealed that 2001 was the first year in which endometrial cancer as a standalone cancer was published in the medical literature as an inherited form of malignancy derived from HNPCC (what LS was previously called) mismatch repair gene mutation[145]. Yet, the physician whom the participant encountered in the Metabolism and Genetics department in around 2013 was unaware of this over 10-year old information. The statement by the genetics physician, declaring that the family did not have LS, led the participant's gynecologist to strongly suggest that a prophylactic hysterectomy with oophorectomy was unnecessary. The suggestion proved detrimental as the participant was diagnosed with endometrial cancer less than a year after the conversation with the gynecologist. Below, the participant describes the sequence

of events and how the strong statement by the genetics physician caused her to doubt her symptoms:

Participant 3: Yeah. She [gynecologist] said I don't think that you need it but you know, it's up to you of course and so she kind of sort of talked me out of it and then I started having some bleeding. So of course, I got worried, but still we didn't have any evidence of Lynch syndrome so I was thinking okay, maybe I'm just being like – because meanwhile my mom's going through chemo and all this, right? So I'm going maybe I'm just being hypersensitive to all this. Maybe I'm just paranoid.

Interviewer: And you were told that you did not have Lynch Syndrome so it makes it more difficult?

Participant 3: Yeah. Like they make you think you're crazy, right? So anyways, then I went back to her [gynecologist] and she said no, like you just had an endometrial biopsy like six months ago. There's no way that you have cancer. Like you know, maybe it's something else. So then I waited and she sent me for an ultrasound and it came back and it was nothing of no trait. So she said well, you know, you're fine but just to be on the safe side why don't we do a D &C [Dilation and Curettage]. So that was like good. So I went and did the D&C and then it came back that I had this endometrial adenocarcinoma…

Even after the participant's diagnosis of endometrial cancer, which made her the fourth person in her family to be diagnosed with endometrial cancer, the genetics physician did not think that LS was a possibility. The participant decided to take ownership of her family's situation and privately paid for LS testing to be completed in the US:

Participant 3: So what we ended up doing is we decided to contact that Myriad Genetics and pay privately for the testing. It cost us $4,000 because my sister didn't want to go and have a surgery [prophylactic hysterectomy oophorectomy] that she might not need if she didn't have it.

So we got the Myriad Genetics test kit and I got – I had to go first because I was – and my [other] sister had passed away, passed away shortly after. So she was never able to be involved in that test, but I did and I got my results back in two weeks and then my sister was able to get her results back and then she was actually negative.

How the participant interpreted her gynecologist's behaviour before and then after the LS testing was completed is described below:

Participant 3: And like I said, as much as I really like my gynecologist she's like well … She kind of thought we were like lunatics, right, going hey, I need to have this hysterectomy and she's like well why? Like you don't know that you've got anything. But anyway, she's … It was good for her, I think, because she learned a lot too and yeah. And she was kind of interacting with the Myriad Genetics too so I think she learned quite a bit too.

Interviewer: Okay. So she'd be like a bit of an expert here in Winnipeg?

Participant 3: Yeah. She's learned quite a bit, I think, too. So I think I was her first Lynch syndrome patient that she probably knew of. I'm sure there's been more, but nobody would even know, right? So because I know people are on the colon side, colon cancer.

I then asked the participant if after receiving the LS diagnosis from Myriad Genetics she or family members ever went back to the department of Genetics and Metabolism:

Interviewer: And when you were meeting with genetics with your ___[genotype affected] positive test were you meeting –

Participant 3: - Oh, you know what? I never did. I never went and met with them here [Manitoba]. You know what we did is because like I said, I work for an amazing company, my mom and I went down to the Mayo Clinic and –

Interviewer: - Okay. In Rochester.

Participant 3: Yeah, that's right. And we met with a genetics counsellor down there at Mayo and he went all through everything and gave us what the recommended surveillance is and I brought that back and gave it to Dr. __ [Endoscopist] and that's kind of what I'm going with.

The participant and her family felt that the best decision after receiving the diagnosis from Myriad Genetics was not to go back to the Metabolism and Genetics department in Manitoba but instead to attend the Mayo Clinic for genetic counselling. The participant described the experience at the Mayo Clinic by incorporating her thoughts of the Metabolism and Genetics department in Manitoba:

Interviewer: And with the genetic testing at the Mayo Clinic how did you feel that it went like?

Participant 3: - The guy was super nice. He charted out everything and he drew me up a little diagram and just sort of went through what the best practices were because – and

> I'm not sure that it was probably anything more than I [would have] got here [Manitoba], but it was just we were kind of just like done with it here [Manitoba], you know what I mean, after all that. So and kind of like I don't want to go meet with them [Metabolism and Genetics department].

The choice of words, "…we were kind of just like done with it here [Manitoba]…" which was followed with "…don't want to go meet with them [Metabolism and Genetics department]" demonstrates a less than satisfactory physician-patient relationship that likely resulted from the lack of knowledge regarding the possibility of a LS diagnosis.

Below the participant describes their experience at Mayo Clinic in terms of, "it was worth it because at least you feel like you're getting modern information". This statement implies that the participant realized that they received outdated information from the Metabolism and Genetics department in Manitoba regarding endometrial cancer not being solely attributed to LS:

Interviewer: No, I understand. So did you feel you had a good experience or not so much at the Mayo Clinic?

Respondent: I felt like I had that at the Mayo Clinic because they know a lot about it and they were like – I think it cost like my – like I said, this place that I work here is amazing. So they paid for it. They said hey, you know, why don't you go down there if it'll make you feel better? So that's just the kind of company this is. So we went down there and I think it cost $2,000 but yeah, it was worth it because at least you feel like you're getting like modern information and you feel like you could come back and go hey, I've got this and …

It is known that before LS testing was implemented in Manitoba the main option was to go to the Press to have LS testing paid for by Manitoba Health (Appendix A). This method demonstrates that the public's confidence in the health system must have been undermined. One participant shared that going to the media was not an avenue with which their family was comfortable moving forward:

Interviewer: That is difficult in light of having to also keep fighting for testing. So for the Lynch Syndrome testing how did that end up going?

Participant 14: People were going to the press and having their names published. They got the testing paid for. But I was not comfortable going to the press and exposing this hereditary condition. It can ruin people's insurance and relationships. My daughter-in-law was also not comfortable having the last name associated with Lynch Syndrome. So we did not go the press, but every time we heard a story we phoned [the] Medical Genetics [department].

It took several letters and discussions before Manitoba implemented LS testing for this family.

The participant described the health system in this way in the closing remarks of the interview:

Interviewer: Is there anything else you would like to share?

Participant 14: Our family had to advocate for testing for years. No family should be treated like this. We need compassion and things implemented to prevent cancer and it does not seem like that is happening here in the health system. No one seems to care and if they do there is nothing they can do about it.

In closing, the Canadian Medical Association Code of Ethics[146] recommends that physicians provide patients with whatever information and thus knowledge that will, from the patient's perspective, have a bearing on medical care decision-making and communicate that information clearly to the patient.

### 4.2.3 Theme: *Central force in the family*

When participants were asked about how a LS diagnosis was communicated to other blood relatives it became apparent that someone in the family became the central force in communicating the possibility of a LS diagnosis to blood relatives. This theme will first present instances where the participant was the central force in the family for communicating their possibility of a LS diagnosis. Described secondly are the participants who experienced the proband in the family being the central force in communicating the LS diagnosis. Thirdly, one participant experienced a non-LS positive family member who at the time took on the role of the communicator regarding the LS diagnosis and became the central force in the family.

Participant 7, who was additionally the proband in the family, described how sharing the LS diagnosis with blood relatives occurred:

Interviewer: So you did contact relatives that weren't very close to you but related by blood, like your aunt and uncle?

Participant 7: You know what? I did actually, so there were a few that – the primary ones I contacted were my dad's siblings. Them I would consider myself close with, but through them I was put in contact with some of the further-out cousins. I also wanted to see if I could get more information on our family history and which cancers we might be able to expect.

So through Facebook I contacted a few more, was able to learn a little bit more but ultimately I don't know that a lot came of it because they weren't from the known line of – like we know that my dad had it so we know his mom had it and his siblings are affected, but we don't know that her – we assume her brother had it and that his family would be affected, so those were the ones I contacted on Facebook and let them know.

Most of them live in the US I never did hear back as to whether any of them proceeded or got testing. I don't see anything on Facebook about cancers or illness so I'd like to think that they dodged that bullet.

The quotes above demonstrate the lengths to which Participant 7 went to communicate the possibility of LS to blood relatives. Participant 9 explained that their family was small and as the diagnosed proband they shared the results with the sibling and first cousin:

Interviewer: And after you received your genetic test result, did you communicate that to family members?

Participant 9: Yeah. So once I got the results, I - you know, because then, once I have, anybody can get the test done, right? So obviously, I let ___[brother] know and then we have, we have a fairly small family. I have my brother and two cousins. That's it. So because of that, both of my cousins actually went and got tested, which I wasn't sure they – wasn't sure if they were going to, but they did.

Another participant shared how they communicated the LS diagnosis to their siblings and immediate cousins:

Interviewer: And I have a question, just for communicating the genetic status to the extended family members, did you do that?

Participant 5: Oh yeah. We did. Like my brother lives in BC [British Columbia] and he has three daughters. So as soon as we had our stuff from the genetics and got the diagnosis - actually even before that we told him what we were going to have that done. And once we did it, and I was said to have Lynch syndrome, we wrote, told him, email there. And said that, you know, you might want to look into that yourself and for the girls and that.

Participant 13 is the father of the family's proband. The proband was tested in Quebec and was unable to receive formal documentation in English to send to relatives. Once the father had his LS testing completed in Ontario, where English documentation was given, he said that:

Participant 13: Now having learned that I had Lynch syndrome, I contacted my surviving brother and his two sons, and the daughter's of my deceased brother, and explained everything and said 'Well, it's hereditary' etc, etc. And the odd thing is we only got one request for release of information, just the one nephew. _____ requested a release of information from the UK.

In another family, the central force in the family was the proband tested, who later rapidly died from a LS cancer. This participant shares how the proband wrote a letter to the siblings and called a family meeting to share the LS diagnosis:

Participant 1: So anyways what happened was she [proband] wrote a letter to my older sister and I and in that letter were many things but one of the things she said was you have to do me this one favour, this one thing - you have to get tested for Lynch because I have it and there's a 50/50 chance you have it. So we started doing research – we didn't know anything about it – and started doing more research. She called a family meeting actually where we all sat around and learned about it…

Participant 16 had a brother living in Ontario who was diagnosed with colorectal cancer in the mid-'90s when Ontario received funding from the US National Institutes of Health to fund the Colon Cancer Family Registries[144]. Participant 16 described what occurred approximately 20 years ago:

Interviewer: And after your brother got tested how did his communication to the rest of the family, for example to you, how did that happen?

Participant 16: Well, this would be something that my brother would send me like after he found out and that's where it all started I guess so yeah he did forward that information to the rest of us.

Interviewer: Okay, so it's the letter from genetic counselling and the cancer registry [participant had the letter from 1998 on the table] that started everything.

Participant 16: I think so, yeah.

The experience of participant 16 demonstrates the role that a cancer registry or genetic counselor can play in preparing a letter to share with family members regarding the LS diagnosis of a blood relative. A letter or a package prepared by a genetic counsellor also had an impact on another participant:

Participant 11: I get a phone call first from my mother and she says your cousin has been tested for something, and it's come back positive, and it appears as though we've got some kind of genetic thing in our family. Back then they were referring to it as HNPCC.

So … and she said I've been tested, and she was living in Victoria [British Columbia] at the time, I've been tested, and my genetic counsellor wants to send you a package of information so that you can get tested as well.

In this instance, the central force in the family seemed to have been passed down from the proband to the closest blood relatives and if that closest blood relative tested positive it was their responsibility to share this information with their children and become the central force in communicating for that particular branch of the family tree. This situation demonstrates that several central forces may be needed within a family to communicate the LS diagnosis.

One participant was 22 years of age when they received their colorectal cancer diagnosis and LS diagnosis. Their mother became the central force in communicating the LS diagnosis with family members:

Participant 15: Once I got the result they recommended you know to tell everyone in the family to get tested. So my Mom told everyone. She was sorta the catalyst I guess. She kinda took control of that.

### 4.2.4 Theme: *Personalized approach*

A recurring theme across many interviews is that persons with LS are unique with needs that differ from the general population. Participant 2 stated, "You're not an average case". This statement applies to all aspects of medical care for persons with LS, particularly: bowel preparation for colonoscopy, chemotherapy, gynecological cancer surveillance, and hormone therapy or treatment options following prophylactic oophorectomy. These aspects fall within the confines of the theme *personalized approach*.

#### Choice in bowel preparation

Bowel preparation for colonoscopy ensures there is adequate colonic cleansing to permit the endoscopist to examine the colonic mucosa for precancerous polyps. The most commonly used bowel preparation laxative regimens have included polyethylene glycol (PEG) in high-volume (≥3L), PEG in low-volume (< 3L), sodium phosphate, oral sulfate solution, or sodium picosulfate magnesium oxide and citric acid. The Winnipeg Regional Health Authority prescribes the high-volume PEG solution[147]. Persons rate the high-volume PEG as difficult to consume[148] and a meta-analysis by Zawaly *et al.* demonstrates that high-volume PEG does not increase polyp detection.[149]. Choice in bowel preparation as part of the theme *personalized approach* describes the experiences of participants in consuming a bowel preparation for colonoscopy and the recommendations they share.

One participant described the difficulty they had in consuming the high-volume PEG solution they were prescribed:

Interviewer: PEG or polyethylene glycol.

Participant 4: Is there another name for it?

Interviewer: Oh, the brand name would be like GoLytely or Colyte.

Participant 4: That's it, yeah. It's horrible. When I first started [going for colosncopies] and I took that I drank it with that no name grapefruit pop from Safeway. I won't drink it again. It was horrible. Like I diluted it with that to change the taste. No, I won't do that. And it makes you nauseous and throw up and you don't end up taking all of it in the end.

The side effects of nausea and vomiting with high volume PEG are well documented[148]. Yet, endoscopists still prescribe this bowel preparation. Below another participant describes their experience with PEG and compares it to another type of bowel preparation common in the US:

Interviewer: Okay. And for your colonoscopies, what have your experiences with bowel preparation been like?

Participant 17: In the States, fabulous. I love the MiraLAX – well, my mom was an endo nurse as well. So I was like, "Hey, docs, like can I do MiraLAX and Gatorade?" And they're like, "Absolutely." And I was like, "Dr. [endoscopist's name], can I do MiraLAX and Gatorade?" [he responded] "No." "What? It's GoLytely again?" Or Peg Lightly or whatever – the big jug.

Interviewer: The four liters?

Participant 17: Yeah.

Interviewer: So he made you take that PEG- GoLytely?

Participant 17: Yeah. That [other] prep is heaven. But GoLytely is just a f**ker.

This experience demonstrates how some endoscopists are unwilling to try a new bowel preparation even when the patient requests it and the literature supports its effectiveness[150]. Participant 17 also shared that they asked the endoscopist a second time, "But no, he likes that prep. [high-volume PEG] I even asked his nurse. I was like, "Can I switch preps?" She's like, "No,

this is the one he likes best". This quote further demonstrates the barriers that persons experience in trying to advocate for a bowel preparation that cause less side effects for them.

Several other participants shared the difficulty they experienced with high-volume PEG, which fortunately they do not have to consume anymore:

Participant 16: The gallon jug. Oh, don't even make me take Gatorade because that's too reminiscent of the taste of that one [high-volume PEG], that one was not pleasant. This now the Pico-Salax or the Purg-Odan, I mean it's not pleasant drinking it but it's only five ounces at a time, right? So it's doable, yeah so I would say this is - this procedure is so much better than the GoLytely [high-volume PEG] one.

In addition another participant shared:

Participant 15: The one Doctor in Edmonton gave me the two big jugs [PEG]. Well when I had my first colonoscopy to find out if I did have cancer- I had the two giant jugs and it was horrible I do not even know if I got half of it down.

After watching family members struggle when consuming the high-volume PEG bowel preparation, Participant 9 shared their strong aversion to this bowel preparation:

Participant 9: But they sent me along with a big four litre jug of stuff and I'm not drinking four liters of crap.

Interviewer: Yes, so did you ever try to drink that four litres?

Participant 9: No, nope. I've seen my dad choke it down, I've seen___ [sister] try and choke it down, I have zero interest in it, I didn't even try it. I just went back to the original prep that I was given that I could handle and it wasn't too evasive and I stuck with that.

As persons with LS are recommended to undergo a colonoscopy either annual or bi-annually, the frequency of consuming a bowel preparation is tens of times higher than the average population. Participant 10 described how undergoing multiple bowel preparation regimens utilizing high-volume PEG, increased their side-effects:

Participant 10: I hate … so my first surgeon made me do GoLytely [high-volume PEG], and that was awful. And it was awful not just as a … you know, four litres, but I got this

> psychological thing where as the years went on it got worse and worse. So, as I did this more often my head started to … so I was … I would often get to three litres and I'm puking it back up again because my body just won't take it anymore. So, it was awful. When … so Doctor ___ 's [second surgeon] protocol is PicoSalax . And that's what he's … yeah. And I don't know if I should say this on the record, I don't want to get him into trouble. He sends out a separate prep from what the central intake does.

Participant 10 describes that their surgeon utilizes a bowel preparation that differs from the one the Winnipeg Regional Health Authority mails to those undergoing a colonoscopy. The surgeon who has the autonomy to do so, made the choice to provide a bowel preparation with fewer documented side effects.

When participants were asked whether they thought a personalized approach for bowel preparation for persons with LS would be useful, they unanimously acknowledged that they should not have to consume the high-volume PEG bowel preparation. One participant shared that their endoscopist tried to find the best bowel preparation for them:

Interviewer: And was your Endoscopist, I'll call them, who was doing the colonoscopy, very supportive of the trying other bowel preparation?

Participant 11: Yeah. Yeah. Absolutely. I mean, he recommended, right? So, the first time it didn't work very well, so let's try this different one the next time, and then another one, and we settled on one that worked and stuck with that.

Participant 16 said that people undergoing a colonoscopy should be a given an option for bowel preparation and know that a different option exists:

Participant 16: Yeah, so yeah I think people should have the option.

Interviewer: Okay and they should know about it [other bowel preparations]?

Participant 16: They should know about it. I think that's pretty pathetic if they don't [laughs]. I'm just thinking from my experience how yucky, just the thought of it [high-volume PEG] almost made me puke.

Another participant also shared this same perspective:

Participant 2: Yeah, exactly. So PICO-SALAX is what I've been on and it works fine and I don't want the GoLytely. I don't want that at all 4 liters bad tasting

Interviewer: You want to be able to choose?

Participant 2: I want to be able to choose, but I also am very educated. I also know what's available and what's not available. I know that my mom has had to take the GoLytely and that is she – it's not fair.

The quotes in this section, describe the negative experiences of participants while consuming high-volume PEG. Some participants were fortunate to receive care from endoscopists who endorsed the use of other bowel preparation formulas proven to be effective. Participants agreed that other bowel preparation formulas besides high-volume PEG should be offered because there is value in prescribing the bowel preparation formula with the least side effects and providing persons who have LS with a choice in the process. This links closely with the theme *personalized approach* found in my study, as a personalized approach for bowel preparation was shared as a recommendation by my participants.

**Care in gynecological surveillance**

It is recommended that women with LS who have not undergone a prophylactic hysterectomy and bilateral salpingo-oophorectomy have annual endometrial sampling performed[138]. However, the sensitivity of endometrial sampling is low in premenopausal women without symptoms of uterine bleeding[151]. Female participants who had not had a hysterectomy and bilateral salpingo-oophorectomy before or immediately after LS diagnosis, shared their experiences. Participant 2 described the pain associated with endometrial sampling and how it forced her to ask for a prophylactic hysterectomy and bilateral salpingo-oophorectomy:

Interviewer: Okay. So for endometrial sampling biopsy what would you recommend because you were going annually? Was it very difficult?

Participant 2: It was very painful. It is not easy. It is – they have to do three samples. Like they have to do three specimens. Oh, yeah. Just as you say that that just gives me the heebie-jeebies. So the first time I had I had – was before children so my cervix wasn't dilated at any point. So that one was very painful. So it would be nice if they could, you know, give you something to relax at least, even take you … I wonder if they could get OR time and that's a whole other story, right? You can't do that but … The second time wasn't as bad. The third time was really, really bad and that was the time that I said that I wouldn't do it again and that would potentially make me noncompliant because it was so painful.

The last statement, "…would potentially make me noncompliant because it was so painful" is significant because when this was shared with the gynecologist they prescribed pain medication:

Participant 2: Dr. [gynecologist]., did then give me a prescription for I believe it was a cervical dilator, like a cervical like cream to help dilate it, I think, and like a stronger pain med so that the next time I would come in I would go fill that prescription, do the stuff like half an hour or whatever, how long beforehand and then do it. But after that then we had the conversation about [the] hysterectomy and I went that way to not have this [endometrial sampling] again.

The pain experienced by participant 2 during endometrial sampling was so great that she decided to undergo hysterectomy and bilateral salpingo-oophorectomy in order to, "not have this [endometrial sampling] again". It is possible that if the participant had previously been prescribed pain medication or had a local anesthetic in the cervix, she may not have had the prophylactic hysterectomy and bilateral salpingo-oophorectomy at such a young age. The participant later expressed regretting the prophylactic surgery:

Participant 2: If I still had those parts like part of me would like to have – you know, I thought maybe another one or two kids. He's [husband] like yeah, me too. I'm like but that's gone, right? You can't go back. Can't go back.

The experience described above demonstrates that it is of value for women undergoing endometrial sampling annually to receive pain medication and/or local anesthetic into the cervix. Pain from endometrial sampling should not be the sole reason for undergoing a prophylactic hysterectomy and bilateral salpingo-oophorectomy. Having this prophylactic surgery removes the

ability to have future children, and this decision should not be made due to painful procedures. Another participant experienced significant pain when undergoing endometrial sampling:

Participant 7: I think if it weren't for the fact that I know my family history and that there is a very good chance, I often consider the fact that I would almost rather get cancer and deal with that than have to continually force myself to voluntarily go through that procedure. Endometrial biopsies are the worst ... and not every doctor is really good at it.

The statement, "I would often consider the fact that I would almost rather get cancer and deal with that then have to continually force myself to voluntarily go through that procedure [endometrial sampling]", demonstrates the extreme discomfort that can be associated with endometrial sampling. The level of skill that a gynecologist has in performing endometrial sampling was also noted by the participant. This provides additional evidence that relational continuity of care for persons with LS and a choice of gynecologist improves the care received.

Another participant described that when they first inquired about receiving endometrial sampling it was discouraged by the gynecologist:

Interviewer: For endometrial sampling how is the procedure for you? If you could change anything what would you like to see occur?

Participant 8: So I went to him [gynecologist] the first time and didn't end up leaving with the endometrial sampling because of the way it was described, that it'd be very painful and it wouldn't be a pleasant experience, and that I should only do it if I'm sure about it, which I also think isn't great advice. I think if they think I need it I should be getting it regardless of pain, and they [gynecologists] should be managing that accordingly.

The quote, "regardless of pain and they [gynecologists] should be managing that accordingly" likely means that this participant had an expectation for pain management.

The section, care in gynecological surveillance, describes the pain that the female participants experienced during annual endometrial sampling. The suggestions made by participants were the use of adequate pain medication and ability to select the gynecologist who performs the procedure.

**Treatment after prophylactic hysterectomy and bilateral salpingo-oophorectomy**

The section, treatment after prophylactic hysterectomy and bilateral salpingo-oophorectomy, describes the experiences that female participants had following surgery and explores the recommendations for a personalized approach that they shared. A common experience shared by multiple participants was the need for self-advocacy in order to have hormone replacement therapy to assist with reducing the negative hormone related side-effects after surgery. Participant 3 shared:

Participant 3: But I was very happy that it was gone [uterus and ovaries] and I went instantly into menopause which wasn't so much fun but anyway.

Interviewer: And were you given any hormone options?

Participant 3: No. In fact, I … Well, what I did is after about two weeks of – after the surgery I was just like I can't take this. I was like drenched. I was just like miserable. I just phoned her [gynecologist] and said can you get me some hormone replacement and she did right away.

Participant 6 shared the negative consequences of not receiving hormone therapy directly after surgery for both herself and her sister:

Interviewer: And so you had the surgery and then you started, being forced into a menopausal state which caused, I would imagine, some issues?

Participant 6: Quite a bit yes. More emotional than anything. I just cried at the drop of a hat. It was horrible. So emotionally messed up, oh my goodness. I didn't suffer from the hot flashes. It was only when I was in bed that I really got really hot. During the day I was hot, but it wouldn't just come and hit me. For my sister she'd be sitting there and all of a sudden was like, oh - she'd be ripping her clothes off. But for me it was just almost constant. You know.

Participant 6 then shared that they scheduled an appointment with their family physician to receive or be referred to a specialist for hormone replacement as their quality of life was decreasing:

Interviewer: And then your family physician looked after your side effects?

Participant 6: Yes. I went to her and that's when I told her what was going on. So she - she said, I want you to go to the Mature Women's Centre. So I got into there and that's when they put me on Estrogel.

Interviewer: Okay, and has that been working well?

Participant 6: Amazing. Amazing. It works great for those side effects because they're gone.

The Mature Women's Centre was closed in 2017 by the Winnipeg Regional Health Authority[152] as the health region's only centre for hormone replacement therapy. Participant 6 explained that having a specialized centre was valuable to her because without the medical support she felt isolated:

Participant 6: It's not something easy, and especially for those of us who aren't post-menopausal, we get thrown to the depths of menopause where it's an emotional train wreck and you don't have anybody - like, you're wondering, "I'm going crazy". And you don't have anybody there to support you. And here with this Women's Centre, it's a God send. I think it is, anyways.

Another participant felt uncertain about undergoing a prophylactic hysterectomy and bilateral salpingo-ophorectomy due to the lack of medical care after surgery regarding side effects from the surgery induced menopause:

Participant 8: So my most recent appointment in fact – and I don't think that it was meant maliciously by any means, but I expressed concern over the fact that the Mature Women's Clinic was closing, that if I was going to go through with a hysterectomy and be well at my age, and was potentially going to compromise my quality of life, I wanted to know that the support would be there.

Participant 8 added that when she expressed this concern to the gynecologist, the response did not include that they were able to provide or refer for hormone replacement therapy for her:

Participant 8: I expressed my concern to the doctor [gynecologist] – and I understand I need the surgery and I understand it's imperative, but I want to know that the support is going to be there for me. And his response was, "Well you're a ticking time bomb, would you rather get cancer?"

The statement by the gynecologist ignored the concerns expressed by their patient:

Participant 8: And it's like, "Well no, that's not at all what I'm saying." And I understand that he was probably a little perturbed at the fact that – you know, it seems very obvious to him that I should just go ahead and get the surgery and get it done with, but there's a lot more implications.

Participant 8 also shared what she would have liked the gynecologist to say to her:

Participant 8: But rather than explain that to me – like if he had just turned to me and said, "Listen, don't worry. You're in our care; if you need additional estrogen therapy or care after the fact to help with your menopausal symptoms, we'll make sure that you see somebody that can help you with that." But he didn't say that.

When a woman considers undergoing a prophylactic hysterectomy and bilateral salpingo-oophorectomy there are, "a lot more implications" [Participant 8] to consider than merely the surgery itself. The experiences described above demonstrate the need for hormone replacement therapy to be discussed with women who have LS and for it to be readily offered after surgery. One participant shared their recommendations for the health care of women after prophylactic hysterectomy and bilateral salpingo-ophorectomy:

Participant 2: Like I would say they need to go home on – from surgery they need to go home on either the patch, like an Estradot patch or they need to go home on Estrogel. I had to go on – I went on Estradot first and I didn't absorb it all and so I started having hot flashes and all that and within three months I was like there's something not right here and I had – luckily, my coworkers had said you should go to Mature Women's Clinic, like that's something you should do.

Participant 2 also explains how in her case she had known to ask for a referral to the Mature Women's Clinic from her family physician. This request proved necessary as the initial hormone

therapy did not work well and another type of hormone therapy was ultimately prescribed. This experience demonstrates the need for follow-up for hormone replacement therapy:

> Participant 2: And so, I had gone to my GP [General Practitioner] prior to my hysterectomy asking for a referral there so that I had it all in place. And so when I went to get my blood work done three months after they're like you don't have any estrogen. You have nothing. You're not absorbing. I'm like okay, right? So then Estrogel and then I was like – I perked up right away as soon as I had that. But for any woman who has no idea, most women wouldn't even know.

Participant 2 recommends a clinic to follow-up and discuss hormone replacement therapy for women with LS:

> Participant 2: A young woman like me and like that's [menopause side effects] not fair. That's not fair at all. And so – and then now the guidelines are – like so for me I stay on estrogen until I hit my typical menopausal age, right? So I'm like I'm good. So yes, there should be something there and wouldn't it be lovely if there was, you know, here's a follow up clinic that you've had – you know, you've had your oophorectomy, you've had all this and let's help you through this stage. Let's help.

Above, participant 2 shared that her ideal setting for women undergoing prophylactic hysterectomy and bilateral salpingo-oophorectomy is a follow-up clinic, and she ended this statement with, "…you've had all this and let's help you through this stage". This outlines a need for ongoing care from the gynecologist who performed the surgery or a gynecologist specializing in hormone replacement therapy. This quote with the word choice, "…you've had all this…" also alludes to the impact experienced by women with LS who undergo prophylactic surgery.

### 4.2.5 Theme: *Ensuring continuity of care*

The theme, *ensuring continuity of care* was prominent in the interviews and a clear definition of the term 'continuity' is needed to ground this theme. The meaning of the term, continuity, is variable, with many definitions that depend on the health care domain in which the

term is used[153]. For example, in the primary care literature, continuity is mainly viewed in terms of a continuing relationship characterized by loyalty and responsibility between the same clinician (or team) and a patient. Over time this relationship produces longitudinality of care and enhances trust, communication, and responsibility[154]. In primary care, continuity is different from coordination of care, which refers to information transfer[153]. In the 1980s some authors defined continuity as a structural element (e.g. medical record)[155] but this view has been largely disregarded[156]. The American Academy of Family Physicians has defined continuity of care as "the process by which the patient and the physician are cooperatively involved in ongoing health care management toward the goal of high quality, cost-effective medical care"[157]. However, the term 'process' in the definition is not defined. As a result of multiple, confusing definitions of continuity, in my thesis I utilize two more exacting terms that fall under the umbrella of continuity of care: informational continuity as described Haggerty *et al.*[153] (defined in the following paragraph) and longitudinality as described by Starfield[158]. Starfield defines longitudinality as the, "building and maintaining of a long-term patient-practitioner relationship, regardless of whether there is a problem" (p.118)[158]. I consider coordination of care as a distinct theme.

The first type of continuity is informational continuity, which is defined as "the use of information on past events and personal circumstances to make current care appropriate for each individual"[153]. Information as the common thread linking care and detailed information regarding a medical condition is needed to provide health care[153]. Yet, for a physician to have extensive knowledge about LS is rare[159]. It has been identified that persons with LS would like to have a physician who utilizes information about LS involved in their care. The high probability of a cancer diagnosis requires a person with LS to trust the physician:

Participant 1: I have an 83% chance of having colon cancer; I'm not just a normal colonoscopy that Dr.[previous Endoscopist] thinks he's doing, and he treats me like a normal colonoscopy. But I have an 83% chance, so that's scary. So that's why I'm excited – because I'm seeing Dr. [new Endoscopist] in a different way now, but I know he's going to look in the right spots, I feel confident and I'm really excited to have him on my team.

Interviewer: Mm-hm.

Participant 1: I trust him. It's about trust with people.

Following their LS diagnosis, another participant also mentioned the lack of informational continuity of one physician, which prompted them to change physicians:

Interviewer: So about your first GI specialist, did you feel there was a lack of knowledge about Lynch syndrome or was your first specialist knowledgeable?

Participant 2: Yes, yes not that much knowledge.

The need for a clinician who utilizes LS information such as guidelines following their patients LS diagnosis was identified by some participants. One participant experienced some difficulty in finding an Endoscopist who took the time to be familiar with LS:

Participant 2: There's a lack of education about it, definitely. You know, the fact that I had to go to the research – like to the Lynch syndrome meeting to find out well, who's the guy? Who's the person that you guys go to [for colonoscopy]? Who's the guy that knows about this? There has to be a specialist here in town who specializes in Lynch, the go-to guy. And so that's when I found out about Dr. ___, right, which was great. And like as soon as I heard about him I'm like – well, I went to my doctor I think the next week and I'm like I need him.

And so you know, there's a lack of education most definitely, education for the public and education for the providers.

In addition to the lack of education of some physicians about LS, it was also identified that without longitudinal care, – where on-going relationships are established with clinicians it was more likely, according to one participant, that the health care provider would forget to share important details about someone's health (see quote below). Below I report having asked the participant a question with the term 'continuity of care' in order to obtain a sense anecdotally of what this term means to participants and here it was closest linked to longitudinal care:

Interviewer: Interviewer: Do you think continuity of care is important for people with Lynch Syndrome?

Participant 16: I would think so, yeah because if you have continuity of care you're less likely to or the medical profession's less likely to forget to tell you something when they should be telling you something I think, yeah.

One participant's surgeon shared a practice with another surgeon. The relationship between the two surgeons allowed for the participant to feel secure with either surgeon performing the colonoscopy as both surgeons had informational continuity, which translated into a form of longitudinal care. The participant shared:

Interviewer: And how do you feel about that [sharing of the practice]?

Participant 7: I feel a great deal better. Not only do I have confidence in both of them but I mean there's a comfort level that comes with that that I firmly believe they know what they're looking for. They know what I looked like last year – yeah, it's a huge comfort. And it's just nice to know it's them, I'm reassured by them. I really appreciate the work they do.

The above quote demonstrates that longitudinal care is not necessarily through a single physician. It can also occur in a shared practice with high levels of informational continuity.

Participant 8 explained that by having the same gastroenterologist perform the colonoscopy they knew it would be, "pretty painless because I have the same doctor". Additionally, it was shared by the participant that by having longitudinal care, "I know what's coming, I know what's

going to happen and he knows what to look for". The high level of importance of having longitudinal care for Participant 8 was affirmed with this statement:

> Participant 8: So yeah, I think it's great that I can go to the same doctor and that they know my case, especially with my family history, that they've now been educated on everybody. And I feel it's great they know that. And that's why, even if I left Winnipeg, I would fly home for my appointments. Like I would make a point of staying with these doctors.

Above, Participant 8 mentioned the security they felt with their physician's knowledge of their family history and the need to have longitudinal care. Two participants elaborated on how it was helpful to have the same physician care for their family members with LS. Participant 10 shared that their sister is their health advocate and as their surgeon's office knows this, the office has scheduled their colonoscopies one after the other on the same day, so they may be together. When an abnormal result was obtained the surgeon scheduled an appointment with both siblings:

> Participant 10: No, I think he [surgeon] understands my attitude towards it, right. So like any time we have appointments [colonoscopies] he brings my sister and me at the same time because then she's asking the questions and I'm getting the answers. Because I probably wouldn't ask questions, I would just go along with whatever. So the one time I did have positive results they made an appointment for both of us together at the same time, we went in together, obviously everything was fine, there was nothing wrong.

Participant 14 expanded on why continuity of care is important at both the family and person levels:

> Participant 14: Yes, it is important that the same person sees the family, it builds a relationship with patients. I do not want to see a different person; I do not know? If you are going for a procedure more often than the general population you deserve the same person. Why do we have to wait for people to get cancer to treat them like people?

The question posed by Participant 14, "why do we have to wait for people to get cancer to treat them like people?", highlights the need for thinking of the 'whole person' in terms of continuity[156]. Participant 12 expanded on the need for clinicians to provide care, which I define

as sincere benevolence[160], by describing an experience with the surgeon following a sub-total colectomy. When the participant re-counted the interaction, the emphasis was around the term 'care':

Participant 11: I'm eight days into recovery from my second surgery, and my intestines refused to wake up and so I can't eat, I'm feeling sick all the time, I'm puking up bile, and it's just horrific. And I'm walking the halls, and I walked out of my room, __ Surgical Ward, and he's [surgeon] down the hallway and he's talking to a nurse at the nursing station, and I turned, and I catch his [surgeon's] eye, and he's walking towards me, right. "___[name of participant], how are you doing"? I said, "I'm awful, I just want to get out of here". And he stops, and he puts his hand on my shoulder, and he said, "it's going to be okay". And that was the most healing experience I have ever had with a physician. It wasn't about how well he cut me, or put me back together again, it wasn't about follow-up, it wasn't about plans. But I knew that he actually *cared*.

**Continuity of care for gynecological surveillance**

Within the theme *ensuring continuity of care* there were instances when participants shared the specific value of continuity of care in gynecological surveillance. Participant 7 shared her experience of lack of longitudinal care:

Interviewer: So did you have the same gynecologist throughout?

Participant 7: I've not once seen the same – in the five years, six years now out, I've one doctor twice and I've seen multiple other doctors once. So I've seen Dr. _[name 1], I've seen Dr. _[name 2], I've seen Dr. ___[name 3], I saw a resident one year, I saw a visiting doctor from another clinic another year whose name I don't even know. Very seldom did I see the same doctor twice.

Interviewer: So there's no consistency with care?

Participant 7: No. Which is not great.

The participant goes on to describe why they would have benefitted from longitudinal care:

Interviewer: Would you be reassured in gynecology to have a consistent care provider?

Participant 7: Absolutely, it would be huge just to not have to every year feel like I have to reiterate my concerns or my stance or everything. Like I mean I know they've got the file and they know the basics, but every year I feel like I have to kind

> of explain myself, that I'm not a hypochondriac and I understand the risks. But I'm constantly having to do that again and again, and on top of it all I don't know how seriously they take me. And I don't necessarily feel like I'm a serious patient for them; I feel like I'm just one more in a long line of people they've got to pump through. It's very impersonal.

In contrast, Participant 8 had experienced longitudinal care for gynecological surveillance and for colonoscopy. Thus, when posed with a question regarding the importance of continuity of care, she shared the following:

Interviewer: And have you seen the same gynecologist for your procedure?

Participant 8: Yeah.

Interviewer: And do you feel that's important to have continuity of care?

Participant 8: Yes, so that's our biggest problem right now with our changing GPs so often is that we don't like that. That's why when I chose my doctors [gynecologist and endoscopist] I chose all doctors that are around the age of 50, could be my doctor for most of my life for of my care if I stayed in Winnipeg type of thing. Dr. __ is the gynecologist; he's probably in his late 40s. So I purposely chose doctors who either know a lot about Lynch or young enough to carry me through.

But yeah, I think continuing care is really important and that Lynch patients are referred to the doctors who know the most about Lynch and aren't on their own to find doctors for these areas.

The above quote demonstrates the selection criteria the participant had when investigating possible physicians for their care. To optimize informational continuity, Participant 8 considered the age of the physician and the physician's knowledge of LS when selecting this person as their specialist physician.

The value in continuity of care has been demonstrated for persons with chronic illnesses[161] who interact with multiple clinicians. Persons with LS are in a similar situation. Studies investigating chronic disease further emphasize that care coordination is of like value to

continuity[161] . The participants with LS in my study explained their need for care coordination as a distinct entity even though it can be viewed as related to continuity.

### 4.2.6 Theme: *Need for care coordination*

The theme of *need for care coordination* describes experiences of participants in organizing organ surveillance and as well as the communication of results between the physician and patient. The definition of coordination that I identify with is, "the delivery of services by different practitioners in a timely and complementary manner so that care is connected and cohesive for the patient".[162] Within this definition communication is necessary for optimal service delivery. Participants shared their powerlessness in trying to coordinate care and manage barriers they encountered, highlighting a need for some type of coordination in the health system.

Participant 11 described the powerlessness experienced in trying to coordinate care:

Participant 11: The problem with patients is that even really smart patients still have no power in the system. I cannot order a colonoscopy, I can't order drugs, I can't order any kind of anything. So, I am a powerless person who has essentially been told to run the show. Well, okay, how does that work? How does an architect with no power get a building built, right? That's the problem that we're in right now. And so, something needs to get fixed in that.

Multiple participants further described the experience of having to manage and coordinate care. Participant 4 described similar difficulty to Participant 12, with the additional experience of having a physician's nurse act as gatekeeper in scheduling care:

Interviewer: But now it sounds you're having to sort of request everything.

Participant 4: I have to request. No one tells me. There's no list of what I should be having or what I should be doing. I'm on my own and it's very hard because you're – you know, I don't know. I'm not a doctor. I don't know. And if I don't, if I don't advocate for myself I don't get it.

Interviewer: Yeah, there's not a lot in the system.

Participant 4: No. And even to make an appointment with my doctor because her nurse will say you don't need to come, you only need to come like every second year now, they're changing things and I'll say no. There's family history and I have Lynch and I need to be seen on a regular basis. So I'll push for that.

It has been proposed that coordination of care is a role of the family physician[75]. Yet, all participants in my study did not experience the family physician coordinating their care. For example one participant shared:

Interviewer: Okay, and does she [family physician] schedule your screening?

Participant 7: No, no, I book my appointments with her once a year and when I go in for that I ask her to prescribe me my urine cytology test at that time. And all the rest of my tests I primarily scheduled myself for the most part, up until this year when the ___[health authority] took – or sorry, last year, when the ___[health authority] took over the scopes [endoscopy]. I used to book those through my surgeon. I would just phone him up. I still book my own dermatology appointments and my gyno-oncology I was referred to shortly after my Lynch-positive diagnosis. And for that one they just keep me in their repertoire, so I just get my automatic yearly appointment requests.

The participant above explains the success of the central intake system that the health authority in Winnipeg enacted for endoscopy. Implementing an automated system that mails out appointment dates and times on behalf of a health authority's surgeons and gastroenterologists freed this participant from arranging their own colonoscopy or EGD. Participant 7 was also part of a gynecology oncology surveillance program through the health authority's cancer center which had an automated system for appointment generation. Another participant who has not experienced the central intake system for endoscopy described an automated system as something they would like implemented:

Interviewer: So you schedule your own screenings, from what I hear?

Participant 9: Right. Well, we go in to the - A colonoscopy is a bit different because it's through the surgeon's office, but we always get - It's never a year. It's always, like, I had one in January, February and then it was a year until June. So it was a year and a few months. This time it's going to be a year pretty much exactly because ____[my

wife] phoned and said we want it in a year. It's pretty easy. Like, I'll say it again, nobody says no, but just that would be nice, though, if it was just automatic.

Interviewer: Would you like an external source to coordinate that? Would that be easier?

Participant 9: It would be easier if, you know, like, when you take your kids to the dentist, you get something in the mail saying he's due for a checkup. That's all I want, right? So it's just automatic. I get my thing. Okay. It's coming up.

The quote below further demonstrates the role that a surgeon's office plays in coordinating endoscopy for a different participant:

Interviewer: So, do you ever use your family physician ever for your referrals?

Participant 10: No, now that it's kind of set in stone where I go every year at the same time and whatnot, I don't need the referrals anymore. I used them the very first time to get my referral and then every time after that it's just been close contact with Dr. ___'s [surgeon] office and whatnot and getting my testing done through them.

Interviewer: So the surgeon's office has taken charge of your care?

Participant 10: Yeah, he's excited and intrigued by the whole situation so he's pretty keen to get into it.

Interviewer: And do you think that's helpful having an office take charge and make the appointments for you?

Participant 10: Oh yeah, I wouldn't go if it weren't for them sending a letter- To be honest, it just wouldn't come to my mind; it wouldn't be something I think about first in my mind for something to do. It's not a fun experience, you know? So, yeah if they didn't send me a letter every year and I didn't have my sister nudging me to do it I probably wouldn't do it.

Participant 10's experience demonstrates that some persons with LS need coordination of care in order to participate in organ surveillance. Thus, the implementation of an automatized system may increase organ surveillance participation and lower the probability of advanced cancer and death.

**Communication between physician and patient**

Within the theme of *need for care coordination*, which depends on communication, all participants identified the issue of communication of results between physicians and patients. Experiences and suggestions for improvements included the need for physicians to communicate both abnormal and normal findings following organ surveillance. Some participants identified that when the results were negative, they did not have communication from the physician's office indicating the result. Instead they reached out to the physician's office to find out the result:

Interviewer: And when they found the polyps and they removed them and sent them to pathology, did you get feedback whether they were clear or whether they were positive?

Participant 17: Again, maybe it's just me being a fussy nurse, but then I called for results.

Interviewer: Okay, so you did have to call?

Participant 17: Yeah, I never did get results. So yeah, I phoned and said, "Can you send me the results?"

When I asked another participant if they would like to receive their results even if they were negative, they shared:

Participant 15: I would want to know either way. Just so you know somewhere along the line they missed it something and it actually a positive result and they missed it. And it is like you don't want to fall through the cracks.

The communication between a physician and patient regarding test results irrespective of the outcome was shared by the participants as being of value to them.

One participant shared an experience where a polyp was removed and they we were not told of its removal until a year later. The passage below demonstrates how a participant would have liked their endoscopist to share that a polyp was removed, as it had caused side effects:

Participant 4: Had a polyp there and now I found out that last year I had a polyp in my stomach and he [Gastroenterologist] never told me. They said it was all clear when I had the colonoscopy, but after I had it the next day I had cramping and I wasn't feeling great but no one told me that … that they removed something and they

should have and I just found that out today. So they're saying it's all clear because it looks clear and they think it's benign but if something is removed you need to know because then there's side effects. Yeah, and like – because after my last colonoscopy I didn't feel great the next day. Like I felt crampy and I – normally I'm okay. So that was why. They stuck some knife inside me and cut the polyp but they should have told me.

Communication therefore not only of results but also of what also what occurred during the procedure is important to persons with LS and likely all persons.

Some participants had received some communication regarding test results. One participant shared the value in receiving information in writing directly after the colonoscopy and/or EGD:

Interviewer: And do you get your results right away, like if you've had a clear scope, like how does that work?

Respondent: Yeah, usually there's a pamphlet waiting for me type of thing and I force anybody who's there to not look at --to read it just in case, like I don't let mom or anybody. I always say ahead of time put it underneath or whatever you have to do, I don't want anyone else to see it until I see it. And yeah, there's always some sort of feedback right after the scope. It will either say something detected, come back and see us, which was just the one time. The other times have been all clear, small other things that are important to Lynch syndrome.

Interviewer: Yeah. And do you find that helpful?

Respondent: I like to know right away, yeah. I don't like getting the note saying come back and see me. But I understand why, they don't really have any results depending on what they've tested for yet. All they can tell me is there was something, right? So I do like knowing right away.

The above quote demonstrates the value that a person with LS puts on having communication regarding the outcome of the endoscopy directly afterwards.

In the past a physician would often schedule a follow-up appointment with a patient following an invasive procedure. This follow-up appointment in the Winnipeg Regional Health Authority has largely been eliminated unless malignancy has been determined[163]. One participant lamented this change:

Interviewer: So, would you like to see if people want to have the colonoscopy and then having a follow-up appointment?

Participant 13: So, having a follow up appointment or having a hundred percent feedback on results would be good. So, I mean, if you have the colonoscopy and there are no polyps, there's nothing remarkable, then you know that day, right? But if you have the colonoscopy and they do take anything for biopsy, you should let the patient know what the results are.

The experiences shared by participants demonstrate the value of communication between the physician and the patient over results from organ surveillance.

### 4.3 Framework

The participants thoughts, their actions, and their adaptations are all profoundly influenced by their experiences in trying to access health care through an ideologically and bureaucratically complex health system. It became apparent that the possibilities for future health care put forth by persons with LS warrant a framework of what could be implemented in the health system. Creating a framework to improve access to health care for persons with LS was a three-fold process: 1) themes from the qualitative findings were organized into a figure, 2) the comprehensive literature search of health care for persons with LS was reexamined for guiding components, and 3) the study's findings and literature review were combined and organized into a figure.

#### 4.3.1 Themes associated with access to health care

First, Figure 2. represents the themes in my thesis and highlights the particular suggestions persons with LS shared to improve access to health care. Three circles – organ surveillance or treatment, communication, and continuity of care – intersect with one another to demonstrate the connection and overlap of each theme. Each circle contains the major research findings. Most are recommendations to facilitate health care for persons with LS (choice of bowel preparation, endometrial sampling pain management, medical support post- oophorectomy, knowledge,

informed of organ surveillance results, longitudinal care, informational continuity, coordination of organ surveillance and treatment). At the intersection of all three circles is the theme of personalized approach. The placement of a personalized approach provides a prominent cue that health care for persons with LS should be personalized, as each genotype has differing cancer risks and persons with LS are "not an average case" [Participant 2].

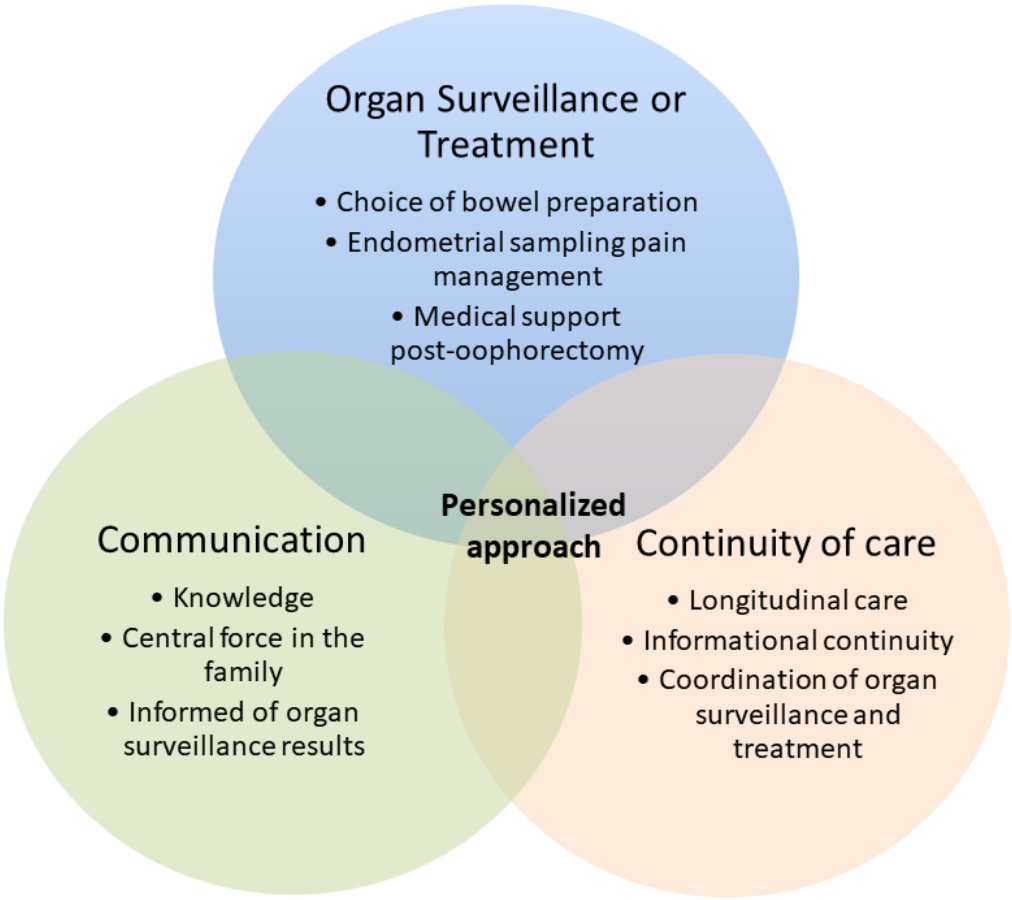


Figure 2. Themes from the thesis and suggestions from persons with LS to improve access to health care.

### 4.3.2 Concepts from the literature

To move forward with the development of the framework I returned to the literature. I read many published population based studies, systematic reviews, qualitative studies, and working group statements/letters as recommended by Imenda 2014[164]. The search strategy included the terms, "Lynch Syndrome", "management", "screening", "organ surveillance" and was run in MEDLINE (Ovid) from 2009 to April 2019. From these abstract screenings and full text retrievals of the articles containing testing and/or management recommendations or outlining clinical experiences, I identified salient concepts to address ways to improve access to health care for persons with LS. The following concepts were identified: Universal tumour testing for a MMR deficiency in colorectal, endometrial, and upper tract urothelial tumours; need for coordination in organ surveillance and cancer treatment; and need for hereditary cancer registries and their potential role. Below I present the literature leading to the development of the concepts.

*Concept: Universal tumour testing for a MMR deficiencies in colorectal, endometrial, and upper tract urothelial tumours*

In 2009, the Evaluation of Genomic Applications in Practice and Prevention Working Group recommended that all newly diagnosed patients with colorectal cancer, regardless of age or family history, should undergo tumor testing for a MMR defect, and those testing positive for the defect should be offered genetic counseling for LS. Since this recommendation there has been additional consensus around and movement towards universal testing for all colorectal tumors for MMR deficiencies[165]. In 2017, the National Institute for Health Care Excellence in the UK recommended universal testing for LS in all people newly diagnosed as having colorectal cancer, which has resulted in many National Health Trusts implementing this guideline[166]. Experiences regarding the

implementation of tumour testing for MMR deficiencies have been documented as successful for identifying persons with LS, who otherwise would not have been diagnosed[166].

In 2018, Adar *et al.*[167] found that expanding the universal testing of MMR deficiencies in colorectal tumours to include universal testing of endometrial tumours identified 50 percent more persons (and families) with LS. The authors recommended that universal testing programs for LS include both colorectal cancer and endometrial cancer.

Persons with LS disproportionally represent the population that develops upper tract urothelial carcinomas, as they have a 22 fold increase in developing upper tract urothelial carcinoma compared to the general population[168,169]. In 2018, Ju *et al*.[170] found three percent of upper urinary tract carcinomas tested positively for LS at their one hospital site. In colorectal and endometrial tumours LS is found in three to six percent of the population[7]. When considering the implementation of universal tumour testing, cost-effectiveness must be considered. Although the economics of the universal testing for MMR deficiencies in upper tract urothelial carcinomas have not been studied, work on universal testing for colorectal tumours has demonstrated that life year gains and cost savings of future cancers offset the cost of universal testing[74]. Given the rarity of upper tract urothelial carcinomas compared to colorectal cancer, the cost impact of universal testing of upper tract urothelial carcinomas for MMR deficiencies is likely to be minor[170].

*Concept: Need for coordination in organ surveillance and cancer treatment*

In response to the National Institute for Health Care Excellence in the UK recommendation for universal testing for LS in all persons newly diagnosed as having colorectal cancer, a poignant letter was written and published in the *British Medical Journal*[171]. The opening line states, "Lynch syndrome is currently under-recognised, underdiagnosed, and undermanaged, so opportunities to

reduce cancer mortality are often missed" (p.1)[171]. The letter goes on to outline that, "many [persons with LS] do not have personalised care strategies, and follow-up is inadequate"[171]. In March 2016, Bowel Cancer UK organized a clinical consensus to address the issues outlined above, and titled the output report from the consensus, "Improving services for Lynch syndrome: who's responsible?"[172]. The term in the report's title, "who's responsible?" relates to the lack of coordination of organ surveillance and communication between medical genetics and the clinicians who provide care to person's with LS. The main recommendations in the Bowel Cancer UK document are to: develop a national registry of persons identified as having LS (following consent from the person), develop a national high-quality surveillance program for person with LS to assure care, and provide every person with LS with a written personalized care plan. It is important to remain cognizant of the value of LS specific cancer treatment as a LS diagnosis can guide oncological decision-making. For example, universal testing of colorectal tumours can influence the choice of chemotherapeutic agents as per the current guidelines[173]. The need for coordination and communication of information across specialties of care was evident in the literature and my qualitative findings.

*Concept: Need for hereditary cancer registries and their potential role*

Literature published between 2006 and 2008 mentioned the possibility of hereditary cancer registries and genetic service centres working in partnership to ensure the latest guidelines are implemented and provide an opportunity for persons with LS to participate in organ surveillance[1,28,93]. How to orchestrate this sharing of responsibilities between the possible hereditary cancer register and genetic service centre was not outlined. The Bowel Cancer UK document[172] recommended a national registry for persons with LS: 1) to increase knowledge and

understanding of LS through anonymized data and 2) to coordinate consistent care services across the UK through automation for the scheduling organ surveillance[172].

### 4.3.3 Developing the framework

When developing the framework, the concepts from the literature review informed multiple features and the qualitative findings from my study also contributed to aspects of the framework. I describe each aspect in the order that they appear visually in Figure 3.

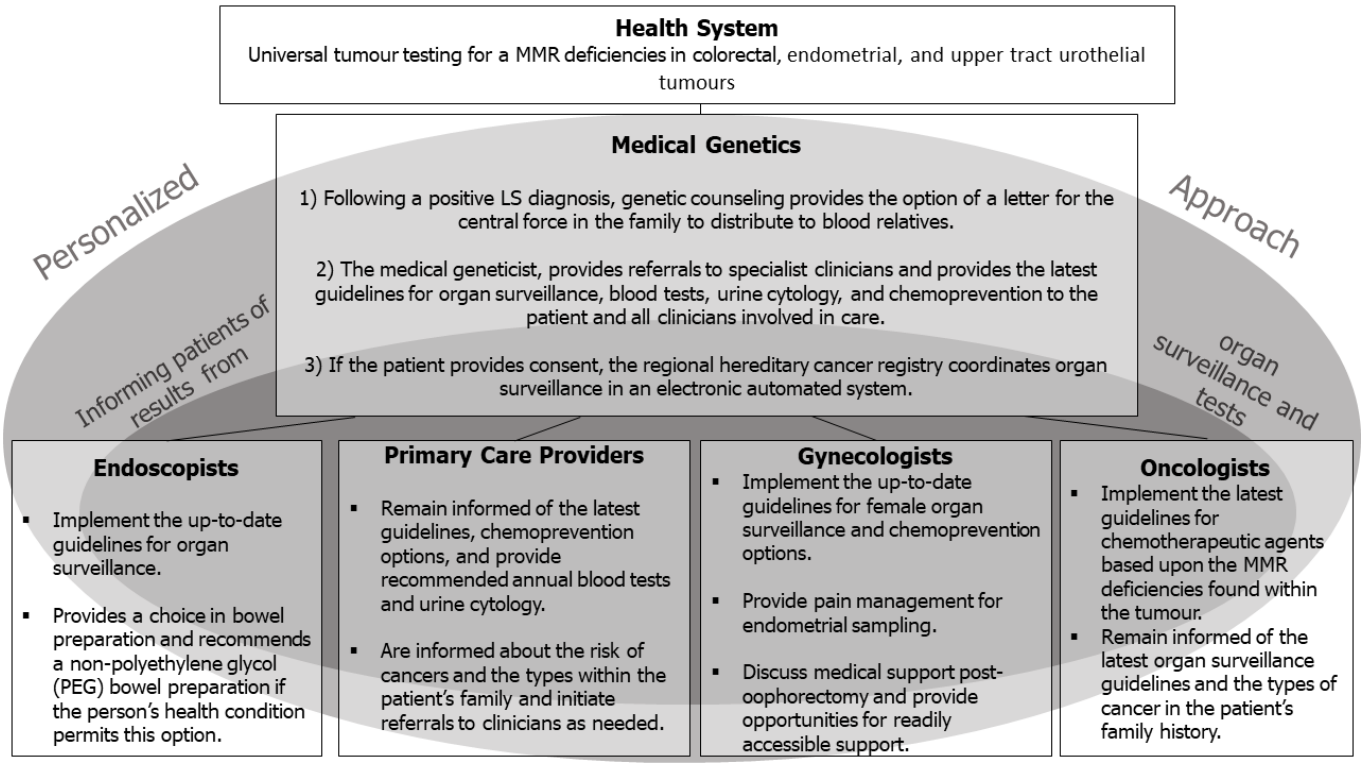


Figure 3. Framework to improve access to health care for persons with LS.

*Rectangle in framework: Health System*

The literature review, suggested that the role of the health system at the policy level, is to implement universal tumour testing for a MMR deficiencies in colorectal, endometrial, and upper

tract urothelial tumours. A recent Canadian study investigating universal tumour testing has suggested the need for national standards and guidance on tumour screening for LS[174].

*Overarching concept in the framework: Personalized Approach*

The Bowel Cancer UK document[172] recommends, "everyone with Lynch Syndrome providing everyone with a written personalised care plan at diagnosis to support involvement in making suitable treatment decisions for their care"(p.125). A recurring theme across many interviews in my study is that persons with LS are unique with needs that differ from the general population. Participant 2 stated, "You're not an average case". This statement applies to all aspects of medical care for persons with LS: continuity of care, coordination of organ surveillance, bowel preparation for colonoscopy, chemotherapy, gynecological cancer surveillance, and hormone therapy or treatment options following prophylactic oophorectomy. Thus, a personalized approach is needed by every clinician (medical geneticist, genetic counsellor, endoscopist, primary care provider, gynecologist, and oncologist) who provides care specific to LS.

*Overarching concept in the framework: Informing patients of results from organ surveillance tests*

My study found that many participants did not know the results of their organ surveillance procedure or tests. It was often the responsibility of the patient to call the office to ensure that nothing abnormal was found. The framework proposes sharing with the patient the results of every one of their organ surveillance procedures and tests.

*Rectangle in framework: Medical Genetics*

Genetic counselling has been mentioned as an integral part of LS testing since the mid-ninties[89]. My qualitative findings reinforced the suggestion for genetic counsellors to provide a

letter to the proband to distribute to family members[80] and demonstrates that the proband may not be the ideal central force within the family to communicate the LS diagnosis to blood relative. It is recommended that genetic counsellors discuss the ideal communicator in the family with the proband. Previous research[175] has suggested that the medical genetics department share the latest LS guidelines with the patient and all clinicians involved in care. My study findings also reinforced the need for persons with LS and their clinicians to be reminded of and have access to the latest guidelines. It has been suggested that hereditary cancer registries take an active role in creating automatic scheduling reminders for organ surveillance[172].

*Rectangle in framework: Endoscopist*

If endoscopists are unaware of the latest LS guidelines under- or over surveillance can occur. Being aware of latest guidelines is only part of implementing evidence based medicine (integrating clinical expertise and the best external evidence), as defined by Sackett[176]. Forty-seven percent of participants in my study had gastroscopies performed on an annual basis, although the guidelines recommend a 3-5 year time period, depending upon the family history of gastric cancer[177]. The side effects from PEG as a bowel preparation regimen are well documented[148] and all participants in my study who had undergone colonoscopy did not fare well consuming PEG. It is recommended that a non-PEG bowel preparation formula be offered to persons with LS undergoing colonoscopy.

*Rectangle in framework: Primary Care Providers*

Primary care providers are usually responsible for providing urine cytology and blood tests, and need to be aware of the latest LS guidelines[175]. Although guidelines[138] recommend annual urine cytology for persons with LS, in my study, 2 out of 17 participants had urine cytology performed. The framework suggests that primary care providers who have patients with LS be

aware of the latest LS guidelines. Primary care providers need to be familiar with the patient and initiate referrals for additional screenings or referrals as guidelines change or concerns arise[75].

*Rectangle in framework: Gynecologists*

In the US, 65 percent of gynecologists have never received formal training in DNA-based genetic testing and do not feel confident in their knowledge of genetics[178]. The role of medical genetics in providing all specialists involved in care for persons with LS updated LS guidelines is also important for gynecologists. Pain was mentioned as a serious deterrent to women undergoing endometrial sampling in my study and the framework suggests that pain management be provided. My study demonstrates the need for gynecologists to discuss hormone replacement options before the oophorectomy and provide opportunities to access medical support post-oophorectomy.

*Rectangle in framework: Oncologists*

MMR results from tumours are known to guide surgical and oncological decision making. A positive genetic test for LS can influence the choice of therapeutic agents[173]. One participant in my study was unable to receive LS testing before her oncologist started her chemotherapy, and later regretted undergoing chemotherapy as it had no benefit for LS tumours.

# CHAPTER 5: DISCUSSION

## 5.1 Contextualising the findings with existing literature

My study demonstrates what barriers and facilitators a sample of 17 persons with LS experience in receiving care. Some of the experiences shared by the participants in my study were previously identified in the qualitative works by: Watkins *et al.* (2011)[75], Etchegary *et al.*

(2018)[179], and Schneider *et al.* (2018)[175]. However, the experiences shared by the participants in my study contributed to new perspectives on some elements that warrant further discussion in light of existing literature. I reiterate here as in Chapter 2, that previous qualitative studies about LS have tended to focus on psychological outcomes and/or emotional issues rather than barriers and facilitators of access to care, which may explain why my work aligns with only three qualitative studies.

Watkins *et al.* (2011) utilized semi-structured interviews for 23 participants with LS, all of whom had MSH2 genotype affected. In contrast the participants in my study had a broad spectrum of genotypes (MlH1, MlH1-Muir Torre, MSH6, MSH2, and EPCAM) affected, which provides a previously absent holistic view of the experiences of persons with LS. The findings from Watkins *et al.*, state that "emotional forces were found to impact a person's willingness to participate fully in organ surveillance" (p.4.). In my study emotional forces were not described as a facilitator or barrier to participating in organ surveillance, but rather a coordinated and automated scheduling system was shared to be beneficial, but rather a coordinated and automated scheduling system was beneficial. I am unable to provide reasons for this, but it is possible that my study participants focused more on 'trying to improve' their care and share facilitators and barriers at the health system level than on discussing emotional barriers. For example, one participant from my study said, "I wouldn't go if it [colonoscopy] weren't for them sending a letter". Another participant who had not experienced an automated system articulated, "that would be nice, though, if it was just automatic". Additionally, Watkins *et al.* described that the participants had, "conflicting emotions about knowing what had to be done" (p.4.) for disease management, whereas my study participants viewed up-to-date information positively and wanted it for themselves and/or their clinicians. For example, one participant shared, "There's no

list of what I should be having or what I should be doing… if I don't advocate for myself I don't get it". It is evident that Watkins *et al.* had a strong psycho-emotional lens. Most studies have focused on psychological outcomes as opposed to barriers and facilitators experienced by persons with LS. Watkins *et al.* continue this line of focus in all but one of their themes. Both my study and theirs identified the limited organization and coordination of organ surveillance as a barrier. Participants from their study and my study experienced challenges when navigating the health system. Watkins *et al.* recommend a more coordinated approach but no concrete solutions in how to achieve that.

The recently published work of Etchegary *et al.* (2018)[179] consisted of telephone interviews of 10 women with LS to explore how they described their quality of life post-prophylactic hysterectomy bilateral salpingo-oophorectomy. Some of our findings were similar. Their participant's had a large reduction in cancer anxiety after the surgery. None of the 11 female participants in my study explained their justification for the prophylactic hysterectomy bilateral salpingo-oophorectomy solely in terms of reducing worry. For example, Participant 3 shared, "but I was very happy that it was gone [uterus and ovaries]". Whereas Participant 8 shared, "I understand I need the surgery and I understand it's imperative". Most participants in my study viewed prophylactic hysterectomy bilateral salpingo-oophorectomy as a necessary procedure for their health. One participant chose prophylactic hysterectomy bilateral salpingo-oophorectomy as an alternative to undergoing painful annual endometrial sampling. Etchegary *et al.* noted that there are gaps in women's knowledge about the sudden onset of menopause and the availability of hormone replacement therapy. This finding was also evident in my study and the participants shared that this lack of knowledge was caused by the gynecologist withholding information regarding the possible side effects. Etchegary *et al.* described a theme titled, *Post-surgical follow*

*up support*, but only one quote supported this theme, directly related to side effects or concerns post-surgery. The quote centered around the participant's wish to speak to a counselor about the impact of surgery on her marriage. The participants in my study shared the significant challenges they faced in receiving hormone replacement therapy post-surgery but it seems that this was not the experience of participants in the study by Etchegary *et al*. They were selected through the same tertiary care setting sharing the same specialist clinician, which the authors identify as limiting the findings generalizability. In contrast my study participants received care from a variety of care centers and clinicians, including outside Manitoba and outside Canada.

Schneider *et al.* (2018)[175], interviewed 12 persons with LS and 10 primary care providers with at least one patient with LS. Their research aimed to understand how LS organ surveillance is coordinated and monitored. They wrote that, "all patients believed their overall LS care was coordinated and felt generally supported by their providers" (p.5). This statement does not align with my research findings. Most of my participants struggled to coordinate organ surveillance in the publicly funded Manitoba health system. However, Schneider *et al.* (2018) recruited participants from a single integrated care-delivery system at Kaiser Permanente Northwest, a private corporation in Portland, Oregon, U.S. Four of the participants relied on their own efforts to track and complete organ surveillance.[175] In my study the Winnipeg Regional Health Authority's implementation of the central intake system for endoscopy limited the number of participants who had to schedule their own gastroscopies and colonoscopies but other organ surveillance had to be scheduled by the majority of participants. Schneider *et al.* (2018) recommend a comprehensive monitoring program with electronic reminder prompts for organ surveillance, but do not provide quotes of persons with LS making these recommendations.

Watkins *et al.* (2011)[75], Etchegary *et al.* (2018)[179], and Schneider *et al.* (2018)[175] make no

mention of participants describing instances where the clinician does not share the results of their organ surveillance procedure. In my study 14 of 17 participants said they had to call a clinician's office to ensure the result was clear of cancerous findings. They described a culture where clinicians or their office would only share the result with the patient, if that result indicated further treatment. Participant 15 suggested that if a person with LS is never informed of their organ surveillance result something could be missed, and in his words "it is like you don't want to fall through the cracks". This unique finding could indicate a possible culture in Manitoba.

Previous qualitative research has not asked persons with LS about their experiences with bowel preparation. My study findings provide the perspectives of persons with LS who have ingested bowel preparation regimens for many colonoscopies. To mitigate the side effects of nausea and vomiting, the utilization of a non-PEG bowel preparation regimen was suggested by the participants. All participants in my study either no longer ingested PEG bowel preparation regimen or had asked their endoscopist for permission to take a different bowel preparation regimen, in which the endoscopist refused to change the bowel preparation regimen.

My study fills a gap in the literature by asking persons with LS to share their experiences in accessing health care for LS, and inviting suggestions on how to improve their access to care. These lines of inquiry move away from previous research concentrating on psycho-social barriers and facilitators in organ surveillance and genetic testing, since my focus is on what the barriers and facilitators are in the health system when persons with LS try to access care.

### 5.3 Findings in relation to bioethics

Beauchamp and Childress (2001)[143], propose using four principles when making ethical decisions in health care: respect for autonomy, beneficence, non-maleficence and justice.

Respect for autonomy is strongly linked to informed consent. The elements of informed consent have evolved to include[180]: 1) competence, 2) disclosure, 3) understanding,4) voluntariness, and 5) consent. My study findings relate to the disclosure (of material information), where the legal doctrine of informed consent, "has been primarily a requirement of disclosure based on a physician's general obligation to exercise reasonable care by providing information" (p.81)[143]. The experiences shared by some participants where physicians either denied the possibility of LS or chose to remain silence regarding the possibility LS have greater bearing when viewed from the moral viewpoint of informed consent. Consent, "has less to do with the liability of professionals as agents of disclosure and more do with the autonomous choice of patients" (p.81)[143]. Some participants and families in my study were not given an autonomous choice to receive enough information to make informed decisions about their care. Yet, within the same health system some physicians chose to share with their patients the possibility of LS. These instances demonstrate that physicians' morals and virtues differ from one another.

The principle of non-maleficence, "asserts an obligation not to inflict harm on others" (p.113)[143]. There may have been time when genetic testing was considered an extraordinary treatment for a health system. Traditionally, it has been thought that extraordinary treatments can legitimately be forgone, whereas ordinary treatments cannot be forgone[143]. Genetic testing falls into a liminal space between the two terms, as earlier in the advent of medical genetics it may have been more towards extraordinary care. The US taskforce[177] and the National Comprehensive Cancer Network®[138] make it clear that genetic testing for LS is necessary.

No clear boundary exists between the principle of non-maleficence and the principle of beneficence. The principle of beneficence, "demands more than the principle of non-maleficence because agents must take positive steps to help others, not merely refrain from harmful acts" (p.165)[143]. Often in terms of beneficence at the health policy level, issues arise in relation to the comparison, relative costs and risks and benefits. Universal testing for MMR deficiencies of tumours commonly associated with LS cancers has been questioned and investigated as to whether there is a cost-benefit. The benefit exceeds the cost when a person with LS shares the diagnosis with three blood relatives so that they can be tested[74]. However, it is assumed that persons will participate in organ surveillance that is accessible and adhere to guidelines. When a health system enacts universal tumour testing, it is recommended to investigate coordination and guideline adherence, and implementing an approach as described in Figure 2., to balance the costs and benefits.

The principle of justice is often related to inequality in health care, yet there is uncertainty, "over how to reconcile goals such as equal access to health care" (p.225)[143]. In my study the historical fact that Manitoba was the last province in Canada to permit LS proband testing[50] does question access to health care. In a country where funds for health care are managed somewhat independently by the Provinces, geographic disparities in access to health are likely to persist.

### 5.4 Limitations

The findings of studies should be interpreted in light of limitations. Although the sample size of 17 participants provided an opportunity to reach data saturation in key themes, the participants were recruited from a LS educational group or attendees of a LS awareness evening. Thus, most participants had a keen interest in information for LS care management. Even so,

many described challenges in accessing care in the health system, raising concern about how other persons with LS fare in having organ surveillance performed. The findings are based on participants recalling how they experienced and reacted to specific events and situations. No attempt was made to validate the objective veracity of events recounted in their narratives.

# CHAPTER 6: REFLECTIONS AND CONCLUSIONS

This concluding chapter brings together the disability studies scholarship I presented in Chapter Two to reflect on how some of the findings may align with the experiences of persons with disabilities. At the end of this Chapter, I present the concluding statements.

## 6.1 Reflections and Connections to Disability Studies

The disability movement famously declares, "Nothing about us without us!"[181], and my person-centred research approach is committed to this declaration, as indicated by getting 15 out of 17 of participants to respond to an invitation to participate in member-checking for the final themes. Some participants highlighted areas (*personalized approach* and *knowledge* amongst specialists) for emphasis and these became prominent statements in the health system framework. I actively tried to prevent my research from following a paternalistic approach by valuing the lived experiences of persons with LS. The participants' narratives are so valued that I consider those experiences to be a catalyst for change, as demonstrated in the framework.

It has been documented by Hansen (2009) that at the clinic level clinicians often do not use lived experience of persons with disabilities as a resource[182]. In my study, the clearest example of a clinician ignoring the lived experience of a person with LS occurred when the participant

asked their endoscopist if they could use the same bowel preparation regimen that was prescribed to them in the US, as the participant had experienced very few side effects with that regimen:

Participant 17: And I was like, "Dr. [endoscopist's name], can I do MiraLAX and Gatorade?" [he responded] "No." "What? It's GoLytely again?" Or Peg Lightly or whatever – the big jug.

Interviewer: The four liters?

Participant 17: Yeah.

Interviewer: So he made you take that PEG- GoLytely?

Participant 17: Yeah. That [other] prep is heaven. But GoLytely is just a f**ker.

As demonstrated by the quote above, when clinicians ignore the lived experiences of persons, they may add unneeded suffering and reduce the quality of life (even temporarily) of their patients.

Less obvious to outright ignoring lived experiences were scenarios where clinicians ignored the concerns of persons with LS. This behaviour was most prominent in the experiences of women who voiced their concerns around the possible side effects and supports available following prophylactic hysterectomy and bilateral salpingo-oophorectomy:

Participant 8: I expressed my concern to the doctor [gynecologist] – and I understand I need the surgery and I understand it's imperative, but I want to know that the support is going to be there for me. And his response was, "Well you're a ticking time bomb, would you rather get cancer?"

The statement by the gynecologist ignored the concerns expressed by their patient:

Participant 8: And it's like, "Well no, that's not at all what I'm saying." And I understand that he was probably a little perturbed at the fact that – you know, it seems very obvious to him that I should just go ahead and get the surgery and get it done with, but there's a lot more implications.

Participant 8 also shared what she would have liked the gynecologist to say to her:

Participant 8: But rather than explain that to me – like if he had just turned to me and said, "Listen, don't worry. You're in our care; if you need additional estrogen therapy or care after the fact to help with your menopausal symptoms, we'll make sure that you see somebody that can help you with that." But he didn't say that.

It is interesting that women with LS in my study experienced additional barriers in accessing health care specific to LS. Quantitative scholarship investigating disparities in health care access in the US for women and men with disabilities and without disabilities, noted that identifying as a woman without disability produced a 1.26 times greater likelihood of having no access to health care when compared to those identifying as a man, and women with disabilities were 3.78 times more likely to have no access to health care[183]. Identifying as a woman with or without disability corresponds with increased barriers to accessing health care.

Hansen (2009)[182] identified the following theme by interviewing women with disabilities, *The "Place" of Technology in Health Care*, where the internet was identified as a key health information tool. In my study many persons with LS relied heavily on information sourced from outside Manitoba. As documented by Hansen, clinicians can be "gatekeepers" to health care and knowledge. Persons with LS in my study also experienced the effects of gatekeeping. To circumvent the lack of knowledgeable clinicians in Manitoba, one participant reached out to the clinician who discovered LS, and was practicing in Nebraska, US:

Interviewer: Did you do outside research before about Lynch syndrome?

Participant 4: I did. Once my friend had told me about Lynch syndrome I did. I did a lot of research about it. I wrote to Dr. Lynch who responded right away and told me all the steps that we needed to take and what we should be doing and the testing that we should have.

The above information exchange demonstrates an opportunity for clinicians to be allies with persons with LS. As Dr. H.T. Lynch is the clinician who discovered LS, he is very familiar with the impact that LS can have on families, and the value of allies to persons with disabilities,

particularly close allies (e.g., family members, partners, and friends), as they are familiar and have a deep understanding of disabilities. This raises the concern that only persons familiar with disability, a syndrome, or an illness can become allies. If this were the case the use of the social model to fundamentally change attitudes may not be 100 percent effective. Rather persons may need personal experiences with those that they view as 'different'. However, the social model highlights the need for equality, and the removal of physical and attitudinal barriers created by society, including the health system[112].

The main concept of the social model drawn upon in my study was the attitudinal barriers present in the health system, as described by the participants. The themes in my study that capture a sense of attitudinal barriers are, *It's health care, not 'Russian Roulette'* and *Knowledge*. Within the theme, *It's health care, not 'Russian Roulette'* the attitudinal barrier encountered was the health system not providing an opportunity for persons to test for LS even though all the other provinces at the time provided LS testing[50]. The attitude encountered in Manitoba before LS testing was initiated, was that *LS testing is not of importance*. In the theme *Knowledge* some clinicians did not seem to investigate the literature concerning what LS was and the clinical guidelines regarding when LS testing should be initiated.

Health care barriers experienced by persons with disabilities need to be investigated, "to avoid the costly and detrimental consequences for both people with disabilities and the society in which they live, which often pays for these consequences"[184]. The same can be said for persons with LS for when LS testing is denied, and barriers are experienced when persons with LS try to access organ surveillance. There can be detrimental effects for the person such as cancer and years of potential life lost, and the health system must expend funds for cancer treatment and palliative care, that could have been avoided.

In reflection, for health systems to provide care that centres around flourishing and the common good, concepts from disability studies can be translated into an approach to disrupt the traditional model that does not view persons as persons, but rather in terms of their disability or health condition. Person-centred health care[86] is a way to illuminate the clinician and patient as moral equals to facilitate an ethic of virtue to respect the occupancy of both social roles by whole persons.

### 6.2 Concluding Statements

The first goal of this research was to explore the barriers and facilitators to genetic testing and organ surveillance in the same health system to identify whether persons with LS receive sufficient care for the genetic testing cost benefit ratio to balance[74]. Persons with LS shared experiences that indicate barriers in accessing health care. The findings underscore gaps in health care coordination for persons with LS from previous studies and add to this growing area of literature.

The second goal of this research was to develop a framework to improve access to health care for LS, utilizing suggestions from persons with this condition. The implementation of the framework at the health system level and clinician level could steer resources to improve the uptake of genetic testing, adequate organ surveillance, and provide high quality care for persons with LS.

## Appendices

Appendix A: Letter I and Letter II from Health Minister

Appendix B: Study Invitation Letter

Appendix C: Survey

Appendix D: Interview Guide

Appendix E: Excerpt of Reflexivity Journal and Process Notes

Appendix F: Ethics Approval and Renewal Certificate

Appendix G: Informed Consent Forms

Appendix A: Letter I and Letter II from Health Minister

MINISTER OF HEALTH

Room 302
Legislative Building
Winnipeg, Manitoba, CANADA
R3C 0V8

March 1, 2011

Dear :

The Honourable Kerri Irvin-Ross, Minister of Housing and Community Development, has forwarded to me your correspondence regarding testing for Lynch syndrome. I would like to commend you for advocating on behalf of people affected by Lynch syndrome and take this opportunity to provide you with the following information.

I would like to assure you that if a physician believes a person should have a certain type of test, such as for Lynch syndrome, that there are avenues the physician can pursue for this testing. This is why it is crucial for people always to speak openly with their physicians.

Diagnostic Services of Manitoba Inc. (DSM) does testing for Lynch syndrome in Manitoba. DSM has advised that 90% of Lynch syndrome cases have no known genetic association. For the 10% of cases that do have a genetic association, 4 genes have been identified, and DSM provides testing for all 4 genes.

Medical professionals at DSM have stated that the most commonly occurring gene in Manitoba is specific to individuals with a Mennonite heritage, which makes that specific gene the most requested for testing in Manitoba. The higher volume of tests allows DSM to maintain testing for that gene locally in Manitoba, while the less frequent testing for the other three genes are sent out of province to larger, more specialized labs that have the expertise for a full gene analysis. Contrary to media reports, should a Manitoban of any cultural heritage require testing for Lynch syndrome, they can receive that testing.

Again, I commend you for your advocacy and thank you for writing.

Regards,

Theresa Oswald

c Honourable Kerri Irvin-Ross

MINISTER OF HEALTH

Room 302
Legislative Building
Winnipeg, Manitoba, CANADA
R3C 0V8

May 11, 2011

Dear :

I have been in touch with Diagnostic Services of Manitoba Inc. (DSM) and regrettably have learned that inaccurate information was sent to you in my March 1, 2011, letter, regarding Lynch syndrome.

DSM and I are sincerely regretful of the miscommunication, as well as any inconveniences caused to you and your family. I understand that Dr. Amin Kabani, Chief Medical Officer, DSM, has been in touch with you to personally address your concerns, as well as to clarify the misinformation that was sent to you. I am assured that Dr. Kabani will remain in contact with you to fully address all of your concerns related to Lynch syndrome.

DSM has provided clarification regarding the misinformation that was provided to you in my March 1, 2011, letter.

- Item 1: Diagnostic Services of Manitoba Inc. (DSM) does testing for Lynch syndrome in Manitoba. DSM has advised that 90% of Lynch syndrome cases have no known genetic association.
- Correction: This statistic should have read that 90% of *colorectal cancers* have no known genetic cause. Further, Lynch syndrome does account for three to eight percent of all colorectal cancers, and in most Lynch syndrome cases, there *is* a genetic association.

- Item 2: For the 10% of cases that do have a genetic association, 4 genes have been identified, and DSM provides testing for all 4 genes.
- Correction: Three to eight percent of colorectal cancer cases do have a genetic association, and these individuals have Lynch syndrome. Four genes are responsible for the majority of these cases.

- Item 3: Medical professionals at DSM have stated that the most commonly occurring gene in Manitoba is specific to individuals with a Mennonite heritage, which makes that specific gene the most requested for testing in Manitoba. The higher volume of tests allows DSM to maintain testing for that gene locally in Manitoba, while the less frequent testing for the other three genes are sent out of province to larger, more specialized labs that have the expertise for a full gene analysis.
- Correction: It is for historical reasons that we are testing for *one* Mennonite mutation. This is not the most commonly occurring mutation in the Manitoba population; however, it is more common in Manitoba than in other provinces given our larger population of individuals of Mennonite descent. Other mutations in these four genes have been found in the Mennonite population.

- Item 4: Contrary to media reports, should a Manitoban of any cultural heritage require testing for Lynch syndrome, they can receive that testing.
- Correction: Only Manitobans where the proband has been tested elsewhere can have testing for a family-specific mutation.

Genetic testing plays an ever-important part in the health of Manitobans with a high-risk of hereditary cancers. Our government is committed to exploring all opportunities for cancer screening that would promote early detection and improved prognosis, including Lynch syndrome testing.

I would like to reiterate my sincere apologies for the oversight that resulted in incorrect information being supplied to you. Thank you for your understanding.

Regards,

Theresa Oswald
Minister of Health

c Mr. Reg Toews, Chief Executive Officer, DSM
Dr. Amin Kabani
Mr. Paul Penner, Chief Operating Officer, DSM
Dr. Laurel Thorlacius, Clinical Biochemistry and Genetics Medical Director, DSM
Dr. Beth Spriggs, Director, Molecular Diagnostic Lab, DSM
Dr. Bernie Chodirker, Director, Prenatal Diagnosis Program, DSM
Ms Arlene Wilgosh,
President and Chief Executive Officer, Winnipeg Health Region
Mr. Milton Sussman, Deputy Minister of Health

## Appendix B: Study Invitation Letter

**INVITATION TO PARTICIPATE IN AN INTERVIEW: Experiences of genetic testing and organ surveillance for persons diagnosed with Lynch Syndrome**

Ethics Protocol J2017:093, HS21063

I am inviting you to participate in a research study that is investigating the experiences of person with Lynch Syndrome in having genetic testing completed and coordinating/participating in organ surveillance. This research has been approved by the Joint-ethics board at the University of Manitoba and you may contact the Human Ethics Coordinator at 204. 474.7122 or via email at humanethics@umanitoba.ca if you have any concerns.

**Advisor:** Dr. Nancy Hansen, Ph.D.
Email: nancy.hansen@umanitoba.ca
Phone number for advisor: 204. 474.6458

The primary investigator is a Master of Science Candidate (Kathleen Zawaly) and this research would be part of her thesis. The first aim of this research is to identify whether or not there is a lack of ease in the health care system that prevent persons with Lynch Syndrome or their families members in having genetic testing completed. The second aim is to identify how and if persons with Lynch Syndrome participate in organ surveillance (for example bi-annual colonoscopy).

The study is gathering this information through a combined interview and a survey. You would be asked questions in the interview about how you experienced genetic testing and how or if you participate in organ surveillance. You would also be asked to complete a short survey form about your Lynch Syndrome diagnosis journey, current organ surveillance routine (if applicable), and basic demographics.

**Costs of Research:** It will take one hour to participate in the interview. Parking will be reimbursed (if applicable).

**Risks or Discomforts**
There should be no risks or discomforts associated with this study other than sitting in a room for at least an hour.

**Benefits**

By participating in this study, you may gain some benefit by being able to share your experience of having Lynch Syndrome. There may or may not be direct benefit to you from participating in this study.

If you would be interested in participating in the interview and survey or have any questions, please contact Kathleen Zawaly, email: zawalyk@myumanitoba.ca or telephone _____.

Thank you for your consideration.

Kathleen Zawaly
B.Sc. (McGill), M.Sc. (Candidate, University of Manitoba)

## Appendix C: Survey

### Descriptive Statistics Survey

**About You:**

1. Your Gender

(*Select only one*)

☐ Female

☐ Male

☐ Other

2. Your Age

(*Select only one*)

☐18-25

☐26-35

☐36-45

☐46-55

☐56-65

☐66-75

☐76 or more

3. Your Relationship Status

(*select the one that describes your current status best*)

☐Single

☐Divorced or Separated

☐Common Law

☐Married

4. In what year did you receive genetic testing for Lynch Syndrome and in what City, Province and Country?

Year_____

City_____Province_____Country_____

5. Did you receive genetic testing after cancer diagnosis?

☐ No

☐Yes

If Yes please specify your cancer diagnosis and if there has been recurrence and if the cancer was identified elsewhere:

_____

_____

_____
_____

6. Did you receive genetic testing due to a family members' positive test?

☐ No

☐ Yes

7. If you were or if you currently have children would you share with them your Lynch Syndrome diagnosis and support them if they choose to pursue genetic testing?

☐ No

☐ Yes

8. What genotype was your Lynch Syndrome diagnosis associated with?

☐ MLH1

☐ MSH2

☐MSH6

☐ PMS2

☐ EPCAM

9. Are you currently involved in scheduled cancer screening?

☐No

☐Yes

10. If Yes, which of the following screening procedures do you receive?

(*select all that apply*)

☐ Colonoscopy

☐ Flexible Sigmoidoscopy

☐ CA 125 Blood Test

☐Pelvic Ultrasound

☐ Transvaginal Ultrasound

☐Endometrial, Uterine Biopsies

11. Have you had any of the following **preventive** surgeries?

☐ Subtotal colectomy — technically, yes. My surgeon and I decided that twice was enough and to PREVENT a third we should take out the colon.

☐ Total hysterectomy (removal of the uterus, cervix, fallopian tubes and ovaries)

☐ Hysterectomy (uterus only)

12. Have you implemented any of the following lifestyle changes or additions to reduce the possible risk of cancer (*check all that apply*)?

☐Exercise

☐ Diet (low in red meat)

☐ Diet (low in sugar intake besides sugars found in fruit)

☐Diet (increased fruit and vegetable intake)

☐Aspirin

If you take aspirin please specify the milligrams?

_____

13. What is your highest level of education that you completed?

☐ Grade 8

☐ Grade 12

☐ College program

☐ University Bachelors degree

☐ Master's degree

☐ Doctorate

Thank you very much for taking the time to fill out the survey!

## Appendix D: Interview Guide

### Interview Guide

Experiences in the uptake of genetic testing and organ surveillance for persons diagnosed with Lynch Syndrome

**Section 1: Preamble, introduction and explanation:**

*1. First the informed consent document will be given and explained to the participant*.
The purpose of my research is to find out what if there were any barriers or difficulties you have experienced in getting or being referred to be genetically tested for Lynch Syndrome and if you feel your screening procedures are scheduled and suggested with relative ease by a Doctor who you feel is knowledge about Lynch Syndrome. The interview should only take 60 minutes. I will be audio recording the interview and I appreciate your willingness to allow me to do so with your informed consent. The audio recording will be kept secure and confidential. Once the interview has been transcribed, and after the data has been analyzed, the audio recording will be erased. There are no right or wrong answers to the questions. Consider this interview as simply a conversation between us where you have the opportunity to tell me whether you think there are issues in getting genetically tested for Lynch Syndrome and in receiving adequate organ screenings.

**Section 2: Initial experience with the term 'Lynch Syndrome'**

***1***. *I am going to start with some general questions. Please tell me about how you first came to know about Lynch Syndrome?*

(*Prompt if need: this study is specifically interested if it was a clinician who suggested this possibility, or from public knowledge*).

**Section 3: Trajectory to Genetic Testing for Lynch Syndrome**

***2.*** *How did your referral or decision to attend genetic testing take place?*

***3.*** *How easy was it for you to initiate genetic testing or did you encounter any difficulties? (Prompt if needed: These difficulties could be in getting referred to have the test performed or personal, for example some people find it hard to get emotionally prepared to receive the results).*

***4.*** *Would you change anything in the process to receive genetic testing?*

**Question number 5, below is for the 'index patient' (person who had genetic testing following cancer diagnosis) and is responsible for communicating LS status to family members.**

***5***. *After receiving your test result how did you communicate this family members?*

***6.*** *Did you contact relatives that were not very close with but related by blood? How easy or difficult was that for you?*

***7.*** *What could be changed to increase participation in genetic testing for Lynch Syndrome?*

**Section 4: Organ Screening and care received for Lynch Syndrome**

***8.*** *Do you have a primary care provider (nurse practitioner or family physician)?*
IF YES PROCEED TO QUESTION **11**, IF NO PROCEED TO QUESTION **12**.

***9.*** *Does your primary care provider schedule your screening procedures for you based on their knowledge of Lynch Syndrome or do you have to ask for certain procedures?*

***10.*** *How knowledgeable about Lynch Syndrome does your primary care provider seem to be? Did they provide you refer you to a specialist? For females, was gynaecological cancer discussed and different screening possibilities or prophylactic surgery also discussed?*

***11.*** *What preventive colon cancer recommendations, if any, did your health care provider offer you? What was the occupation of that person? Did you research and preventive measures on your own and did you implement them? What were these preventive measures?* (Prompts: lifestyle changes, medications*)*

***12.*** *Where do you think you received the most information about Lynch Syndrome? (Prompts: genetic counselling, primary care provider, oncologist, gastroenterologist,* surgeon, other persons with Lynch syndrome, Internet, medical journals, conferences)

***13.*** *Before we finish, is there anything else you would like to add that you haven't already mentioned?*

**Section 5: Closing remarks:** Thank you for agreeing to be interviewed. Once the data are analyzed if you are interested the results will be sent to you so you can verify what ideas came out of this research.

## Appendix E: Excerpt of Reflexivity Journal and Process Notes

Example of excerpt from Principal Investigators Reflexivity Journal

Interview number 4:

The participant today had an appointment with their new specialist at the Health Sciences Centre (HSC). She wanted the interview to occur afterwards at the Faculty of Medicine attached to the Health Sciences Centre. I met her in the G.I. clinic at HSC and walked her to the conference room I booked for the interview. She seemed relieved to speak to me and told me about what happened in the consult appointment before the interview started.

I asked the questions as per the interview guide but added very little additional dialogue. The participant shared information easily. She mentioned the need for a personalized approach to LS organ surveillance based on family history and the need for persons undergoing a colonoscopy every year to have the easiest type of bowel preparation formula. She used the word 'horrible' multiple times to describe the 4 liter polyethylene glycol she had to drink. She now uses only the pico-salax for bowel preparation.

After I had stopped the audio-recording the participant shared with me that she often does not have opportunity to speak about her concerns regarding Lynch Syndrome and the impact cancer has had on her family. She also mentioned that the Lynch Syndrome diagnosis is not something she shares outside her family because of the genetic component and she worries about how this knowledge would impact her other family members.

How did my background influence the interview and interpretation?

I listened and responded with genuine empathy as the participant shared how many family members passed away from Lynch Syndrome cancers before the province implemented testing. My background as a qualitative health researcher on project with older adults provided me the understanding that it is often not one person involved in care when a family member experiences illness but rather the whole family is often involved and impacted in some way. This led me to postulate an idea of family and communication being vital. It is my familiarity of previously interviewing caregivers and being a caregiver myself that influenced my investigation of the role of family.

How did the participant's narrative change me?

The connection I have with a participant during the interview is like a thread that is woven into tapestry, it is always there but becomes part of the greater narrative, providing the strength for the tapestry. This experience made me reflect upon the highly guarded aspect of a genetic condition that the participant shared. It made me value the need to remove the possibility of participation identification through the quotes. It led me to think about how communicating the genetic condition occurs in the family. Something I have not personally experienced but can imagine can be difficult. This narrative has led to think heavily on the role of communication and families.

## Appendix F: Ethics Approval and Renewal Certificate

UNIVERSITY OF MANITOBA | Research Ethics and Compliance

Human Ethics
208-194 Dafoe Road
Winnipeg, MB
Canada R3T 2N2
Phone +204-474-7122
Email: humanethics@umanitoba.ca

**PROTOCOL APPROVAL**

**TO:** **Kathleen Zawaly** **(Advisor: Nancy Hansen)**
Principal Investigator

**FROM:** **Kevin Russell, Chair**
Joint-Faculty Research Ethics Board (JFREB)

**Re:** **Protocol J2017:093 (HS21063)**
**"Experiences of genetic testing and organ surveillance for persons diagnosed with Lynch Syndrome"**

---

**Effective:** October 13, 2017 **Expiry:** October 13, 2018

**Joint-Faculty Research Ethics Board (JFREB)** has reviewed and approved the above research. JFREB is constituted and operates in accordance with the current *Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans*.

This approval is subject to the following conditions:

1. Approval is granted only for the research and purposes described in the application.
2. Any modification to the research must be submitted to JFREB for approval before implementation.
3. Any deviations to the research or adverse events must be submitted to JFREB as soon as possible.
4. This approval is valid for one year only and a Renewal Request must be submitted and approved by the above expiry date.
5. A Study Closure form must be submitted to JFREB when the research is complete or terminated.
6. The University of Manitoba may request to review research documentation from this project to demonstrate compliance with this approved protocol and the University of Manitoba *Ethics of Research Involving Humans*.

**Funded Protocols:**
- **Please mail/e-mail a copy of this Approval, identifying the related UM Project Number, to the Research Grants Officer in ORS.**

UNIVERSITY OF MANITOBA | Research Ethics and Compliance

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**RENEWAL APPROVAL**

**Date: September 25, 2018** **New Expiry: October 13, 2019**

**TO:** **Kathleen Zawaly** **(Advisor: Nancy Hansen)**
Principal Investigator

**FROM:** **Julia Witt, Chair**
Joint-Faculty Research Ethics Board (JFREB)

**Re:** **Protocol #J2017:093 (HS21063)**
**"Experiences of Genetic Testing and Organ Surveillance for Persons Diagnosed with Lynch Syndrome"**

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**Joint-Faculty Research Ethics Board (JFREB)** has reviewed and renewed the above research. JFREB is constituted and operates in accordance with the current *Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans*.

This approval is subject to the following conditions:

1. Any modification to the research must be submitted to JFREB for approval before implementation.
2. Any deviations to the research or adverse events must be submitted to JFREB as soon as possible.
3. This renewal is valid for one year only and a Renewal Request must be submitted and approved by the above expiry date.
4. A Study Closure form must be submitted to JFREB when the research is complete or terminated.

**Funded Protocols:**

- **Please mail/e-mail a copy of this Renewal Approval, identifying the related UM Project Number, to the Research Grants Officer in ORS.**

## Appendix G: Informed Consent Forms

UNIVERSITY OF MANITOBA

### PARTICIPANT INFORMATION AND CONSENT FORM

**STUDY TITLE: Experiences of genetic testing and organ surveillance for persons diagnosed with Lynch Syndrome**

**Ethics Protocol:**
J2017:093 (HS21063)

**Principal Investigator:** Ms. Kathleen Zawaly, B.Sc. (McGill), M.Sc. (Student, University of Manitoba). This research is for Kathleen's M.Sc. thesis.
**Advisor:** Dr. Nancy Hansen, Ph.D.
Email: nancy.hansen@umanitoba.ca
Phone number for advisor: 204. 474.6458

**You are being invited to participate in a research study. Please take your time to review this consent form and ask any questions you may have. This consent form may contain words that you do not understand. Please ask Kathleen Zawaly any questions you may have. She can explain any words that are not familiar. This research has been approved by the Joint-ethics board at the University of Manitoba and you may contact the Human Ethics Coordinator at 204. 474.7122 or via email at humanethics@umanitoba.ca if you have any concerns.**

**Purpose of Study:**
The key purpose of this study is to provide information regarding the barriers within the health care system that persons diagnosed with Lynch Syndrome encounter regarding organ surveillance and access to genetic testing for themselves and family members.

**Study Procedures: The interview**
**If you agree to participate in the study, Kathleen Zawaly will contact you to set up an interview time. I expect the interview to take approximately one hour but it may take a little longer. I will have a series of questions to ask about:**

1. How you were referred for genetic testing (following tumor testing, or did a family member communicate with you).
2. Your experience of any difficulties in being refereed for genetic testing?
3. Your experience in communicating or not communicating a positive test result to family members
4. Who coordinates your organ screening and what is their knowledge about Lynch Syndrome?

In the interview you will be free to discuss any aspect of your life, lynch syndrome and your health. You can decide how much you would like to say in response to the questions and you may decline to answer any of the questions if you wish.

The interview will be audio recorded and later typed word for word in a written document. At that time your name and any identifying information will be removed and replaced by a code number. The audio recording will be erased once the written document has been verified by my advisor.

**Follow-up:** Once interviews from most, or all, of the people in the study have been completed, I will mail or email a draft summary of the findings to you. I will ask you to read the summary and tell us if you feel your views have been accurately reflected. The anticipated date of disseminating preliminary findings will be in the summer of 2018. The results would be published as part of my master's thesis.

**Risks or Discomforts**
There should be no risks or discomforts associated with this study other than sitting in a room for at least an hour.

**Benefits**
By participating in this study, you may gain some benefit by being able to share your experience of having Lynch Syndrome. There may or may not be direct benefits to you from participating in this study.

**Counseling Resources**
If you think you may benefit from access to free counseling resources after the interview, the following is one option that is available. Klinic at 870 Portage Ave, for drop-in counseling, please call 204.784.4067 for hours or to make an appointment.

**Confidentiality**
Your name and contact information will be stored on a password protected computer that only the primary investigator will have access to. When the results of the study are published, your identity will remain confidential. Your confidentiality will be protected according to the law and in accordance with the Personal Health Information Act of Manitoba (PHIA).

Despite efforts to keep your personal information confidential, absolute confidentiality cannot be guaranteed. Your personal information may be disclosed if required by law. The University of Manitoba Research Ethics Board may review research records for quality assurance purposes.

**Further Information**
If you would like any further information please contact the primary investigator Kathleen Zawaly at _____.

**Voluntary Participation/Withdrawal from the Study**
Your decision to take part in this study is voluntary. You may refuse to participate or you may withdraw from the study within 60 days of participating. If you provided informed consent to be

audio-recorded you can contact Kathleen Zawaly, if you wish to withdraw within 60 days and have the information you provided deleted. You understand that information regarding your personal identity will be kept confidential, but that confidentiality is not guaranteed. The University of Manitoba Research Ethics Board may look at the data collected, for quality assurance purposes.

By signing this consent form, you have not waived any of the legal rights that you have as a participant in a research study. You have now agreed to be audio-recorded for the interview.

| **Consent Form** | **Yes** | **No** |
|---|---|---|
| I agree to participate in this study | ☐ | ☐ |
| I agree to be contacted for follow-up related to this study | ☐ | ☐ |

| _____ | _____ | _____ |
|---|---|---|
| Name of Participant (printed name) | Signature of Participant | Date |

| _____ | _____ | _____ |
|---|---|---|
| Kathleen Zawaly (P.I.) (printed name) | Signature of Person Obtaining Consent (P.I.) | Date |

If you have agreed to be contacted for follow-up (summary of findings for member checking) from this study, how would you like to receive and check the results from this study?

Email: _____

Or mailing address:

_____

_____

## Appendix H: Medical Terms

**Adenoma:** a benign tumour in the epithelia tissue of a gland or gland-like region in the body. Adenomas found in the colon are a common type. There is a possibility that adenomas can become malignant.

**Antrum:** in the stomach the antrum is the portion before the outlet to the small intestines.

**Autosomal dominant inheritance:** a person with an autosomal dominant disorder has a 50 percent chance of having a child affected the mutated gene and a 50 percent chance of having a child with two nonmutated genes.

**Bowel preparation regimen:** consists of a solution that is consumed orally to flush the colon and may be combined with an adjunct such as a stimulate laxative to further the cleansing of the colon.

**Chemoprevention:** is the use of medication, vitamins, herbs or supplements to prevent cancer.

**Colonoscopy:** is a procedure where a long flexible tube with a video camera on the end is inserted through the rectum to view the mucosa of the colon.

**Endometrial biopsies/sampling:** a tube is inserted into the vagina through the cervix into the uterus and through the use of suction a sample of the lining of the uterus will be taken.

**Esophagogastroduodenoscopy (EGD):** is a procedure where a tube with a video camera on is inserted into the mouth and advanced into the stomach.

**Germline mutation:** is also called a hereditary mutation. This is a gene change in the body's reproductive cell (sperm or egg) that becomes incorporated into the DNA of every cell in the body of the offspring.

**Immunohistochemistry**: is a lab test often performed on tissue to test for certain tumour markers. Certain tumour markers can be linked to a particular enzyme or a fluorescent dye that is activated when it comes into contact with the tumour marker. The enzyme or dye then can be seen under a microscope and can be used to identify the mutations within the tumour.

**Hereditary nonpolyposis colorectal cancer (HNPCC):** is a term that was used to previously to describe Lynch Syndrome. This term was used when the research on Lynch Syndrome focused on patterns of colorectal cancer.

***H. pylori* infection:** Helicobacter pylori is a bacterium that can cause a stomach ulcer which can possibly lead to gastric cancer. *H. pylori* bacteria may be passed from person to person through

direct contact with feces, saliva, or vomit. *H.pylori* may also passed on through contaminated water or food.

**Keratoacanthorna:** are an aggressive skin cancer that may be invasive and lead to metastasis.

**Lynch Syndrome (LS):** is an inherited disorder that increases a person's risk of multiple types of cancer.

**Microsatellite instability (MSI)**: is the condition where there is predisposition for a mutation to form from DNA not repairing itself in an ideal way (impaired DNA mismatch repair (MMR)).

**Polymerase chain reaction (PCR):** is a technique used in the field of genetics that allows for the analysis of DNA samples containing small amounts of DNA. This analysis can identify MSI in the DNA.

**Muir-Torre Syndrome:** is a form of Lynch Syndrome, that is characterized with specific skin cancers in association with internal cancers.

**Proband or index patient:** is the first person in a family that identifies as having a genetic disorder to the clinician(s) investigating the hereditary condition.

**Prophylactic hysterectomy and bilateral salpingo-oophorectomy:** is a risk reducing surgery where the uterus and ovaries are removed to prevent the possibility of cancer.

**Polypectomy:** is the removal of a polyp in the colon and can often be performed during a colonoscopy.

**Sebaceous carcinoma:** are skin cancers that can spread rapidly.

**Subtotal colectom**y: is the surgical resection of part of the colon, often performed to reduce the incidence of future cancers.

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