Hereditary Nonpolyposis Colorectal Cancer: Factors Contributing to Adherence and Non-adherence to Surveillance for Mutation Carriers in Rural Areas of the Northern and Western Cape

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DECLARATION

I, Brenda Julie Kruger hereby declare that the work on which this research project is based is my original work (except where acknowledgements indicate otherwise) and that neither the whole work nor any part of it has been, is being, or is to be submitted for another degree in this or any other university.

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ABSTRACT

Hereditary nonpolyposis colorectal cancer (HNPCC) is the most common hereditary form of colorectal cancer (CRC) accounting for 20-35% of such cancers and 1-7% of all CRCs. Once a disease-causing mutation in one of the mismatch repair (MMR) genes has been identified in an affected individual, predictive genetic testing can be offered to first-degree relatives. This service has been available at the Division of Human Genetics, University of Cape Town (UCT), since 1994. Those testing positive are at increased risk of developing CRC and thus advised to attend frequent CRC surveillance (colonoscopies or flexible sigmoidoscopies). Limited literature is available internationally and is non-existent in South Africa on adherence to such screening recommendations. The aim of this study was to explore possible factors that may affect non-adherence and adherence to surveillance guidelines for mutation positive individuals who are at high risk of developing CRC in the areas of the Northern Cape of South Africa.

The study took place in rural, impoverished areas of the Northern Cape of South Africa and used a qualitative, exploratory research design to prospectively study the participants. Purposive sampling was used to select participants with the intention of gaining a deeper understanding of the research topic. Although a total of 17 individuals (6 adhering and eleven non-adhering) provided verbal consent to participate in semi-structured interviews, only 8 individuals participated in the study. Three individuals (1 adhering and 2 non-adhering) agreed to take part in a follow-up group discussion 3 months later. The data obtained from the interviews, participant observations and the follow-up group discussion was analysed using content analysis.

A total of 8 individuals (5 adhering and 3 non-adhering) participated in the study. The findings indicate that barriers to adherence include the preparation, the discomfort during the colonoscopy procedure, a family member's painful experience during a colonoscopy, not realising they were at increased risk of developing CRC and being unaware that they had received their genetic test result. While adherent participants attended regular surveillance to maintain their health and know their cancer status, non-adherent participants were unaware or misunderstood the reason for regular screening.

The findings from this research project provide a framework for a more comprehensive investigation into the non-adherence of mutation carriers. Although absolute adherence to surveillance guidelines is unrealistic, there is a potential for genetic counselling to help motivate, educate and support those not attending screening to maximise adherence.

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LIST OF ABBREVIATIONS

CAPP	Colon Cancer Prevention Project
CRC	Colorectal Cancer
BRCA	Breast cancer gene
DNA	Deoxyribonucleic acid
FAP	Familial Adenomatous Polyposis
GSH	Groote Schuur Hospital
HBM	Health Belief Model
HD	Huntington disease
hMLH1	human MutL homologue 1
hMSH2	human MutS homologue 2
hMSH6	human MutS homologue 6
hPMS1	human postmeiotic segregation 1
hPMS2	human postmeiotic segregation 2
HNPCC	Hereditary Nonpolyposis Colorectal Cancer
ICG-HNPCC	International Collaborative Group on HNPCC
IHC	Immunohistochemistry
InSiGHT	International Society for Gastrointestinal Hereditary Tumours
IR	Ileorectal Anastomosis
LCPG	Leeds Castle Polyposis Group
MMR	Mismatch Repair
MSI	Microsatellite Instability
MSI-H	MSI-High
MSI-L	MSI-Low
MSS	Microsatellite Stable
RN	Registered Nurse
TAC	Total Abdominal Colectomy
UCT	University of Cape Town

GLOSSARY

The terms in the glossary are adapted from Nussbaum et al. (2001).

Autosomal dominant inheritance: The expression of a trait in the heterozygous state, which is located on an autosome.

Colonoscopy: An examination in which the doctor looks at the colon through a flexible, lighted instrument called a colonoscope.

DNA: The primary carrier of genetic information. It is a macromolecule usually consisting of a polynucleotide chain, phosphate and deoxyribose sugar.

Exon: The DNA base sequences of a gene that encode amino acids. Exons are interspersed with non-coding regions called introns.

Gene: A sequence of DNA that codes for a particular protein.

Microsatellite: A small run of tandem repeats of nucleotides (usually less than 0.1kb) of a simple DNA sequence, usually 1-4 base pairs in length.

Mutation: Alterations in the DNA sequence or chromosome structure that damages the function of a gene and may cause disease.

Polyp: A mass of tissue that projects into the colon.

Proband: An individual through whom a family comes to the attention of an investigator (index case).

Prophylactic surgery: Surgery performed before a particular phenotype manifests itself in an individual.

Sigmoidoscopy: A procedure in which the doctor looks inside the rectum and the lower part of the colon (sigmoid colon) through a lighted tube.

Tumour: An abnormal mass of tissue that results from excessive cell division. They may either be benign (not cancerous) or malignant (cancerous).

PLAN OF THESIS

This research project is divided into five chapters and the layout is as follows:

Chapter one is the introduction and is divided into the following sections: an overview of HNPCC; a literature review on the factors affecting adherence to surveillance guidelines for HNPCC and a background to the HNPCC genetic service at UCT. It also provides a motivation for the study and the aims and objectives of the project.

Chapter two describes the methodology used in this research project.

Chapter three explains the results of the research project according to the themes obtained from the interviews, participant observations and follow-up group discussion.

Chapter four discusses the themes in the context of published literature.

Chapter five provides the summary and the conclusion of the study as well as recommendations for the genetic service and further research.

The referencing method used is in accordance with the American Journal of Human Genetics and internet references are referred to as "Href" within the text.

CHAPTER 1: INTRODUCTION

CHAPTER 1

INTRODUCTION

1.1. HEREDITARY NONPOLYPOSIS COLORECTAL CANCER – AN OVERVIEW

1.1.1. Introduction

Colorectal cancer (CRC) is the third most common cancer in females and the fifth in males in South Africa (Sitas et al. 1998). Most CRC's are sporadic (65-85%) with no evidence of people having inherited the disorder. In the remaining 15-35%, a potentially definable genetic component exists (Burt 1996). In the past decade, germline genetic mutations giving high lifetime risk of CRC in carriers have been found, accounting for 3-8% of all CRC cases. Inheritance of a single altered gene can result in a marked susceptibility to CRC in two distinct syndromes, familial adenomatous polyposis (FAP) and hereditary nonpolyposis colorectal cancer (HNPCC) (Jo and Chung 2005) (Figure 1). Both these hereditary CRCs are the result of a multistep process of carcinogenesis that typically develops over decades and appears to require at least seven genetic events for completion (Href 1). HNPCC is the most common hereditary form of CRC accounting for 20-35% of such cancers and 1-7% of all CRCs (Aaltonen et al. 1998; Samowitz et al. 2001; Müller and Fishel 2002; Yu et al. 2003; Hadley et al. 2004).

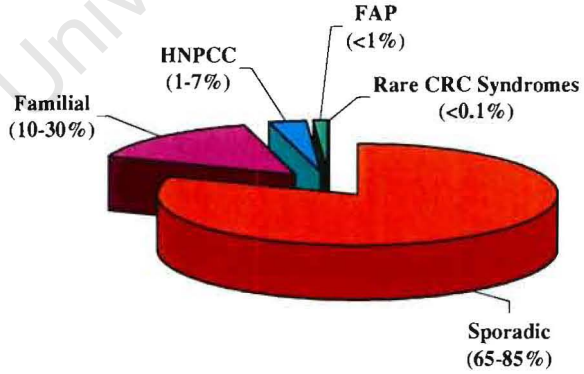

Figure 1. Causes of CRC, including sporadic, familial, HNPCC, FAP, and rare CRC syndromes (adapted from Burke et al. 1997).

1.1.2. Incidence

Differences exist in the incidence of colon cancer amongst the various population groups in South Africa. According to the National Cancer Registry of South Africa, the incidence for CRC in the indigenous African and Caucasian population group is 3 and 23 per 100 000, respectively (Sitas et al. 1998). The incidence of CRC in the Caucasian population in South Africa resembles those of Westernised countries (Oettle and Strijdom 1997). Gender related differences also exist; with the lifetime risk being 1 in 91 for males and 1 in 134 for females (Albrecht 2004). Despite considerable effort being made, the incidence of inherited CRCs, in particular HNPCC, in the South African population has not been ascertained as yet.

1.1.3. Clinical Features

The earliest reports on HNPCC include the first by Warthin (Warthin 1913) and later by Lynch who described the familial collection of colorectal, stomach and endometrial cancer with the name Cancer Family Syndrome (Lynch et al. 1966) but later it was termed Lynch Syndrome. Unlike FAP, where CRC is preceded by multiple polyps, the tumour in HNPCC usually arises from a single colorectal lesion. HNPCC may be subclassified into Lynch I and Lynch II based on the absence or presence of extracolonic cancers, respectively (Müller and Fishel 2002). The characteristic clinical traits are mentioned below in Table 1.

Table 1. The clinical features which typify HNPCC.

- CRC diagnosed at an average age of 42 years (Stanley et al. 2000).
- Autosomal dominant inheritance resulting from mutations in one of several DNA (deoxyribonucleic acid) mismatch repair (MMR) genes. The penetrance of mutations in *hMSH2* (human MutS homologue 2) gene and *hMLH1* (human MutL homologue 1) gene is incomplete and is significantly higher in males (about 80%) than in females (about 40-60%) by age 70 years (Mitchell et al. 2002).
- Improved survival stage-for-stage compared to those with sporadic CRC (Gryfe et al. 2000).
- An increased susceptibility to extracolonic cancers of the endometrium, ovary, stomach, small bowel, pancreas, hepatobiliary tract, brain, and upper uroepithelial tract (Stanely et al. 2000; Lynch and de la Chapelle 2003; Yu et al. 2003).
- Rapid adenoma to carcinoma progression (Yu et al. 2003).
- An increased proportion of colorectal adenomas are villous and dysplastic.
- Location, macroscopic and microscopic features of the cancer:
  - Predilection for the right side of the colon (proximal to the splenic flexure) (Lynch et al. 1993);
  - Development of multiple synchronous and metachronous CRCs (Vasen et al. 1999);
  - Increased proportion of tumours that are poorly differentiated, abundant in extracellular mucin, and distinguished by a lymphoid host response to the tumour (Vasen et al. 1999).

1.1.4. Diagnostic Criteria

The variable clinical characteristics seen in HNPCC patients and families emphasised the need for a specific definition of the disorder, which would assist more directed HNPCC research. In addition, there are no predictive physical signs of susceptibility to HNPCC. For these reasons, family history has been the major method for identification. The Amsterdam I Criteria were proposed by the International Collaborative Group on HNPCC (ICG-HNPCC) during an Amsterdam conference in 1990 to provide a standardised clinical definition for identification of families at risk for the disease (Vasen et al. 1991). The patient must meet all of the following criteria:

Amsterdam I Criteria

- At least three relatives with CRC
- One affected case is a first-degree relative of the other two
- Two or more successive generations affected
- One or more affected relatives received CRC diagnosis before 50 years of age
- FAP excluded
- Tumours verified by pathological examination

However, the original criteria failed to take family size or extracolonic cancers into account. To resolve this problem, new clinical criteria were introduced, known as the Amsterdam II Criteria. The revised criteria changed the first requirement to: "At least 3 relatives with an HNPCC-associated cancer (i.e. cancer of the colon and rectum, endometrium, small bowel, ureter, or renal pelvis)" (Vasen et al. 1999).

In 1996, the Bethesda Criteria were proposed by the National Cancer Institute workshop (Rodriguez-Bigas et al. 1997*a*). These criteria allow for the inclusion of smaller families and individuals with cancer in organs besides the colon and rectum, thereby maximising sensitivity but necessarily reducing specificity (Hampel and Peltomaki 2000). The Bethesda Criteria were updated and simplified in 2004 and state that the patient may meet any one of the following criteria (Umar et al. 2004):

Modified Bethesda Criteria

- CRC before 50 years
- Synchronous and metachronous CRC or associated HNPCC-related cancer* regardless of age

- CRC with one or more first-degree relatives with CRC or HNPCC-related cancer*; one of the cancers diagnosed before 50 years
- Individuals with CRC or endometrial cancer diagnosed at age < 45 years
- CRC with microsatellite instability-High (MSI-H) morphology (characterised by the presence of tumour infiltrating lymphocytes, mucinous differentiation or signet ring cell carcinoma, peritumoral Crohn's-like lymphocytic reaction, medullary growth pattern) before 60 years
- CRC with two or more relatives with CRC or other HNPCC-related cancer* regardless of age

(*Includes: endometrial, ovarian, gastric, small intestine, urinary tract, biliary tract, pancreas, brain, and sebaceous gland)

1.1.5. Genetic Defect

In 1993 linkage was shown between HNPCC and a locus on chromosome 2 (Peltomaki et al. 1993). Two large kindreds with HNPCC were investigated and the disease gene was localised to chromosome 2p15-16. This gene was later fine mapped to chromosome 2p22-p21 and identified as *hMSH2* (Fishel et al. 1993; Leach et al. 1993). Since the disorder did not map to this gene in some families, it became evident that *hMSH2* was not the only gene involved in disease pathogenesis and that heterogeneity existed for HNPCC. The defective genes causing HNPCC in subsequent subsets of families with HNPCC have been found to include: *hMLH1* on chromosome 3p21-23 (Bronner et al. 1994); *hPMS1* (human postmeiotic segregation 1) gene on chromosome 2q31; *hPMS2* (human postmeiotic segregation 2) gene on chromosome 7q11 (Nicolaides et al. 1994); and *hMSH6* (human MutS homologue 6) gene on chromosome 2p16 (Baba 1997).

The above-mentioned genes function as MMR genes to maintain the fidelity of DNA in replication. The MMR genes encode for a family of proteins that recognise, excise and correct mismatches that occur during DNA replication. Germline mutations in one allele of one of these genes, followed by the somatic loss or inactivation of the second allele leads to a defective MMR mechanism. This process results in an accumulation of mistakes in DNA replication, an increase in mutations, and an acceleration of oncogenesis (Yu et al. 2003). Mutation rates in tumour cells with a deficient MMR system are 100 to 1000 times higher when compared with normal cells (Lynch et al. 1997).

Germline mutations of *hMSH2* and *hMLH1* account for more than 90% of HNPCC cases (Lynch and de la Chapelle 2003; Jo and Chung 2005). There is significant clinical variation between the

two genotypes, with carriers of *hMSH2* mutations showing a considerably higher chance of extracolonic cancers than those with an *hMLH1* gene defect (Lin et al. 1998).

Recent studies show that there might be other genes that have a role in the development of HNPCC. A study by Yamamoto et al. (1998) found that 32% of the families fulfilling the Amsterdam Criteria had no detectable MSI, which led them to conclude that other genes other than those involved with MMR may play a role in the tumourogenesis of HNPCC. These target genes include receptors for growth factors (transforming growth factors-β receptor II, insulin-like growth factor II receptor), cell cycle regulators (*E2F4*), regulators of apoptosis (*BAX*), and some of the MMR genes (*hMSH3* and *hMSH6*) (Yagi et al. 1998; Jo and Chung 2005).

1.1.6. Management

1.1.6.1. Clinical surveillance

More frequent colonoscopic surveillance is required in HNPCC than that received by the general population, since there is an earlier age of onset of CRC (average age of 42 years), a predominance of right-sided CRC, and an accelerated carcinogenesis (Yu et al. 2003). Screening for CRC in HNPCC is aimed at finding and removing adenomatous polyps as well as detecting early-stage cancer. The ICG-HNPCC proposed a set of guidelines for the clinical surveillance of HNPCC individuals (Weber 1996). The most recent guidelines for people with a genetic or clinical diagnosis of HNPCC or who are at increased risk for HNPCC are: colonoscopic screening initiated at age 20-25 years, or 10 years earlier than the youngest age a family member was diagnosed with HNPCC, whichever is earlier. It should be repeated every 1-2 years until 30 years of age and every year thereafter (Touwbridge and Burt 2002; Winawer et al. 2003).

In 2003, the International Society for Gastrointestinal Hereditary Tumours (InSiGHT) was founded to combine members of the ICG-HNPCC and the Leeds Castle Polyposis Group (LCPG). The role of this international society is to expand knowledge of hereditary CRC and improve care for patients and families affected by it (Lynch et al. 2004).

Screening for HNPCC through colonoscopy and removal of polyps is an effective approach for preventing CRC. A prospective 15-year screening study reported that colonoscopy repeated every three years can reduce the risk for CRC by 56-62%, prevent CRC deaths and decrease overall mortality by approximately 65% in HNPCC families (Jarvinen et al. 2000).

There is an increased risk of extracolonic cancers associated with HNPCC. The collective incidence of extracolonic cancers was determined in HNPCC gene carriers up to age 70 years in the Finnish Cancer Registry, and was as follows: colorectal (82%); endometrium (40-60%);

stomach (13%); ovary (12%); bladder, urethra and ureter (4%); brain (3.7%); kidney (3.3%); and biliary tract and gall bladder (2%) (Aarnio et al. 1999; Lu 2005). Since the endometrium and ovaries are the sites of the most common extracolonic cancers, screening is suggested in patients with and at-risk of developing HNPCC.

1.1.6.2. Treatment

The recommended treatment for an affected HNPCC patient is total abdominal colectomy (TAC) and ileorectal anastomosis (IR) in order to prevent metachronous colon cancer (Rodriguez-Bigas 1996). A subtotal colectomy with IR is suggested in HNPCC-associated mutation carriers with colon cancer. In addition, this procedure may also be considered prophylactically for selected HNPCC-associated mutation carriers who have adenomas in the colon at the time of screening (Burke et al. 1997). A proctocolectomy should be offered to individuals with rectal cancer and is recommended to mutation carriers who present with adenomas in the rectum during examination. Patients who have had surgery are recommended to continue regular surveillance due to the lifetime risk of rectal cancer and extracolonic cancers. According to a study by the ICG-HNPCC, the risk of developing rectal cancer was 12% at 12 years post abdominal colectomy (Rodriguez-Bigas et al. 1997*b*).

Chemotherapy is not usually recommended as a treatment for HNPCC since MMR-deficient cells are tolerant to being killed by alkylating agents (Branch et al. 1995).

1.1.7. Genetic Testing

Commercial genetic testing is presently available for *hMLH1* and *hMSH2*. The algorithm suggested by Jo and Chung (2005) for the molecular diagnosis of HNPCC is shown in Figure 2. If the diagnosis is based on the Amsterdam I Criteria, then one can advance directly to germline mutation testing. About 70% of families who fulfil the Amsterdam Criteria and undergo genetic testing will test positive for an HNPCC genotype (Schoen 2000).

If HNPCC is suspected by patients meeting any of the modified Bethesda Criteria, MSI testing and/or immunohistochemistry (IHC) on the colon cancer tissue from an affected individual should be performed first (Umar et al. 2004). MSI is found in 90% of colorectal tumours from HNPCC patients, but only about 15% of sporadic colorectal tumours (Boland et al. 1998; Kouraklis and Misiakos 2005). MSI is a marker of genetic instability, which may indicate the presence of a defective MMR gene and thus patients who test positive for MSI should then have further testing by mutation analysis. Colorectal tumours have been classified into three groups according to the extent of MSI:

- MSI-H: tumours with high frequency MSI, more than 30% unstable loci

- MSI-Low (MSI-L): tumours with low frequency MSI, 1-30% unstable loci
- Microsatellite stable (MSS): Tumours with 0% unstable loci (Hussein and Wood 2002).

Germline mutation testing in *hMLH1* and *hMSH2* is indicated for tumours showing MSI-H, but not for tumours exhibiting MSI-L and MSS.

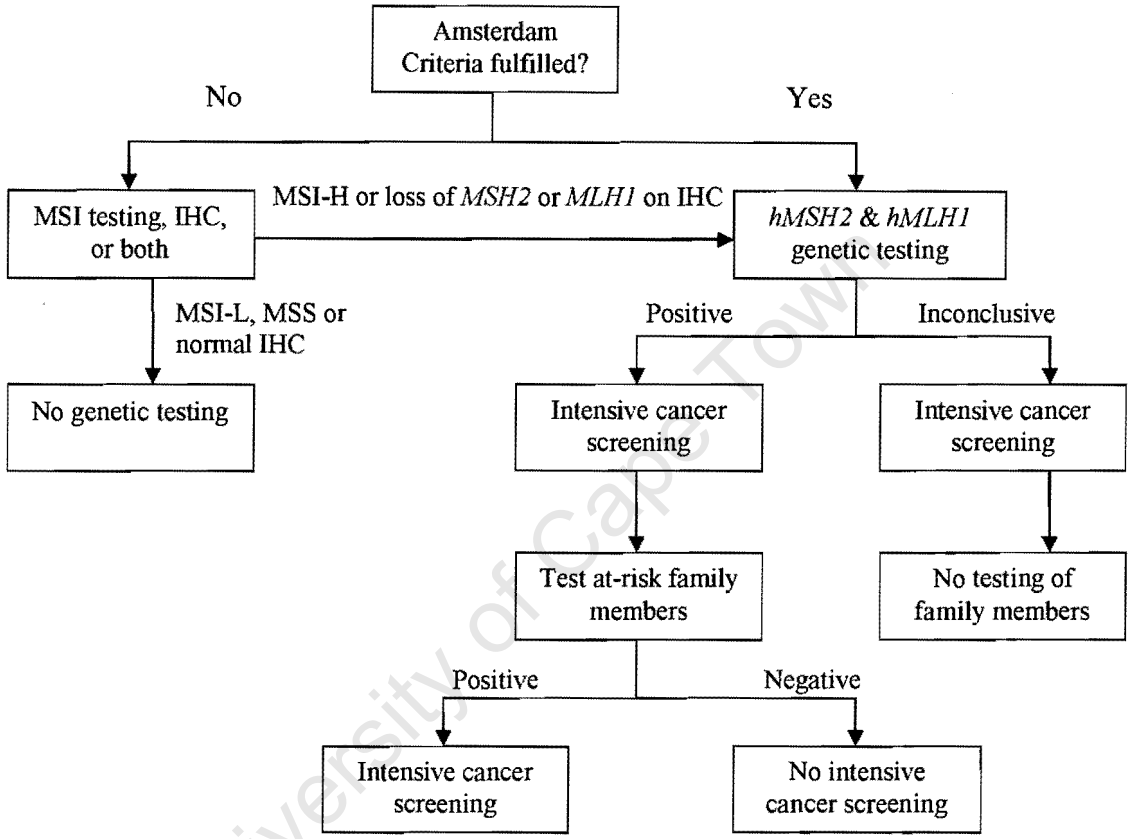

Figure 2. Algorithm suggested for genetic testing of an affected proband from a suspected HNPCC family (Jo and Chung 2005).

An alternative to MSI testing is IHC of tumour DNA, which can provide diagnostic information by specifying which MMR gene is most likely to be mutated. This information can then be used to determine if further testing should focus on *hMSH2* or *hMLH1* (Yu et al. 2003).

Once a germline mutation in *hMLH1* and *hMSH2* has been identified, predictive genetic testing can be offered to other family members. This allows differentiation of family members who carry the mutation and have definite increased risk from non-carriers who have the population risk of 5-6% (Lynch and de la Chapelle 2003; Stanley et al. 2000). This, in turn, allows mutation negative individuals to be released from surveillance, while those with the disease-causing mutation are advised to follow regular CRC screening as described in section 1.1.6.

1.1.8. Predictive Genetic Testing

"The main purpose of offering predictive genetic testing for hereditary cancer is to reduce unnecessary worry among those with a low risk of cancer (mutation negative) and to recognise those with a high risk (mutation positive), so as to promote preventative measures" (Aktan-Collan et al. 2001). Predictive testing allows for the early start of therapeutic or prophylactic regimes if any exist for the given disorder, and for life decisions to be informed in a way that would not otherwise be possible (Marteau and Richards 1996).

Predictive testing for late onset genetic disorders became possible initially for Huntington disease (HD) in the early 1980s (Harper 1991). The experience gained from testing and setting up guidelines for HD testing has since been used to establish predictive testing protocols for other genetic disorders, such as HNPCC. The predictive testing programme for HNPCC should include at least two pre-test educational sessions and several post-test counselling sessions, to minimise misunderstanding regarding the test result, combined with psychological support (Aktan-Collan et al. 2000).

The aim of the pre-test sessions is to educate the individual about the hereditary nature of HNPCC, the tests available, the cancer risks associated with a positive genetic test result, the medical aspects, and the recommended screening guidelines. This should give the individual the ability to make an informed, voluntary decision whether to undergo testing or not. Individuals should also be informed that they may withdraw from the protocol at any stage. A post-test session follows, in which the individual may elect to be informed of the result of the predictive genetic test. A follow-up session is advised for individuals who tested mutation positive to decide whether they require any additional support and to discuss early management options. The potential consequences of a predictive result are listed in Table 2.

Table 2. Potential consequences of predictive testing for HNPCC (adapted from Trimbath and Giardiello 2002)

If result is mutation positive

Positive consequences

- Removal of uncertainty
- Early detection of polyps and prevention of CRC
- Improved ability to plan the future, including family and career decisions
- Increased compliance with recommended surveillance
- Choice of surgical and medical management

Negative consequences

- Psychosocial distress, including anxiety, depression, anger, or denial
- Changes in family psychosocial dynamics
- Stigmatisation
- Increased fear about surgery or death
- Knowledge that children are at 50% risk of CRC
- Guilt or worry about children
- Genetic discrimination – insurance and employment
- Possible lifestyle changes after surgery

If result is mutation negative

Positive consequences

- Removal of uncertainty
- Children not at-risk
- Fewer medical exams and costs
- Better insurability

Negative consequences

- Survivor guilt
- Disturbance in family psychosocial dynamics

1.1.9. Genetic Counselling

According to Harper (2004) genetic counselling is: "the process by which patients or relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it and the ways in which this may be prevented, avoided or ameliorated." This process helps the individual or family to:

- Comprehend the medical facts, diagnosis, probable cause and available management
- Comprehend the genetic aspects of the disorder and its recurrence risks
- Understand the available options for dealing with the recurrence risk
- Come to a decision appropriate to them and to act in accordance with that decision to make the best possible adjustment to the disorder and/or the recurrence risk (Fraser 1974).

Hereditary cancers, such as HNPCC, require a specialised form of genetic counselling, taking the following into consideration:

- Patients have individual reactions and may be apprehensive about their and their family's risk of cancer.

- Since genetic counselling is medically oriented, the counsellor has both an interest in and an obligation to explore the disease status of the patient using available diagnostic, preventative and therapeutic procedures.
- The genetic counsellor needs to inform relatives at high risk of the natural history of the disease and their genetic risk in order to benefit from this information.
- The genetic counsellor should attempt to recognise and effectively manage the disease-related emotional content that may be present in the patient and their family.
- Accurate genetic information is provided to individuals who are emotionally ready for understanding its significance (Lynch and Lynch 1996).

In the past, genetic counselling in HNPCC was vague due to the lack of reliable predictive physical signs of genetic susceptibility to CRC. Diagnosis was based solely on the natural history and the pattern of cancer distribution within an extended HNPCC family. The risk of first-degree relatives of patients affected with HNPCC for developing CRC could be assessed at only approximately 50%. Thus, it was not known whether family members had inherited the deleterious gene and were at about 80-90% risk for developing CRC or whether they were mutation negative and therefore reverted to the general population risk of 5% for CRC. Fortunately the genetic risk assessment for HNPCC has changed since the discovery of the causative genes. This information is now used for highly definitive genetic risk assessment in families where the mutation has been found. Mutation carriers are recommended to follow vigorous surveillance and management guidelines, while mutation negative individuals would be released from the regular management and follow the recommendations for the general population (Lynch and Lynch 1996).

When there are living affected family members, genetic testing often begins with one or more of them in order to identify the specific disease-causing mutation. Family members may differ in their opinion of the desirability and value of testing and the potential implications of test results on family members and their relationships to one another. The rights of individual family members must be respected and supported with respect to confidentiality, privacy, and the right to not participate or not be informed of the test results (Weil 2000).

Once the HNPCC diagnosis has been established, the genetic counsellor's role is to facilitate information distribution within the family without causing additional anxiety and unnecessary concerns about cancer. It is important that genetic counselling reduces cancer-specific distress, since depressive symptoms may reduce genetic test acceptance (Lerman et al. 1999). Also, a high level of distress before predictive testing seems to be a key predictor of post-test anxiety, as

found in breast cancer gene (BRCA) testing (Lodder et al. 2001). By reducing pre-test distress, one can possibly reduce adverse affects after testing (Keller et al. 2002).

Another consideration is whether members of HNPCC families want to know their genetic cancer risk status. The anxiety of coping with uncertainty must be considered. It is questionable whether is it psychologically more advantageous for the individual to know whether they are a gene carrier or to remain unaware of their status (Keller et al. 2002).

Taking into account the fact that preventative options can substantially reduce the incidence of, and mortality from, CRC, another important role of genetic counselling for HNPCC is to improve the motivation to participate in, and adherence to, recommended screening procedures of high-risk individuals. While this decision remains the individual's, a counselling model based on a medical recommendation that also recognises the patient's role in the decision-making process may be the most appropriate (Weil 2000). Promoting the individual's sense of control of the situation and confidence in the effectiveness of early detection can reduce the perceived threat associated with HNPCC-related cancers (Keller et al. 2002).

Genetic counselling, with its developing benefit in cancer control, is an emerging resource that signifies a hopeful future for those at high risk for the onset of cancer and particularly HNPCC. However, several issues are yet to be resolved about the potential ethical, financial, legal, psychological, and social repercussions of genetic testing and also the real benefit for patients and their families (Bonadona et al. 2002). Genetic counsellors need to acknowledge the implications associated with confidentiality, stigmatisation, and psychosocial effects that often go with counselling and to be prepared to effectively manage these psychodynamic factors (Lynch and Lynch 1996).

Finally, it should be emphasised that the goal of genetic counselling is to "facilitate comprehensive consideration of medical, psychological, and social issues part of the testing process, inform the patient's decision-making, facilitate the adaptation process, and promote behaviour consistent with medical recommendations with respect for personal and cultural values and beliefs" (Hadley et al. 2004).

1.2. FACTORS AFFECTING ADHERENCE TO SURVEILLANCE GUIDELINES

Despite the demonstrated beneficial effects of CRC screening, adherence to screening recommendations is less than optimal. Compliance rates vary from about 50-80% in studies

including first-degree relatives of patients with CRC and from 63-93% in studies of HNPCC families (Bleiker et al. 2005).

Only a limited number of international studies have investigated the factors that play a major role in adherence and non-adherence with colon cancer screening advice for those at high risk for HNPCC (Table 3), and no studies have been carried out on this same issue in South Africa or Africa, to date.

A possible reason for the few articles published worldwide on this topic could be that predictive testing has only been available since 1994 to individuals in which a family-specific mutation has been found in a close relative.

Table 3. Factors shown in studies to affect adherence behaviour to recommended screening guidelines for HNPCC mutation carriers.

Adherence
• Perceived control over developing CRC (Halbert et al. 2004)
• Physician recommendation for screening (Hadley et al. 2004, Bleiker et al. 2005)
• Being sedated (Bleiker et al. 2005)
Non-adherence
• Embarrassment and discomfort of screening (Bleiker et al. 2005)
• Fear that a tumour would be detected during the screening (Bleiker et al. 2005)
• The absence of symptoms or other health problems (Stoffel et al. 2003)
• Low perceived risk of CRC (Lynch et al. 1999)
• Misunderstanding the predictive genetic test result connected with less worry about developing CRC (Aktan-Collan et al. 2001)
• Younger age (Hadley et al. 2004)
• Younger age associated with more discomfort (Liljegren et al. 2004)

1.3. BACKGROUND TO HEREDITARY NONPOLYPOSIS COLORECTAL CANCER GENETIC SERVICE AT THE UNIVERSITY OF CAPE TOWN

1.3.1. History of Genetic Testing

In 1991, formal recruitment of individuals diagnosed with CRC under 50 years of age for genetic research studies to identify disease-causing mutations began at the Division of Human Genetics, University of Cape Town (UCT). Since the mutations causing HNPCC are family-specific, research starts with an individual affected with colon cancer. Mutation screening of all the exons in the *hMLH1*, *hMSH2* and *hMSH6* genes is currently being offered to affected individuals. Once a family-specific mutation has been found, predictive genetic testing can then be offered to first-degree at-risk family members identified through genealogical assessment.

Since 1994, genetic counselling and predictive testing has been available at UCT for at-risk asymptomatic individuals over 18 years of age.

To date a total of 351 families (1285 individuals) had been recruited into the HNPCC registry. Most individuals from these families live in the Northern and Western Cape (Figure 3). Fifteen of these families are involved in predictive testing – in which 145 individuals are mutation positive and have received their result. Of these high-risk individuals, only 61% are adhering to the recommended surveillance programme while 39% are not since they received their positive mutation status (search of HNPCC database until 5 August 2005). There is concern for this sub-group of carriers who do not undergo cancer screening at the recommended levels, especially in light of the recognised benefits of regular screening.

Figure 3. Map of the Northern and Western Cape of South Africa, with the various coloured dots (red, white, yellow, pink and black) representing individuals in which a different HNPCC mutation has been identified.

1.3.2. Predictive Testing Protocol

Routine pre-test and post-test genetic counselling is offered to all individuals in the predictive testing programme at UCT. The initial pre-test meeting of an at-risk family member with a genetic counsellor focuses on information and implications of predictive testing in an atmosphere of support and education. During the post-test meeting with the genetic counsellor the at-risk individual receives their genetic testing information and has the opportunity to ask further questions while receiving the supportive counselling they require. Carriers of an HNPCC mutation are advised to adhere to a lifelong screening programme for early detection of colorectal polyps, while those testing negative are not (Bleiker et al. 2003). The HNPCC team of UCT manages colonic as well as extracolonic cancer surveillance using the management guidelines as outlined in section 1.1.6.

1.4. MOTIVATION FOR THIS STUDY

Since a preventative service like colonoscopic surveillance of mutation carriers is an expense, it is difficult to sustain in a resource-poor country like South Africa (Goldberg et al. 1998). Many of the individuals involved in the HNPCC project come from previously disadvantaged backgrounds and reside in rural under-resourced impoverished areas. To overcome this problem, a specialist team including gastroenterologists, surgeons, pathologists, geneticists and specialist colorectal registered nurses (RNs) travel to a medical facility in the area to offer colonoscopic surveillance to those at high risk. Despite it being a free service, some individuals do not attend these clinics and are consequently regarded as non-adherent to surveillance recommendations. For this reason, this study investigated a small group of mutation positive individuals living in rural impoverished areas in the Northern and Western Cape to explore the extent to which these and other factors affect compliance behaviours to surveillance guidelines.

This study focused on whether the following factors influence adherence and non-adherence to surveillance guidelines for mutation carriers:

- Low socioeconomic background
- Geographical isolation
- Understanding of HNPCC status
- Family history of HNPCC
- Experience of CRC screening
- Concerns about CRC screening
- Concerns about CRC

These factors were chosen on the basis of:

- Careful and thorough literature review of factors previously documented to influence compliance to surveillance guidelines for those at high risk for HNPCC.

- Perceived factors of influence according to expert opinions of Sr. Ursula Algar and Ms. Pat Craig, both of whom have had close contact with the HNPCC families and thus have knowledge regarding the potential factors of contribution to compliance behaviours. Sr. Algar is the clinical genetic RN for the familial CRC unit at Groote Schuur Hospital (GSH) and has been closely dealing with the HNPCC families since the predictive genetic service started in 1994. Ms. Craig was the genetic counsellor for the HNPCC genetic service at UCT from 2002 – 2005.

Hadley et al. (2004) proposed that careful and comprehensive consideration of the individual's personal and family situation and experience through the genetic counselling process influences to a certain extent subsequent adherence to recommended guidelines. For this reason, the knowledge gained from this research project and a future comprehensive investigation will be used to help improve genetic counselling services in maximising surveillance adherence in individuals who are at high risk of developing CRC in South Africa.

1.5. AIMS AND OBJECTIVES

The aim of this study was to identify factors associated with adherence and non-adherence to surveillance guidelines in a small group of mutation positive individuals in rural areas of the Northern and Western Cape of South Africa. The study objectives were as follows:

- To explore the sociodemographics of the study group.

- To hypothesise tentatively the factors associated with adherence and non-adherence to surveillance guidelines, through the identification of themes in the interview transcripts, participant observations and the follow-up group discussion.

- To use selected information gained from this pilot study to provide recommendations for a comprehensive investigation of mutation carriers' perception of factors affecting non-adherence to surveillance guidelines, which will either prove or disprove the hypothesis proposed by this pilot study.

- To ultimately help to improve the genetic counselling services in motivating, educating and supporting individuals who do not adhere to surveillance with the end goal of maximising surveillance adherence for those at high risk for CRC.

CHAPTER 2: METHODOLODY

CHAPTER 2

METHODOLOGY

2.1. INTRODUCTION

The main purpose of this research project was to provide a pilot investigation to explore and describe what influences the clinical screening compliance of individuals who had been informed of their positive mutation status and were thus at increased risk of developing CRC. There is currently no literature investigating this in the South African social and cultural context. The information gained from this research will be used to generate provisional hypotheses, which will then be verified by further research.

2.2. RESEARCH DESIGN

The present study used an exploratory, descriptive, cross-sectional, prospective study design to obtain information appropriate for the goals of the study.

Qualitative research develops understanding of human experiences, which is important for genetic counsellors who focus on communication and interaction. This method focuses on people within their social and cultural context in order to find out the meanings they give to their experiences and the way in which they interpret them (Holloway and Wheeler 1996). The result is an in-depth account of their emotional and experiential phenomena (Giacomini et al. 2000).

The objective of an exploratory study such as this is not to generalise to a large population, but to strive towards increased knowledge and insight in the specific area of investigation (Neuman 1999). Further research can be conducted at a later stage to investigate each finding more in-depth, utilising other levels and instruments of research.

The primary purpose of thick descriptive research is to acquire comprehensive and exact information about a particular observable fact, through examination, explanation and organisation, thus providing new information on that particular event or incident (Neuman 1999; Terre Blanche and Durrheim 1999). It presents an accurate picture of the specific details of a situation, social setting, or relationship with the purpose of discovering new meaning, describing what exists and categorising information (Neuman 1999).

The present study was a cross-sectional study in that it collected data at *one* point in time from the selected sample of participants, not at *several* points in time, as done in a longitudinal study (Grinnell 1988).

2.3. SETTING

The setting is the location where the research takes place (Holloway 1997). The research setting was limited to areas of Nababeep, Okiep, Port Nolloth and Komaggas in the Northern Cape, due to the limited time (6 months) available in which to complete the project. These remote areas are fairly close to Springbok, a town situated 550 km from Cape Town (Figure 4). Many of these towns developed in the mid 1800's to early 1900's for either copper or diamond mining. For this reason, most of the breadwinners in Komaggas and Nababeep work at operating mines in the region. Port Nolloth, being a small harbour town, has small-scale diamond recovery and crayfishing as the main sources of income of the residents (Href 2).

The interviews were conducted in either a private room in one of the local Primary Healthcare clinics in the area, or in the participants' homes. The problems with interviewing in the participants' home could be that privacy may be limited and that the physical surroundings may not be best-suited to interviewing. On the other hand, the participants might feel more relaxed, behave and respond more naturally and be less inhibited at home. In a home setting, the researcher had the opportunity to observe interactions of members of the family, the environment in which they live and the home circumstances that are likely to affect their lives, without the need for direct questioning (Mouton 2002). It is clear, however, that limited value should be placed on observations of interactions and/or behaviour during a single visit.

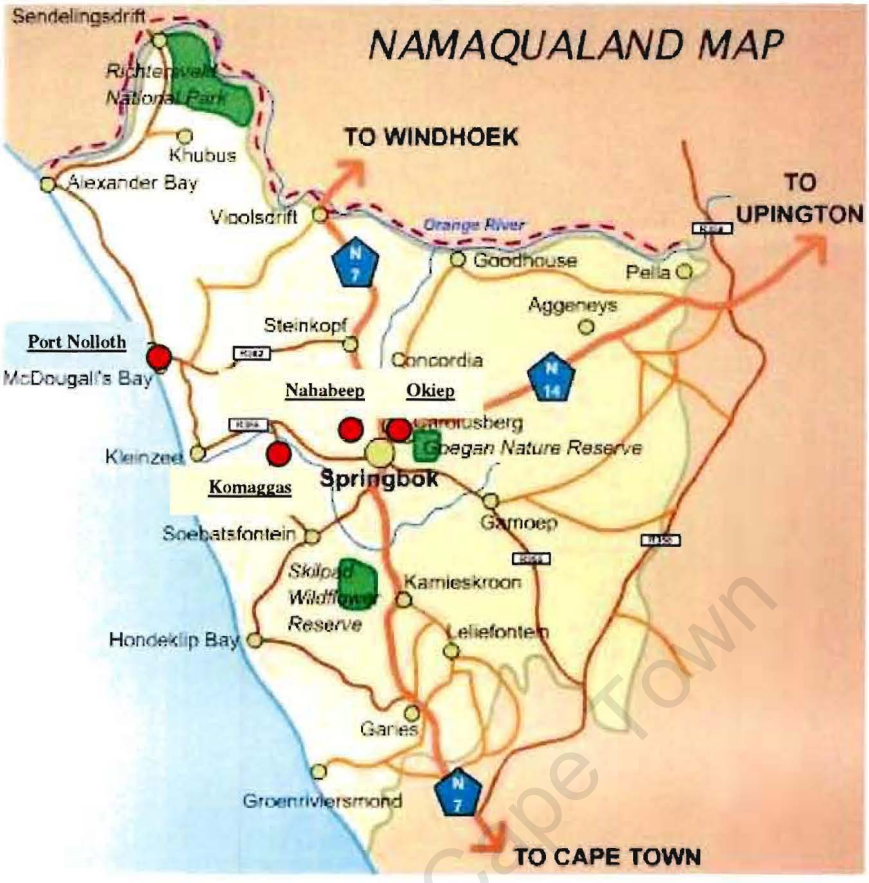

Figure 4. Map of the Northern Cape of South Africa showing the location of the towns where the interviews (Nababeep, Okiep, Port Nolloth and Komaggas) and follow-up group discussion (Port Nolloth) took place (represented by red dots) (Adapted from Href 3).

2.4. SAMPLE

Non-probability or non-random purposive sampling was used for the qualitative study. Non-probability sampling is often used for exploratory studies, where the main interest is in obtaining as much unique data on a research question as possible and not in representing or generalising to a larger population (Rossouw 2000). It is a useful method when each sampling unit (participant) represents a key position to observe or experience the phenomenon being investigated (Grinnell 1988).

Purposive sampling aims to select cases with a specific purpose in mind for in-depth investigation (Neuman 1999; Rossouw 2000). It is used less to generalise to a larger population than it is to gain a deeper understanding of types. The sampling units are selected according to previously specified criteria. Useful participants are those who have undergone

are undergoing experiences about which the researcher wants to gain information (Holloway and Wheeler 1996).

The interviews consisted of a heterogeneous sample. This type of sample contains individuals who differ from each other in a major aspect, in this study the participants were either adhering to surveillance guidelines or not. The participants were placed into one of two adherence categories, which were taken from the time they received their predictive genetic test result:

1. **Adherent** were those who underwent colon screening at the frequency advised.
2. **Non-adherent** were those who delayed being screened at least once for more than 1 year, those who underwent screening only once, or those who never underwent screening for a period of at least 5 years.

2.4.1. Inclusion and Exclusion Criteria

The participants were ascertained from the HNPCC Genetic Service database at UCT. The following criteria were necessary to be eligible for the study:

- Tested positive for a family-specific mutation in either *hMLH1*, *hMSH2* or *hMSH6* gene loci and have been through the predictive testing protocol at UCT
- Live in a geographically isolated part of the Northern Cape
- Come from a socioeconomically disadvantaged background
- Over 18 years of age

Socioeconomic disadvantage is defined as: "a relative lack of financial and material means experienced by a group in society which may limit their access to opportunities and resources that are available to the wider society (Href4)." The study group had a low education level, which may have resulted in their unemployment for a lack of opportunities and the adequate requirements. The resulting lack of or little income may have affected access to acceptable housing and healthcare and a reliance on public transport. They were consequently not able to offer their children adequate opportunities for schooling, thus perpetuating the cycle of disadvantage.

Exclusion Criterion:

- Did not remember receiving their predictive genetic test result

2.4.2. Selection Process

The researcher queried the HNPCC computer database from 1991 to 2004 to obtain the number of participants fulfilling the above-mentioned criteria for the pilot study and the study. Of the

145 individuals who tested mutation positive and had received their result, 40 individuals for the pilot study and 50 for the study were eligible. Ms. Pat Craig, who knew these individuals well, selected individuals from the group of 50 based on their willingness to participate in the study, their contactability and that they had undergone a colonoscopy in the last two years or not. Sr. Ursula Algar, Ms. Craig and Ms. Bonita Williams contacted these selected individuals telephonically and informed them of the purpose and nature of the study and obtained verbal consent from those willing to participate. Sr. Algar and Ms. Craig are known to the participants, unlike the researcher whom they had never met.

The researcher approached those eligible for the follow-up group discussion directly at the beginning of the interview on the first trip and telephonically a month later. All the participants were required to live in the same town for the follow-up group discussion to take place and Port Nolloth was the most convenient location for those who participated.

2.4.3. Total Number of Participants

Qualitative sampling generally consists of small sampling units studied in depth. Although there are no rigid rules for sample size in qualitative research, research text often mentions that 12-20 participants are needed for a heterogeneous sample (Holloway and Wheeler 1996). For this study, 17 participants (six adhering and eleven non-adhering) provided verbal consent for the interviews, but only eight (five adhering and three non-adhering) individuals took part.

The researcher ideally wanted the follow-up group discussion to consist of non-adhering participants. However, only two non-adhering participants could participate in it. For this reason one follow-up group discussion took place in Port Nolloth and consisted of two non-adhering and one adhering participant. The participants in this second part of the study were sisters and one of them was not involved in the interview stage of the study.

2.5. METHOD AND INSTRUMENTATION

2.5.1. Interviews and Participant Observations

A qualitative in-depth interview is a 'conversation with a purpose' in which the researcher aims to obtain the perspectives, feelings and perceptions of the participants (Holloway 1997). A semi-structured interview uses an interview guide to provide a focus on the in-depth issues to be covered (Holloway and Wheeler 1996). However, considerable freedom is given to the researcher to explore in their own way topics related to the research question being studied (Mouton 2002).

In this study, a semi-structured qualitative in-depth interview was used. It was designed by the researcher with the help of Sr. Algar, Ms. Craig and Ms. Vivian. The majority of questions were open-ended to encourage participants to engage with the researcher in open communication. Structured questions were used only to elicit sociodemographic data. Individual interviews were carried out since they tend to be more useful for evoking personal experiences and perspectives, particularly on sensitive topics such as losses due to CRC (Giacomini et al. 2000).

The researcher observed the participants in their natural settings during the interviews and follow-up group discussion and used this technique as an additional tool for data collection and analysis. "Observation is a technique of conducting research which involves the researcher simply observing what goes on in natural settings" (Hicks 1995). During this study the researcher recorded participant observations to help remember activities, events and the participants in the setting. They included raw data from observations and impressions about what the researcher found in the field as well as speculations, analytical comments and other thoughts (Holloway 1997).

The participant observations allowed the researcher time to observe and take notes of the environment, relationships and interactions between family members, and critical events that influenced the lives of the participants. Such events included the impact of deaths in the family, unemployment, poverty, disease burden to self and family members. The impression and feelings gained from these observations gave the researcher a more in-depth understanding of their lives and surroundings than would have been obtained from a self-reported survey.

The interview guide (Appendix D), consisting of 82 items, was constructed in such a way that there were nine sections: sociodemographic and socioeconomic information, understanding of CRC status, family history of CRC, colonoscopy experience, worries about colonoscopy, their opinion of the cancer support team, debriefing, opinion of the interview and an information session. Each section is discussed below.

To help the participant to feel more relaxed the interview progressed from non-threatening questions of sociodemographic and socioeconomic information to more sensitive questions.

A. Sociodemographic and Socioeconomic Information

The demographic details were recorded and measured to describe the cross-section of study subjects. The following factors were included:

a) Age and Sex

b) Marital status
c) Number and age of children
d) Education level and qualification
e) Employment status and income

Participants were also asked about the following factors:

a) Use, location, transport to day hospitals
b) Medical aid status
c) Cost, location, time and transport for colonoscopies

B. Understanding of HNPCC status

The participants' awareness and knowledge of their mutation status was assessed. The researcher explored how they felt after receiving a positive result, who they shared their result with, how their result had influenced their life and how their support networks and families had responded. The participants were asked to explain what it meant to their health to have the disease-causing mutation.

Since all participants in this study had been part of predictive testing and had tested positive for their family-specific mutation, they had been counselled about their result and the implications thereof. Thus all participants should answer "yes" to question no. 29 (Do you have the gene that causes CRC in your family?).

C. Family History of HNPCC

This query assessed how long participants had known about CRC in their families and their awareness of their own risk of developing CRC. It also determined the number, relationship, and loss of family members with CRC and how they had coped with these losses.

D. Colonoscopy experience

This section confirmed which participants had or had not been adhering to surveillance guidelines and explored participants' reasons for their behaviour as well as their experience of the colonoscopy procedure (from the preparation till after the colonoscopy itself). Their view on the appropriateness and the regularity of the procedure and the influence that family member's attendance had on them was explored. They were asked what their feelings were if a polyp or cancer was found during a colonoscopy. It was determined how they would cope with developing a cancer and how scared they were of this happening.

E. Concerns about colonoscopy

Section E requested details of the participants' worries, fears, dislikes, and suggestions to improve the surveillance procedure.

F. Opinions about HNPCC service

Participants were asked their opinion on how supportive, helpful and/or unhelpful the HNPCC team had been, as well as what would help them to manage their CRC risk better.

G. Debriefing

The researcher allowed the participants to share their feelings and ask questions throughout the interview, but specifically during the debriefing section. Participants were offered the opportunity to see the genetic counsellor of the HNPCC team if they needed counselling. Since participants in this study live in remote areas of the Northern Cape, where there is limited access to genetic counsellors, telephonic counselling with the HNPCC genetic counsellor was offered in the first instance. Face-to-face counselling was offered to all participants when the genetic counsellor was next in the area.

H. Opinion of Interview

The participants were asked their opinion of the interview and whether they would like a copy of the completed research findings.

I. Information Session

Finally, the researcher provided an information session using visual aids on: the anatomical location of structures of the digestive system; the genetic basis of CRC; the benefit of going for regular surveillance; and a description of the colonoscopy procedure.

2.5.2. Pilot Study

A pilot study is a small-scale trial run of the research with a very small number of participants chosen by the same criteria as those in the research. Pilot studies are not concerned with the participants' answers but rather with the difficulties experienced in answering the questions. Although pilot studies are unusual in qualitative enquiry, novice researchers should use it to gain confidence especially when using the interview technique (Holloway and Wheeler 1996; Holloway 1997).

A pilot study was conducted to test the constructed qualitative interview guide to determine if the items were understandable and accessible to participants, who were Afrikaans-speaking and of low socioeconomic background. The pilot study also ensured questions were clear and

unambiguous and approximated how much time would be needed to complete the interview (Grinnell 1988). The pilot study also helped the translator and the researcher to establish a close working relationship during the interviews in terms of when and how to translate for the researcher and the participants.

Due to time and budget constraints, the researcher was not able to conduct the pilot study in the Northern Cape and it was thus carried out on three participants (two adhering and one non-adhering) living within the Cape metropolitan area. To ensure reliability; the socioeconomic background of the participants in the pilot study was chosen to match those in the study.

The mean time in hours per interview was 1.5 hours, with a range of 40 minutes to 2 hours. Several questions were found to be ambiguous and were adapted accordingly, and questions were also added to aid the exploration of certain sections more in-depth.

2.5.3. Follow-up Group Discussion

A follow-up group discussion is a form of group interview that capitalises on communication between participants in order to generate data. Participants are encouraged to talk to one another: asking questions, exchanging anecdotes and commenting on each other's experiences and points of view. The idea behind the follow-up group discussion is that group processes can help participants explore and clarify their views in ways that would be less easily accessible in individual interviews (Kitzinger 1995).

The follow-up group discussion commenced once the information generated from the interviews was analysed, sorted into themes using content analysis and then used to formulate questions. The researcher used five open-ended questions in the follow-up group discussion guide (Appendix E) to add depth to and clarify the issues that were established to be important from the interviews. The follow-up group discussion also allowed participants to introduce ideas the researcher had not considered for the interviews.

2.6. DATA COLLECTION

The interviews and follow-up group discussion were tape recorded (with the participants' permission) and later transcribed by the researcher for analysis. The tapes were dated and labelled with a participant code only.

In qualitative research, new concepts are developed throughout the process of data collection until the end of research, and they are continuously adapted. In this study, the researcher

collected the data from the initial interviews and participant observations and started to analyse it at the same time that the other interviews were being conducted (Holloway and Wheeler 1996).

The researcher travelled to participants living in the Northern Cape to collect the data. The first trip was in April 2005 where the interviews were carried out, followed in July 2005 by a second trip to facilitate a follow-up group discussion.

2.7. DATA ANALYSIS

Data analysis in qualitative research is breaking down data and searching for codes and categories that are then reassembled to form themes. Data analysis occurs from the start of data collection, but the focus becomes progressively clearer (Holloway 1997).

Content analysis, a method of data reduction, was used to organise the qualitative data collected from the interviews and participant observations into conceptual frameworks to identify common themes. The conceptual frameworks or themes were used to formulate questions that were then discussed in the follow-up group discussion, where data collection was resumed to explore and challenge this conceptual framework.

Content analysis required the researcher to label phenomena and develop conceptual categories to describe and explain participants' reasons for adherence and non-adherence (Neuman 1999). These categories were derived inductively – obtained gradually from the data. The data were read and reread to identify and index themes and categories. All the data relevant to each category were identified and examined by constant comparison – each item is checked or compared with the rest of the data to set up analytical categories (Pope et al. 2000).

Table 4. Steps used for content analysis (Holloway 1997)

- Ordering and organising the collected material
- Re-reading the data
- Breaking the material into manageable sections
- Identifying and highlighting meaningful phrases
- Building, comparing and contrasting categories
- Looking for consistent patterns of meanings
- Searching for relationships and grouping categories together
- Recognising and describing patterns and themes
- Interpreting and searching for meaning

2.8. TRUSTWORTHINESS/ VALIDITY

The quality in qualitative research should be assessed differently from quantitative research. For this reason trustworthiness is used in qualitative research to measure the validity of it (Holloway 1997). Qualitative research needs to recognise that objective reality and subjective experiences possibly occur together in the research data. For this reason a decision trail must be shown (Holloway and Wheeler 1996). The decision trail is a detailed record of the methods and decisions made by the researcher before and during the research process. It aims to provide information necessary to judge the trustworthiness while doing an inquiry audit, which, according to Holloway is a: "systematic evaluation process of research to establish its quality" (Holloway 1997). The elements of the decision trail in this research project were recorded as follows:

- A description of the design (Section 2.2) with the aims and intentions of the research (Section 1.5)
- An explanation of the sampling process (Section 2.4)
- A detailed description of the setting (Section 2.3)
- A description of the data collection (Section 2.6) and analysis process (Section 2.7)
- A record of decisions about ethical issues (Section 2.9)
- Excerpts from the data (Chapter 3: quotes from interview transcripts and/or participant observations)

According to Lincoln and Guba (1985) trustworthiness involves the following options: credibility, transferability, dependability and confirmability. These four alternatives provide the foundations for demonstrating trustworthiness and the decision trail in qualitative research.

2.8.1. Credibility

To establish credibility, the researcher must ensure that the participants are identified and described accurately (Holloway and Wheeler 1996). This can be achieved by prolonged involvement, persistent observation, triangulation, peer-debriefing or member checks (Lincoln and Guba 1985). In this study, peer debriefing was used to improve the credibility of the study. The researcher ensured that the data was unbiased and a fair account of the participants' responses, known as inter-rater reliability, by meeting regularly during the research process with the neutral co-supervisor to analyse and interpret the data (Cutliffe and McKenna 1999).

2.8.2. Transferability

Transferability is when findings in one context can be transferred to similar situations or participants. Thick description was used to describe accurately and in detail the data in context so that peers and readers can decide if the participants described may be transferred to other settings or participants. Through purposive sampling specific information was described about the participants, giving a richer source of data to determine if transferability can be applied (Holloway and Wheeler 1996).

2.8.3. Dependability

If a study is to be judged dependable, it must be consistent and accurate. This was proved though a decision trail, in which the researcher provides detailed descriptions of the path of the research, so that the readers can follow the decision-making process (Holloway 1997). Thick description creates the opportunity for repeating the research, thereby ensuring dependability (Rossouw 2000).

2.8.4. Confirmability

The concept of confirmability means that the findings are the result of the research and not an outcome of the biases and subjectivity of the researcher. The data should be connected to their sources for the reader to determine if the conclusions and interpretations evolve directly from them (Guba and Lincoln 1989). Confirmability, like dependability, is best demonstrated through a decision trail (Holloway and Wheeler 1996; Holloway 1997).

In the current study, a genetic counsellor and a clinical genetic RN of the HNPCC service as well as a qualitative researcher/medical anthropologist reviewed the questions in the interview guide. This enhanced content validity and appropriateness by ensuring that the questions were easily understood, comprehensive and applicable to the participants.

2.9. ETHICAL CONSIDERATIONS

Research must always maintain ethical principles. This research project maintained ethical principles of participant autonomy, anonymity and confidentiality.

Application to the Research Ethics Committee of UCT for ethical clearance was undertaken prior to commencing with the study. Approval was granted with the project reference number 393/2004 (Appendix A).

The researcher was a second year genetic counselling student, who had been trained in basic counselling skills and had been interacting with patients in the genetic clinics since March 2004 under professional mentorship. The researcher could thus provide supportive counselling when necessary.

The autonomy of study subjects was respected - voluntary participation was essential. Participants were informed of this during the telephonic communication and were given an information sheet (Appendix B) containing this information at the start of the interview. They were made aware that they could withdraw from the research at any time and that it would not affect their subsequent treatment. Verbal as well as written consent was obtained from all participants.

The information sheet and informed consent form (Appendix C) state that all information is strictly confidential and that any identifiable information will be safely stored. Participants were asked for their permission for the interviews and follow-up group discussion to be tape recorded and were made aware that within one year the tapes would be destroyed. A coding method was used to identify each participant. Only the principal investigator for this study had access to the code.

All the interview guides, information sheets and consent forms were in Afrikaans, since all the participants in the study were Afrikaans-speaking. Although the researcher could speak Afrikaans, she was limited in her ability to understand the in-depth, emotional responses of participants, and for this reason a translator was used. The translators were qualified psychiatric RNs working in the HNPCC team, and translated the interviews and the follow-up group discussion.

2.10. RISK BENEFIT

The participants in this study were asked about sensitive and distressful experiences in their life during the interview and for this reason the researcher paid particular attention to the needs of the participants and was aware of their emotional state throughout the study. Participants were debriefed at the end of their interview, which gave them the opportunity to share their feelings and concerns and ask further questions. Participants were offered referral for genetic counselling so that any issues that come out in the study could be dealt with in-depth.

An information session was given to participants after the interview, which gave the researcher an opportunity to reinforce information that all participants had previously heard during the

predictive testing period. Participants, thus, had time to ask questions about the genetics of HNPCC and improve their understanding of this disorder.

The long-term benefit of this study will be to use the information gained to improve the genetic counselling process in motivating, exploring and supporting individuals who do not adhere to surveillance with the end goal of maximising surveillance adherence, especially since screening for HNPCC through colonoscopy and removal of polyps is an effective approach in decreasing overall morbidity and mortality due to CRC in HNPCC families (Jarvinen et al. 2000).

2.11. ASSUMPTIONS

The researcher assumed that the responses of the participants were honest and a true reflection of their lives and feelings.

2.12. LIMITATIONS

- The sample size of eight individuals might have led to incorrect inferences.
- The lack of international literature and no literature on South Africa and other developing countries about the research topic.
- Researcher Bias: The participants' responses might have been what they thought was appropriate rather than their true attitudes (Holloway 1997).
- Rumination Bias: The participants were confronted with questions to which they previously had not given much consideration (Sackett 1979).
- Selection bias: non-compliers were over-represented among those who decline to participate in the study, which is a common limitation in compliance studies (Neilson and Whynes 1995).
- Since the interview guide was based on the literature and the expert opinions of a RN and a genetic counsellor, their perception of the factors of influence were questioned and thus the interview guide might have missed relevant points from the participants' perspective.
- The small sample size and the ethnic homogeneity of the study sample may limit the generalisability of the study results.

CHAPTER 3:
FINDINGS

CHAPTER 3

FINDINGS

3.1. INTRODUCTION

The aim of this study was to provide a pilot investigation to understand and discover what influences people who are at high risk for CRC to be adherent or non-adherent to surveillance guidelines. This chapter presents the reasons for non-participation, the sociodemographic and socioeconomic background and the themes that emerged from the adherent and non-adherent interviews, participant observations and the follow-up group discussion.

Within the text the non-adherent participants are referred to as P1, P2 and P3; the adherent ones as P4, P5, P6, P7 and P8; and R refers to the researcher. P9 was a non-adherent participant who declined to take part in an interview, but consented to be involved in the follow-up group discussion. The data obtained from P9 during this discussion was included in this chapter where applicable. For clarity, it is important to note that the total number of participants discussed in Chapter 3 is thus nine, unless otherwise stipulated.

3.2. PARTICIPATION

All participants who were selected for this study had received their positive genetic test result. Of the participants who provided verbal consent, 27% (3/11) of the non-adhering compared to 83% (5/6) of the adhering participants took part in the interviews (Table 5).

Table 5. Summary of the number of adherent and non-adherent participants who gave verbal consent compared to those who were interviewed for the study.

Participants	Number giving verbal consent	Number interviewed	Reason for non-participation
Adherent	6	5	- Did not remember receiving result
Non-adherent	11	3	- Did not remember receiving result(x3) - Ill at time of interview (x2) - Declined to participate (x2) - Did not arrive for interview (x1)

One of the reasons for the high non-participation in this study was because before the start of four of the interviews (3 non-adhering and 1 adhering) it was evident that these individuals believed they had not yet received their predictive genetic test result and for this reason were

excluded from the study. However, one non-adhering participant, who was also confused about her genetic test result, was not excluded from the study since it only became apparent during the interview that she thought that she had not received her result. During her interview, questions pertaining to the test result (section B of the interview guide, Appendix D) were left out. These five individuals will be followed up by the CRC team to deliver their genetic test results again and to provide supportive counselling. The reason given by the two individuals who declined to participate in the study was that they did not feel comfortable to talk to the researcher about the research topic.

3.3. SOCIODEMOGRAPHIC AND SOCIOECONOMIC BACKGROUND

All participants in the study were of mixed ancestry and living in Port Nolloth, Nababeep, Okiep or Komaggas in the Northern Cape Province of South Africa as indicated on the map in Figure 4 (page 18). The sociodemographic and socioeconomic background for each participant is shown in Table 11 and 12 (Appendix F), while the details for the group are tabulated below (Table 6).

Table 6. The sociodemographic characteristics of the non-adherent and adherent participants in the study (n=number of participants).

Variable	Non-adherent [n=3]	Adherent [n=5]	Total [n=8]
Sex			
Male	1	2	**3**
Female	2	3	**5**
Age[a]	38	39	**39 (33–46)**
Marital status			
Divorced	3	-	**3**
Married	-	4	**4**
Separated	-	1	**1**
Participants with children	3	5	**8**
Number children[a]	2	2	**2 (1–3)**
Education			
Low (< grade 7)	-	2	**2**
Middle (grade 7-11)	3	2	**5**
Grade 12	-	1	**1**
Qualification after school	-	-	**-**
Employment status[b]			
Unemployed	2	2	**4**
Full time employed	-	2	**2**
Housewife	-	1	**1**
Household income/month[c]			
R801 – R1 600	1	-	**1**
R1 601 – R3 200	-	1	**1**
R3 201 – R6 400	-	1	**1**
R6 401 – R12 800	-	2	**2**

[a] Average value and range in brackets. [b] Not applicable to non-adherent participant who was in jail, total n=7. [c] Not applicable to non-adherent participant who was in jail, and two participants who did not know their household income/month, total n=5.

Most (75%) participants completed primary school and one (12.5%) participant had matriculated, with this being the highest level of education for the group. The four unemployed participants were female, but had family members who were employed and generated the monthly household income. Two of the male participants were full-time employed, while the third male participant was in jail at the time of the interview.

The socioeconomic status of the participants in this study was low, with exception of the two full-time employed participants. As show in Table 7, the two most disadvantaged participants had a household income per month of R2 000 or less to support six people. The remaining three participants had a higher income, but this was still low to support either three or four people.

Table 7. The household income per month for each participant and the number of people it supports (n=5).

Household income/month	Number people income supports
R1 460	6
R2 000	6
R5 000	4
R8 000	3
R10 000	4

Not applicable to non-adherent participant who was in jail, and two participants who did not know the household income/month, total n=5.

3.4. COMPLIANCE AND SURGERY BACKGROUND

Compliance data were derived from the HNPCC database and was taken from the year the participants received their genetic test result. The adherent participants in this study had attended all surveillance procedures, whereas the non-adherent participants ranged from no attendance to attending three out of seven colonoscopies. Table 8 provides the attendance rates for adhering and non-adhering participants for colonoscopies and flexible sigmoidoscopies.

Table 8. Participant attendance rates to colonoscopies and sigmoidoscopies.

Surveillance Method	Attendance[a] (%)	
	Non-adherent	Adherent
Colonoscopy[b]	25 (0-43)	100
Sigmoidoscopy[c]	0	100

[a] Average value and range in brackets. [b] Non-adherent n=3, adherent n=5. [c] Non-adherent n=1, adherent n=2.

Three participants were advised to attend annual flexible sigmoidoscopy after they underwent surgery (TAC-IR) for CRC between 1999 and 2003 with the average age at diagnosis of cancer being 32 years. This type of surveillance is used to detect cancer in the

remaining section of colon and the rectum. Two of these participants (adhering) were undergoing regular colonoscopies when a CRC was detected, while the third participant (non-adherent) had never been for a colonoscopy until he was admitted into GSH for CRC symptoms.

The cost of surveillance and transport to the Primary Healthcare hospitals where screening takes place are compensated for by De Beer's Diamond Mines or by the Provincial Health authority. Most participants (75%) correctly stated that the transport was free of charge, while two participants had the impression that they paid for it. The CRC team offers screening in Springbok, Nababeep, Okiep and Kleinsee and the reported distance (in time) from the participants' place of residence to the venue ranged from 5 to 90 minutes. The location and costs of colonoscopies and transport for each participant is shown in Table 13 (Appendix G).

3.5. THEMES

The themes that emerged from the interviews, participant observations and the follow-up group discussion were obtained using content analysis. These themes were derived by ordering and organising the data and then re-reading it to break it down into manageable sections, of which categories were identified. These categories were compared and once consistent patterns of meaning were identified they were grouped together to form themes.

In this section, each theme is described within the dimensions of the following aspects of intervention or cancer and is summarised in Table 9:

- Intervention
  - Genetic service
  - Surveillance
  - Misunderstanding
- Cancer
  - Fear and concern of cancer
  - Personal and family history of cancer
  - Social stigma of cancer

The researcher has used direct quotes (presented in *italics*) from the participant interviews to illustrate and give depth to the themes described as well as provide validity thereof. To maintain confidentiality and anonymity the full transcripts were not included and names were replaced with "***". Omitted phrases were indicated by "(---)"

Table 9. Summary of the themes for non-adhering and adhering participants.

Theme	Non-adherent Participants [n=3 or 4]	Adherent Participants [n=5]
GENETIC SERVICE		
• Remembered dates of counselling and result-giving session	1/4	0/5
• Remembered geneticist who counselled and delivered result	2/4	2/5
• Knew genetic test result was positive	3/4	5/5
• **First attended colonoscopy:**		
- Before counselling or	1/3	0/5
- After counselling or	0/3	4/5
- After result given	2/3	1/5
UNDERSTOOD the following:		
• Reason for annual surveillance	0/3	5/5
• Function of surveillance	2/3	5/5
• Meaning of genetic test result	1/2 (1 did not mention)	4/5
SURVEILLANCE ISSUES		
• **Reasons for adherence**		
- To know health and cancer status	N/A	5/5
- To have treatment or surgery options	N/A	3/5
- Experienced deaths in the family from CRC	N/A	2/5
• **Influenced by family members':**		
- Non-adherence	Did not mention	2/5 (P6 motivated, P7 discouraged)
- Adherence	Did not mention	1/5 (motivated)
- Painful experience during colonoscopy	4/4 (discouraged to go) 2/4 (*reason for non-adherence*)	Did not mention
• **Personal experience of colonoscopy**		
Before colonoscopy:		
- Nervous or anxious	2/3 (all female)	3/5 (all female)
- Disliked preparation	3/3	4/5
- Preparation was worst or one of worst parts of colonoscopy	2/3	2/5
- Preparation was *reason for non-adherence*	2/3	N/A
During colonoscopy:		
- Discomfort	3/3	5/5
- Discomfort was worst or one of worst parts of colonoscopy	2/3	3/5
- Discomfort was *reason for non-adherence*	2/3	N/A

- Painful	0/3	4/5
- Embarrassed	0/3	2/5 (only for 1st colonoscopy)
- Sedative decreased pain and/or discomfort	1/1	4/5
After colonoscopy:		
- Relieved	3/3	5/5
- Cramps	0/3	4/5
• **Suggestions to improve colonoscopy**		
- No preparation	2/3	Did not mention
- Stronger sedative	2/3	4/5
- Have colonoscopy immediately (no waiting)	Did not mention	1/5
- Specific gastroenterologist to perform colonoscopy	Did not mention	2/5
- Allow him to have colonoscopies at Kleinsee *(reason for non-adherence)*	1/3	N/A
CANCER		
• **Realisation CRC in family**		
- Parent died of or developed CRC or	1/3	2/5
- Parent died of cancer or	-	1/5
- Family members died of CRC or	1/3	-
- First counselling session	1/3	2/5
• **Realisation of being at risk of developing CRC**		-
- Informed by parent with CRC or	1/3	5/5
- First counselling session or	-	-
- Result-giving session or	1/3	-
- Researcher unsure if she realises	1/3	
• **Reaction if CRC found for first time [n=5]**		
- Accept and handle it	2/2	3/3
• **Reaction if CRC found AGAIN [n=3]**		
- Not handle it or	-	1/2
- Upset or	-	1/2
- Did not realise could develop CRC again	1/1	-
• **Scared of developing CRC**	0/1 (2 did not answer)	2/5
• **Social stigma of cancer**	2/3 (1 did not mention)	2/5 (3 did not mention)

3.5.1. Genetic Service

This section illustrates the participants' response to and recall of the genetic service offered to them by the CRC team in terms of the first counselling session, the result-giving session and their general impression and opinion of the CRC team.

3.5.1.1. Initial counselling session

As can be seen from Table 10 below, all participants were initially counselled either in 1994 (1/9), 1995 (2/9), 1996 (4/9) or 2002 (2/9). However, all but one (P3) participant either could not remember or was incorrect in their answer to the year in which they were counselled. P3, in contrast, remembered the year and month that he was counselled.

Table 10. The year blood was taken for predictive genetic testing and when participants received their results compared to the first year they had a colonoscopy.

Participant	P1	P2	P3	P4	P5	P6	P7	P8	P9
Year Blood Taken	2002	1996	2002	1995	1996	1996	1995	1994	1996
Year Received Result	2003	1997	2003	1997	1997	1997	1997	1997	1997
Year of First Colonoscopy	-	1997	2002	1995	1997	1996	1995	1994	1997

Four adhering participants (P4, P6, P7 and P8) were compliant with surveillance recommendations from when they were first counselled, which means that they were attending colonoscopies before they had received their positive genetic test result. This implies that these participants went for colonoscopies based on a 50% risk of developing CRC.

3.5.1.2. Genetic test result session

As with the counselling date, the same participant (P3) was the only one to correctly remember the date, and to the day, when he received his genetic test result. P4 correctly remembered that she received her result two years after her blood was taken, but could not recall the actual year. P1 was the only participant to believe that she had not yet received her genetic test result. She claimed that she and her sister did not understand what the people who initially counselled them had explained to them.

"Hulle het net ons bloed getrek, net datums gegee(---) Hulle het glad nie met ons so gepraat nie, soos ons nou sit en praat nie, want as ons na die kliniek toe gaan vir bloed toetse dit is 'n klomp. Daar is nie kans om te sit en praat nie." (P1)

Four (P2, P4, P5 and P9) out of nine participants (44%) remembered the geneticist who delivered their genetic test result. P8, in fact, believed that it was the gastroenterologist who provided him with his result and P3 recalled that one of the HNPCC RNs was the person who initially counselled him.

All participants, except for P1, correctly knew what their result was. Most (71%) referred to their result as 'positive', but their understanding of the meaning thereof will be discussed in section 3.5.2.1.

In contrast to the four participants who first attended colonoscopies after the initial counselling, one adhering (P5) and two non-adhering (P2 and P9) participants went for colonoscopies once they had received their result (Table 10).

3.5.1.3. Participants' opinion of the colorectal cancer team

All participants found the CRC team to be helpful and friendly. The adhering participants found that the team made them feel comfortable and less scared before and/or during their surveillance, with P4 and P5 finding it most reassuring when one of the team members held their hand and talked to them during the procedure.

"Hulle is mos maar altyd vriendelik. Hulle probeer jou net gemakliker maak. Hulle gesigte wys al klaar jy kan maar, hulle het oop gesigte. Hulle weet wat jou naam is, jy voel gemaklik saam met hulle." (P4)

One of the HNPCC RNs was a key person whom three participants (P3, P5 and P8) found to provide them with much support. P5 and P8 would talk to this RN when they felt scared and did not want to go for the colonoscopy. P5 also stated that if this specific RN (referred to as "***" in the quote below) was not present at the colonoscopy that she would then not go.

*"As *** ook nie hier is nie, dan gaan ons nie. Dit is eintlik 'nice' as sy hier is." (P5)*

*"Elke jaar, veral vir Sr. ***, ek ken nou die ander nie, hulle laat jou baie goed voel. Al was daar 'n tydjie wat jou 'n bietjie terughou en as jy bang is, dit voel hulle het jou so opgemaak. Hulle voel asof jou jare ken." (P8)*

All five adhering and three of the four non-adhering participants had no complaints about the people in the CRC team. However, even though P1 found the CRC team to be pleasant, she

mentioned that their explanation on genetics was above her level of understanding, which left her feeling confused and scared.

"...hulle is so bo met ons, dis hoekom ek en my suster bang is. Hulle praat nie 'straight' met ons nie, sodat ons nie kan verstaan nie." (P1)

3.5.2. Misunderstanding versus Understanding

This section explores and compares participants' misunderstanding or understanding in terms of what it meant to be mutation positive and their reason for regular surveillance.

3.5.2.1. Genetic test result

As stated earlier, all participants in this study, except for P1, knew that they were mutation positive. Participants demonstrated different levels of understanding of the meaning of such a result, from basic to a higher level of comprehension.

Four of the participants (P2, P5, P8 and P9) explained that a positive genetic test result meant that they were at increased risk of developing CRC. The risk, however, differed for each participant: P8 stated that he had a 70% chance of developing CRC; while P5 went from thinking it was 99% to 90% to higher than 50% chance; and P2 and P9 recalled that it was simply a high risk.

*"Hulle het spesiaal gekom na hulle bloed getrek het in Komaggas. Die ou dokter ***, daai Professor, hy het vir ons kom gesê wat hom kan kry, die moontlikheid. Jy het 'n sewentig persent kans om dit te kan kry." (P8)*

In comparison to the participants mentioned above, P6 and P7 explained their positive result with respect to surveillance for cancer, without mention of risks. P6 understood the implications of testing positive saying that he then knew how to handle his health because his father died of CRC.

"Ek het geweet hoe moet ek myself hanteer, want my vader is van daai siekte oorlede toe het ek geweet waar bestaan ek met die siekte." (P6)

P4 did not mention the meaning of a positive result. She only revealed that she was not surprised that she had the "mistake" since her father died of cancer a month after she was born, and so she actually expected such a result. Her statement, quoted on the following page, implies that because her father had cancer she would test positive.

This is an incorrect assumption since even though her father was affected with cancer, she still had a 50% and not 100% chance of inheriting the defective gene. It is difficult to gauge how much P4 understood of the meaning of her genetic test result, since her family history of CRC and not her high risk of CRC seemed to be her motivation for compliance.

"Maar ek was nie eintlike so verbaas nie, want my pa was net toe ek gebore 'n maand daarna toe sterf my pa van die kanker. So dit was eintlik te wag." (P4)

P3 also did not mention his understanding of a positive result. He explained that during the session a photograph was taken and the disease was discussed. This could suggest that either P3 did not understand what was talked about during the result-giving session, or that he did not mention what he understood.

3.5.2.2. Regular surveillance

All participants (except for P1 and P3) recalled that, since they were over 30 years of age, they should be attending surveillance annually. The adhering participants understood why they should attend regular surveillance, while the non-adhering participants did not.

Early in P3's interview he explained that the doctors had advised him to attend flexible sigmoidoscopies since there was a chance he could develop cancer after his surgery. He also described how his father had developed cancer after he was operated on and for this reason he believed he too had a chance of it happening again. But later on in his interview he was unsure as to whether he could develop cancer again.

"Hoekom het die dokters vir meneer gesé dat jy na die operasie vir die ondersoeke moet gaan?" (R)
"Want dit kan weer begin groei. En die rede hoekom ek vir hulle glo, omdat my Pa hy was klaar geopereer, huis toe, en toe kom die mense weer om die kolonoskopie te doen en die ambulans het vir hom gewag toe se hulle nee hulle het weer iets gevind. Dit is hoekom dit vir my net so belangrik is dit kan met my ook gebeur." (P3)

"Is daar 'n kans dat ek kan dit weer kry?"(P3)

P2 and P9 understood the purpose of regular colonoscopies, but were confused by the fact that their mother had died of breast cancer and not CRC, and were thus unsure at to why they should be going for yearly surveillance of their colon and not their breasts.

"My Ma het nie van die dikderm gesterf nie. Toe sê hulle nee, maar dit is nou die saak en ons moet gaan vir die toetse(---). My Ma het nie gesterf van daardie probleem nie. Hoekom moet ons elke jaar gaan?" (P2)

Besides being unsure as to the purpose of surveillance, P1 and P3 were also confused with what the actual colonoscopy procedure entailed. P1 was unsure where the colonoscope was inserted, thinking that it was through the mouth or the breast and believed that the colonoscope was checking for a fungus. P3 described the worst part of the colonoscopy being the pipe they put down his throat - a gastroscopy, which is not routinely done with a colonoscopy.

"...ek is rêrig net bekommerd of the pyp hier afgaan of waar hy afgaan. Dis al my probleem."(P1)

"Toe hulle die bloed getrek, wat het hulle vir jou gesê van die kolonoskopie?" (R)
"Hulle het net gesê daar is 'n swam, so iets. Maar hulle het nie verder gepraat nie. Waar dit is nie, het hulle vir my Ma gesê, toe verstaan my Ma."(P1)

"Wat was die slegste ding van daardie kolonoskopie." (R)
"Die ding wat hulle hierbo insteek." (P3)
"Dié wat jy drink?" (R)
"Nee nie drink nie." (P3)
"Ooh, die gastroscopy." (R)
"Ja." (P3)

3.5.3. Surveillance Issues

The data in this section reports on the following issues related to surveillance: participants' self-reported reasons for adherence and non-adherence; the influence of family members; their personal experience of the procedure; and suggestions given to improve the colonoscopy.

3.5.3.1. Adherence to surveillance

- **Reasons for non-adherence**

All non-adherent participants, except for P2 and P9, stated different reasons for not being compliant with surveillance guidelines. P1 had never been for a colonoscopy and her reasons for non-attendance in 2004 were because her brother died and someone she knew went into labour. She also mentioned that she was willing to go but was very scared because she did not know how they performed the colonoscopy.

"...ek is gewillig maar ek is baie bang. Sal ek laas jaar gegaan het toe kry sy bevalling toe kon ek nie gaan nie."(P1)

Since receiving a positive genetic test result in 1997, P2 had attended three out of seven colonoscopies, while P9 had only ever had one colonoscopy. Their reasons for non-adherence were based on the procedure itself and specifically the preparation drink before and the discomfort during the colonoscopy. They also mentioned that their sister's painful experience during the procedure had influenced them not to attend surveillance for the fear of feeling the same.

"...is die water en daai gevoelig(---) Dit is gevoelig, regtig waar(---). Maar, met al hulle respek, as hulle daai twee goed 'n verandering maak. Maar verder, regtig waar, gaan ek nou bly en tot hulle daai ding uitge'sort', want dis verskriklik seer." (P2)

The only surveillance P3 had attended was when in 2002 he was admitted to GSH for CRC symptoms and then diagnosed with cancer after a colonoscopy. Blood was also taken at this time at GSH for predictive genetic testing. His reason for not going for both the genetic test and surveillance before 2002 was that he was not allowed to have it done in Kleinsee, a restricted area belonging to De Beer's Diamond Mines, because of his criminal record. Due to the fact that P3 could not enter Kleinsee, transport had been organised for him to attend surveillance in other areas. However, he remained non-compliant because he did not want to travel from one place to the next compared to going directly to Kleinsee for surveillance:

"Nou wat eenkeer gesê het vir myself, jong ek moet waai na Springbok toe, en dan moet ek weer daar van daan gaan na Nababeep, van een plek na 'n ander plek. Dan moet ek nou weer terug kom, en dan moet ek nou daar vandaan huis toe(---) ek wil graag, as ek kon, kon ek lankal gegaan het. Van dié het ek nie hierdie maand gegaan nie, omdat ek nie daar (Kleinsee) *kan uitkom nie." (P3)*

- **Reasons for adherence**

The adherent participants went for colonoscopies for their health and to know their cancer status, for treatment or surgery options if cancer was found and because family members had died of CRC.

  - Health and Cancer Status

Even though the participants had varying levels of understanding, all five adherent participants went for colonoscopies for their health and so that they would know their cancer

status. P6 explained that the colonoscopy would tell him whether he was busy developing cancer and thus he would be aware at an earlier stage what his disease status was.

"Ek kan sien dat die kolonoskopie gaan vir my op 'n vroeër stadium sê ek is besig om kanker te kry. So ek sal op 'n vroeër stadium sal ek al bewus wees of ek besig is om kanker te kry."(P6)

P7 reasoned that she would not know her disease status unless she went for a colonoscopy and thus attended surveillance for this assurance. The knowledge of his cancer status gave P6 a sense of control, so much so that he actually enjoyed going for colonoscopies. P5 associated going for colonoscopies with staying alive to take care of her child. Two participants (P4 and P8) had undergone surgery and were thus advised to go for flexible sigmoidoscopies, which they attended because they were aware that they could get cancer in the remaining areas of the colon and rectum.

"Omdat ek sien hierdie jaar kan dit miskien gebeur, dit pla my nog nie, nou weet hulle dit mos nie. Daarom gaan ek maar elke jaar. Dan het ek daai versekering, okay ek is 'fine', ek is skoon."(P7)

- Treatment or surgery options

Three adhering participants (P6, P7 and P8) reasoned that by not going for surveillance they would not know their cancer status and without this knowledge, P7 explained, the doctors would not be able to help her if she did have cancer. It was justified by P6 that the advantage of adherence over non-adherence was to know if one had cancer for which there would be a chance for treatment. One of the participants (P8) went regularly for flexible sigmoidoscopies so that if another cancer was detected he could have surgery again to remove his entire colon and the cancer.

"As hulle by my dit kry, dan is daar 'n kans vir geneesing. Maar wat sal ek maak as ek nie vir die kolonoskopie gaan nie, hoe sal ek weet ek het kanker." (P8)

- Deaths in the family from CRC

Two participants (P5 and P6) were motivated to go for colonoscopies by the death of a parent and/or other close relatives of CRC. It seemed that P5 was suggesting that because many of her family members had died of cancer, that this made it even more necessary for her to attend colonoscopies. P6, on the other hand, explained that his father – who died of CRC – would have gone for colonoscopies if they had been available when he was alive and this encouraged him to remain adherent.

3.5.3.2. Influence of family members

Non-adherent and adherent participants were influenced to a certain extent by family members' adherence, non-adherence, or experience of colonoscopies.

- Non-adherent family members

Two participants (P2 and P9) did not mention the effect non-adherent family members had on them. P4, P5 and P8 stated that they were unaffected by their non-adherent relatives who also carried the causative mutation and were thus advised to attend regular surveillance. This compared to P6 who was motivated to be adherent by his father's non-adherence, and P7 who was discouraged when other family members did not attend colonoscopies.

"Beïnvloed dit vir mevrou as mense nie vir die kolonoskopie gaan nie?"(R)
"Dit raak my nogal, want ek praat mos met hulle baie (---) Ek het ook op 'n stadium gesê: "hoekom moet ek?" Verstaan? Toe ek nou moet gaan, "hoekom?" het ek so gevraae?"(P7)

- Adherent family members

P3 mentioned that his adherent father had always advised and encouraged him to attend surveillance. However, P3's non-adherence shows that this did not influence him to attend. On the other hand, P6 and his sister encouraged each other to go. The other five participants did not mention any affect adherent family members had on them.

- Family members' painful experience

All non-adherent and no adherent participants were affected by a family members' experience of the colonoscopy. P1's mother, P2 and P9's sister, and P3's brother had painful experiences during and after their colonoscopy(ies). This was one of the self-reported reasons P2 and P9 gave for their non-adherence.

3.5.3.3. Personal experience of colonoscopy

During this section, the common themes of the participants' colonoscopy experience before, during and after (except for P1 who had never attended surveillance) will be described.

- Before the colonoscopy

All the female participants and no male participants felt nervous, scared or anxious at some point before the colonoscopy. P2 and P9 felt nervous the day before, when they had to drink the preparation. Two participants (P4 and P7) felt tense when they arrived at the clinic the day of the procedure, explaining that the longer they had to wait to go for their colonoscopy the more anxious they felt. P4 also was more scared before the colonoscopy than during it.

"Ek is nie bekommerd by die huis nie, ek is bekommerd as ek daar is. Ek is nie so bekommerd nie, ek is net bang (---) Maar as ek die ding rerig doen, dan is dit nie so erg nie."(P4)

- Colonoscopy preparation

Three participants (P3, P7 and P8) disliked the preparation, but found the discomfort during the procedure to be worse, while P5 did not mind taking it. In comparison P2, P4, P6 and P9 found the preparation to be the worst or one of the worst part(s) of the whole process, with complaints that it made them nauseous (P2, P4 and P9); it was unpalatable (P2, P6 and P9) and it made P9 vomit. For P2 and P9, the preparation was one of the main reasons why they did not go for colonoscopies.

- During the colonoscopy

All participants found the colonoscopy to be uncomfortable and five participants (P2, P5, P7, P8 and P9) found this discomfort to be the worst or one of the worst part(s) of the process. One of the reasons four of these participants (P2, P5, P8 and P9) found the procedure to be uncomfortable was that they could feel the colonoscope moving from the moment it was pushed into the anus until it was pulled out at the end of the procedure. P2 and P5 both mentioned that the movement of the colonoscope at the central part of the diaphragm near the solar plexus was the most uncomfortable part of the procedure. P5, P7 and P8 also attributed their discomfort to the wind that was pushed into the colon during the procedure causing them to feel bloated and to develop cramps.

"As die pyp ingaan dan voel jy tot hy uitkom, jy voel alles, alles" (P5)

"...as hulle daardie pypie insteek hoe hulle daardie wind so blaas, dit maak dit of jou maag bars. Dit is mos baie ongemaklik, nou dis dit."(P7)

All but one (P6) adhering participant found the procedure to be painful or sore, compared to the non-adhering participants who did not. P5 attributed the pain she experienced to the tenderness she felt during the procedure, while P8 found the cramps to cause him the most pain. P4 was unsure as to whether her fear caused her to feel pain or tenderness during the procedure.

"Seer kry, ek wil nie seer kry nie. Dié is gevoelig, só die vrees maak dat ek seer kry. Só ek kry nie rerig seer nie, maar dis net die gevoel."(P4)

Five of the six participants (except for P8) who had experienced at least one colonoscopy with and at least one colonoscopy without a sedative, indicated that the pain and discomfort was reduced as a result of it.

None of the participants reported feeling embarrassed during the surveillance. However, P4 and P8 admitted that during their first colonoscopy they did because the people and the process were unfamiliar to them.

"Die eerste keer jy voel onbekend. Van die tweede jaar af, het ek gevoel dit is 'alright'. Die mense ken mos jou storie. Ek hoef nie om vir hulle skaam te wees nie." (P8)

- After colonoscopy

All participants indicated a sense of relief at the end of the procedure. P5 described that she felt better as they pulled the colonoscope out because she knew that it was near the end of the procedure. Compared to none of the non-adhering participants, four (P4, P5, P7 and P8) of the five adhering participants complained of cramps after the procedure. For P5 and P8, these cramps resulted in them being absent from work the following day. P5 experienced winds after the colonoscopy and felt embarrassed to pass the wind in front of the people who were waiting for surveillance at the hospital.

"Jou maag is vol winde en as die winde uit is, sal dit beter voel. Ek kan ook nie wind voor mense nie, want dit stink nie, dit is nou net 'n geraas(---)maar dan kry ek krampe."

3.5.3.4. Suggestions to improve colonoscopy

All participants, except for P6 and P8, who had been for a colonoscopy suggested ways in which to improve the procedure for them. The following changes to the process, according to these participants, would help to make it a more pleasant experience.

- No preparation

P2 and P9 found the preparation to be one of the worst parts of the procedure. They stated that not less, but no preparation to drink would be enough of a difference for them to go. They suggested that something like a pill would be a suitable replacement for the preparation.

- Stronger sedative

Six participants (2 non-adhering and 4 adhering) felt that a stronger sedative would improve the procedure for them by reducing pain and/or discomfort. P5 even threatened not to go unless she was given a stronger sedative for her next colonoscopy.

"Nou hulle kan mos iets beter gee, iets sterker gee. So jy hoef nie só te voel hoe dit ingaan (---) Miskien iets wat sou jou doodmaak iewers. Jy kan wakker lê, maar jy voel alles van die begin af, oor al die draaitjies en ek het dit alles voel tot wanneer dit uitgetrek word." (P5)

- Specific gastroenterologist to carry out the procedure

P7 admitted that she preferred it when a certain gastroenterologist did the colonoscopy, since she felt more comfortable with him, which helped her to relax and thus feel less pain during it.

*"Nou ek verneem, en dis ook nie reg nie, mos alles met Dr. *** wees. Nou as ek by hom is, dan is ek meer gemaklik, omdat hy, ek sê nie die anders stel nie belang nie, maar ek voel dit nie eintlik nie met hom. En ek is nie so gespanne nie, ek ken hom. As jy gespanne is dan trek dit jou liggaam en dan is dit seer." (P7)*

- Less time to wait before procedure

P4 and P7 found that the longer they waited to go for the procedure, the tenser they became and the worse the procedure was for them. P4 would choose to go first for her colonoscopy so that she would have it immediately, and would thus feel less tense and thus less pain during the procedure.

- Handcuff him so that he could have his surveillance in Kleinsee.

As P3 was prohibited from entering Kleinsee, he suggested that the CRC team handcuff him, take him to Kleinsee for the flexible sigmoidoscopy and then let him loose when he was out of Kleinsee again.

"...as 'n mense van die Kaap kom, hoekom maak ek so, as u nie wil hê ek moet daar ingaan nie, hoekom boei my nie vas nie, dat ek gaan by die hospitaal bring my toetse uit, bring my uit, maak my los, laat ek huis toegaan." (P3)

- Cannot be improved

For P8, the colonoscopy could not be improved because it was an efficient procedure for its function. He acknowledged that it was painful, but stated that this could not be changed.

"Ek sou sê, daai ding kan nie beter gemaak gewees nie soos dit nou is. Omdat dis voldoende. Dit moet so wees om dit te kan doen. Dis net seer gewees, dit kan nie nou nie meer seer geword nie." (P8)

3.5.4. Cancer

This section provides some insight into the participants' awareness of CRC in their family as well as when they first realised that they were at risk of developing the disease. Participants' response to a polyp or cancer being detected during surveillance was also explored, together with their fear of developing CRC. Since P9 did not participate in the interviews, she was not asked the questions that lead to this section, and for this reason the total number of participants is eight.

3.5.4.1. Realisation that colorectal cancer was in their family

Three participants (P3, P5 and P6) first realised that CRC was in their family when one of their parents either died of or developed CRC. P3's father and P5's mother developed CRC when they were 27 and 30 years old, respectively, and P6's father died of CRC when he was 22 years old. P4's father died of liver cancer and not CRC when she was one month old and this was when she knew that cancer was in her family. After multiple members of P1's family died of cancer she became aware that the disease was in her family.

P2, P7 and P8 first became aware that CRC was in their family when a member of the CRC team first counselled them. P7 described that this initial counselling session was the first time she realised why several people in her family had died.

3.5.4.2. Realisation of being at risk of developing colorectal cancer

Five participants (P4, P5, P6, P7 and P8) first realised they were at risk of developing CRC when they were initially counselled (Table 10, page 36). P2 became aware of her own risk for CRC when she received her genetic test result and for P3 it was when his father told him that he was at risk, which was years before he was counselled by the CRC team. P1 did not seem to be aware at the time of the interview to know that she was at risk of developing CRC. This was indicated when she stated that she was worried about her mother getting cancer and not for herself, since her mother was older than she was.

"As 'n mens praat van kanker is ek baie 'worried' vir my ma, nie vir myself nie. Ek is jonger, sy is al oud." (P1)

3.5.4.3. Reaction when cancer was found during a colonoscopy

As stated before, P3, P4 and P8 had undergone surgery after a cancer was detected in their colons. P3 described the shock he experienced when he realised that he had CRC, which compares to P8 who had expected it. P3 had never been for a colonoscopy until he was ill with cancer, whereas it was P4's sixth and P8's fourth colonoscopy when CRC was detected. This could explain why P3 and not P8 was surprised when cancer was found. P4 and P8 described how they had been worried for the pain and recovery after the operation.

3.5.4.4. Reaction if polyp or cancer found during surveillance

Five participants' (P1, P2, P5, P6 and P7) felt that they would accept and handle the news if something was found during surveillance. In comparison, P4 felt that she would not handle it if another cancer was found during her surveillance and was certain that she would not have another operation because of the complications she experienced for her first one. P8 sensed that he would be upset if cancer were found again. He knew that it was his choice to have another operation and felt that he probably would have the surgery.

"As dit gebeur moet ek maar sien. Maar ek sal nie weer vir 'n operasie gaan nie. Ek wil nie weer gesny raak nie (---) En dit vat elke keer langer om gesond te raak. Nee ek sal dit nie weer wil hê nie." (P4)

"Dan ek sal 'upset' wees as hulle nou weer moet kry. Dan gaan hulle my weer sny, maar dit is ook my keuse daai. As ek nou Augustus maand gaan vir die toets en hulle sê daar is fout, dan gaan hulle maar sny. Dit was iets wat ek moet 'face'. Afhangend hoe jy dit vat." (P8)

3.5.4.5. Fear of developing colorectal cancer

P4 and P6 where scared of developing CRC, compared to P1, P5, P7, P8 who were not. P2 and P3 were not willing to answer the question. P1 explained that because many people had cancer she would accept it and not be afraid if she too developed it. She also reasoned that since her mother was older (55 years old) than she was, she was worried that her mother would develop cancer and not that she would. P5 was not afraid of developing CRC because she had Jesus in her life, while P7's reasoning was that if cancer was found that the doctors could provide her with treatment. P8 described that since he had already had one operation he knew what to expect and thus he was less worried if he were to have surgery in the future.

"Kanker kan jy nou nie bang wees nie. Elke minuut in die lewe kry mense kanker. Ek kan nie eintlik sê ek is bang nie. So ek kan nie eintlik sê ek is bang nie, as die werklike kom dan moet ek dit maar aanvaar." (P1)

3.5.5. Social Stigma of Cancer

Stigma, in terms of cancer, presented itself during five of the eight interviews. For three participants (P1, P2 and P7) this came up when they were questioned as to who they would share their genetic test result with. All three stated that they would only tell close members of their immediate family, but would never share this information with other people in the community because they believed that they would then gossip about them and not keep it a secret. P1 explained that the community believed that cancer was, in a way, contagious by stating that if someone had cancer people would not eat or sleep with them.

"Kyk, sê maar nou soos ek en (R) ons ken nie mekaar, ek gaan en ek sê vir (R) ek was na die dokter en die dokter het uitgevind dat ek kanker het, dan sê (R): "ek is jammer, sorry". Maar as ek loop (R) gaan vir almal sê: "oppas vir daardie meisie sy het kanker", dan sê hulle: "moenie saam met hulle eet nie of slaap nie". (P1)

Another aspect of stigmatisation apparent during two of the participants (P2 and P3) interviews was the use of a clan name to distinguish families with their same surname who had HNPCC from those who did not.

*"Toe wil sy (*nurse at Kimberley hospital*) vir my 'ignore', my maag wat ek weet terselfde tyd dat hier is nie 'n dokter nie. Toe hoor sy ek is ook van die *** van Komaggas af, toe sê sy vir die dokter, toe sê die dokter nee, as jy een van die *** van Komaggas is, dan moet jy Kaap toe. Nou die oumense wat sê ek moet Kaap toe."(P3)*

Three adherent participants (P4, P5 and P6) did not mention stigma of cancer during their interviews. One adherent participant (P8) stated that he was not embarrassed or ashamed to tell people that he had cancer and an operation, indicating that he did not believe that there was stigma of cancer in his environment.

"...ek is nie skaam vir dit nie, en ek praat nog by die werk ook. En hulle weet vir wie ek ken, hulle weet ek het so 'n operasie gehad. Dit is nie 'n ding wat jy wegsteek nie." (P8)

CHAPTER 4:
DISCUSSION OF FINDINGS

CHAPTER 4

DISCUSSION OF FINDINGS

4.1. INTRODUCTION

From 1991 the Division of Human Genetics at UCT starting collecting blood for a research project to identify the disease-causing mutation for HNPCC in affected family members in the Northern and Western Cape Provinces of South Africa. The causative mutation was found four years later by Prof. Ramesar (Goldberg et al. 1998; Ramesar et al. 2000). This finding allowed for predictive genetic testing to be offered to first-degree at-risk family members identified through genealogical assessment.

The predictive testing programme at UCT routinely provides pre- and post-test genetic counselling to individuals involved. During the initial pre-test counselling session, first-degree relatives are informed of their 50% risk status and the implications of predictive genetic testing. They are also advised to attend colonoscopic surveillance at this stage. During the post-test result-giving session, individuals are informed of their mutation status and subsequent recommended screening. Genetic testing has helped to simplify management since all mutation negative individuals are released from surveillance, while those testing mutation positive are recommended to go for regular screening because of the recognised benefits (Jarvinen et al. 2000).

Many of the individuals involved in the HNPCC research project reside in rural and under-resourced areas of the Northern Cape and are from a socioeconomically disadvantaged background. For this reason, a specialist team travels to these isolated areas to offer screening to those at risk. Since De Beer's Diamond Mines and Provincial Health authority fund this service, it is brought to these individuals free of charge.

As mentioned in Chapter 1, a total of 1285 individuals from 351 families have been recruited onto the HNPCC research project, to date. The majority of these individuals live in the Northern and Western Cape. Fifteen of these families are involved in predictive testing – in which 145 individuals have the disease-causing mutation. Of these high-risk individuals, 61% are adhering to surveillance recommendations while 39% are not. There is concern for those who do not attend screening, since delays in surveillance can significantly increase the risk for developing interval cancer (Vasen et al. 1995).

The limited literature internationally and its non-existence in South Africa highlighted the need for a study to investigate the reasons why this sub-group of individuals do not adhere to surveillance guidelines, despite it being a free service. Therefore this research study aimed to identify, using qualitative methodology, factors associated with adherence and non-adherence to surveillance guidelines in a small group of mutation positive individuals, living in rural and isolated areas of the Northern Cape of South Africa. The study group consisted of eight individuals (5 adhering and 3 non-adhering) for the individual interviews. Two of these individuals (1 non-adhering and 1 adhering) took part in the group discussion with the addition of a non-adherent individual who did not take part in the interview. Ultimately, however, the end goal of this research project together with a more comprehensive investigation would be to improve genetic counselling services in motivating, educating and supporting individuals who do not adhere to surveillance, in order to maximise screening adherence for those at high risk for CRC.

4.2. SOCIODEMOGRAPHIC AND SOCIOECONOMIC BACKGROUND

All participants in this study were living in resource-poor areas of the Northern Cape Province of South Africa. More than half of the participants were unemployed and none had achieved a tertiary education. The majority of the participants had a low household income per month for the number of people it supported. Compared to the other participants – where sociodemographic information only appeared during section A of the interview (Appendix D); issues around poverty together with family and social issues dominated one non-adhering participant's interview. The researcher wondered if these issues also dominated her life and whether problems of survival and family strife were more important and more of a reality in her life than the threat of developing cancer. There is a chance that these issues played a part in her non-adherence to surveillance guidelines. Although one cannot assume that similar issues do not play a major role in the other participants' livelihood, this was not indicated in their interviews.

4.3. THE PROGRESSION AND IMPACT OF THE GENETIC SERVICE

As the genetic service has progressed in the participants' lives (Figure 5) – from predictive genetic testing to surveillance to detecting a cancer for some – it has affected and influenced their understanding and actions in terms of cancer screening. This section will discuss the role each step of the genetic service has played on the participants with reference to Figure 5.

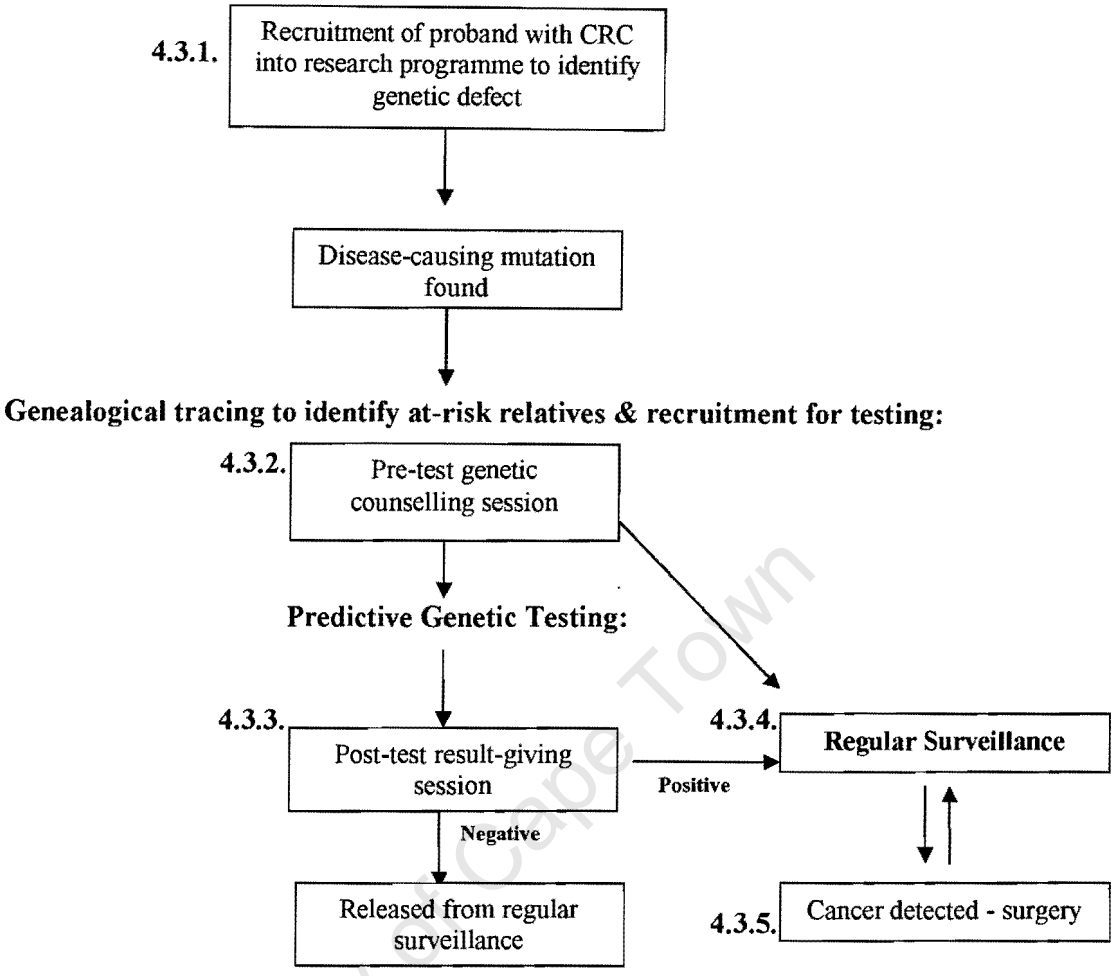

Figure 5. The process involved in the genetic service offered to HNPCC families.

4.3.1. Family Member with Cancer

Before the genetic service had a part to play in the participant's lives, the majority (88%) had already experienced cancer in a first-degree relative such as a parent or sibling. Although prior to this, participants had lost several relatives from cancer, it was either the death or the onset of cancer in a parent or sibling that made four of these seven participants aware for the first time that cancer was in their family. One participant had not lost a first-degree relative from cancer, but had lost several close relatives, which made her realise that cancer was in her family. At this stage, only one non-adhering participant realised that, because of his family history of cancer, he was thus also at-risk of developing CRC. Even though he had this knowledge, it did not influence him to attend the surveillance that his affected father went for. None of the participants had undergone colonoscopies at this first stage of their process.

In the literature on screening practices in HNPCC families more than half of the study group had attended at least one screening procedure before they were approached for genetic counselling and testing, compared to none of the participants having undergone a colonoscopy prior to this

(Johnson et al. 2002; Hadley et al. 2004; Halbert et al. 2004). This suggests that at this stage the participants in this study were unaware of their elevated risk for CRC and thus did not realise that they required intensive screening.

4.3.2. Pre-Test Counselling Session

During the pre-test counselling session the participants were approached and offered predictive genetic testing by the genetic counsellor and counselled about the effect their family history of CRC had on them. The participants' actions and reactions thereafter demonstrate their understanding of the information explained to them by the genetic counsellor.

It was during this session that all participants had their first contact with the genetic service. It was also during this time that three participants (two adhering and one non-adhering), who had not yet realised that CRC was in their family, were made aware of this for the first time. This means that at this stage all the participants had the knowledge that CRC was a disease in their family.

The adhering participants realised for the first time, during this session, that they were at-risk of developing CRC. Interestingly, the two remaining non-adhering participants were still unaware of their risk for developing CRC after this initial pre-test counselling session.

All but one adhering participant compared to none of the non-adhering participants went on to attend and adhere to colonoscopy surveillance after the initial counselling session, which was also when they first realised they were at-risk of developing CRC.

The literature on adherence behaviours shows that the rate of colonoscopy adherence increases from pre- to post-test counselling, but these studies have not measured the effect of pre-test genetic counselling on compliance (Johnson et al. 2002; Hadley et al. 2004; Halbert et al. 2004). This study, however, illustrated that the pre-test session influenced the adhering participants to attend surveillance, while it had no affect on the non-adhering individuals.

4.3.3. Post-Test Result-Giving session

The majority (78%) of the participants received their predictive test result in 1997, and two did in 2003. The turn-around-time from when blood was taken during the initial counselling session to when the result was delivered to the participants during the post-test session ranged from one to three years, but for the majority (67%) it was within one year.

Most (88%) of the participants could recall that their genetic test result was positive, except for one non-adhering participant who believed that she had not received her genetic test result and

was also unaware that she was even at-risk of developing CRC, despite their being evidence to the contrary. This confusion was also the reason why 44% of non-participants were not eligible to take part in the study, of which the majority (75%) were non-adherent. It is not known whether other individuals who have received their positive predictive genetic test result also do not recall that they have, and thus is an area of interest for future research.

The majority (71%) of the participants who knew they had received their predictive genetic test result understood, with differing levels, the meaning of it. A study by Aktan-Collan et al. (2001) found that misunderstanding the predictive genetic test result was associated with less worry about developing CRC, which was an influencing factor for non-adherence. This study, however, shows an equal number of adherent and non-adherent participants who misunderstood the result.

It was also during this session that the geneticist recommended and explained that the participants should attend regular surveillance, since they were at a high risk for developing CRC. None of the non-adherent participants understood the reason for attending annual surveillance compared to all of the adhering participants, who did. One of the reasons the adhering participants' gave for attending regular surveillance was to have control over and knowledge that they had cancer. This perception of control was also shown to influence adherence to surveillance in a study by Halbert et al. (2004). Another reason given for annual surveillance was that if cancer was detected that they could then be offered treatment or surgery.

4.3.4. Surveillance

4.3.4.1. Reasons for adherence

The compliance rates in this study were assessed over a period of eight years for 78% (7/9) and two years for 22% (2/9) of participants. Of the non-adhering participants: one (P1) had never experienced a colonoscopy; two participants (P3 and P9) had attended one; and one (P2) had had three colonoscopies. Thus, only the three non-adhering participants who had experienced a colonoscopy could give an aspect of the procedure as a reason for non-attendance, which was the case for two of them. The other two participants' reasons for non-attendance were a family death and pregnancy, and transport issues. The researcher believes factors such as misunderstanding and confusion around aspects of the genetic intervention, and socioeconomic and psychosocial issues might have also affected their non-adherence.

As mentioned previously, adhering participants reported that they attended regular colonoscopies to: (a) have control of developing CRC; (b) know their cancer status and (c) for

treatment and surgery options if cancer was detected. The participants need for control over developing CRC was also reported as a reason for compliant behaviour by Halbert et al. (2004). Previous literature has not, however, identified factors (b) and (c) as influencing screening adherence. The death due to CRC of several family members of one participant made her realise the seriousness of the cancer and motivated her to *continue* attending screening, not to start attending. Such a factor of influence has not been previously reported in the literature.

4.3.4.2. Family influence

All four non-adherent participants, compared to none of the adhering participants, had family member's whose painful experience(s) during a colonoscopy influenced them, and for two of them, was one of their reasons for them not attending screening. Some adhering participants, on the other hand, only mentioned the influence either adherence (motivated to attend) or non-adherence (motivated or discouraged to attend) of family members had on them. However, one adhering participant was not influenced by the non-adherence of relatives. She remarked that as a result of her relatives' non-attendance to surveillance their CRC was in the late stages when it was discovered. The literature has not identified the effect of a family member's painful experience during surveillance as a factor influencing non-adherence.

"Family interactions such as coping skills, communication and openness influence how each member responds emotionally and behaviourally to adverse circumstances" (Weil 2000). This study shows how the dislike of the surveillance procedure by one individual has profoundly affected the adherence behaviour of other family members, depending on the closeness of their relationship.

4.3.4.3. Participants' experience of the procedure

The following aspects of the colonoscopic procedure affected the participants' experience of it. The participant who had never experienced screening was not included in this section.

All five female participants were nervous or anxious before a colonoscopy compared to none of the male participants. This gender difference could be the result of the male participants not wanting to admit being nervous before a colonoscopy, tending to play down the difficulties they experienced. The male participants in the study were uneasy in talking about emotional subjects and did not open up during their interviews.

Even though all the participants disliked the preparation, only half of them found the preparation to be the worst or one of the worst parts of the screening. This differs from a study by Bleiker where it was found that for the total sample (adherent and non-adherent); the

preparation was considered the worst part of the procedure (Bleiker et al. 2005). The preparation was also the reason why two non-adherent participants refused to attend colonoscopies. They also claimed that they might consider attending surveillance if the preparation was changed to a pill.

All the participants found the colonoscopy to be uncomfortable, while 63% (5/8) found the discomfort to be the worst or one of the worst aspects of the screening. The same two non-adherent participants, whose reason for non-adherence was the preparation, also claimed that the discomfort during the colonoscopy was their other reason.

The majority (83%) of participants who had ever been sedated during screening found that it had decreased the pain and discomfort of the procedure, which agrees with a study by Bleiker et al. (2005). However, Bleiker went on to determine whether being sedated had positively influenced the participants' decision to undergo future screening, which it did. This was not asked in the present study, but would be appropriate to find out during future work. The majority (75%) of participants in this study, however, suggested that stronger sedation would improve the procedure for them by reducing discomfort or pain. This decrease in pain was so much so for one adherent participant that she refused to attend colonoscopies unless she was given a stronger sedation for her next surveillance.

Three participants described that the procedure was the most uncomfortable when the colonoscope was in the vicinity of the central area of the diaphragm near the solar plexus. According to Donaldson (2005), a practising clinical psychologist, this could be due to anxiety symptoms brought on by the tension they experienced during the colonoscopy procedure, rather than the movement of the colonoscope.

This study found that no participants felt the colon screening to be embarrassing, except for two adherent participants who did for their first procedure only, which was unlike a study by Bleiker et al. (2005) where non-adherent participants tended to rate the colon screening as being more embarrassing than the adherent ones and was thus a perceived barrier to screening. Interestingly, the majority (80%) of adherent participants found the colonoscopy to be painful and felt cramps after the procedure compared to none of the non-adhering participants. All the adhering participants had experienced six, seven or nine screening procedures, compared to the non-adhering participants who either had none, one (x2) or three (x1) colonoscopies. This lack of colonoscopic exposure of the non-adherent group could explain why most of the adherent and none of the non-adherent participants felt pain or developed cramps after the procedure.

4.3.5. Cancer Detection and Surgery

For two of the three participants in whom a cancer was detected and surgery performed, this occurred after several years of colonoscopy attendance and in the sequence of events shown in Figure 5. The non-adhering participant (P3), however, had the following sequence of events, with the first three steps occurring within a period of one month (symptoms of CRC → colonoscopy → pre-test counselling → surgery → post-test session). This participant had a colonoscopy because he was showing signs of CRC and it was when he went to GSH for investigation for CRC that he was offered predictive genetic testing. It was a year after his surgery and initial counselling session that he received his result, and had been non-adherent since. He was also unaware that another cancer could develop. This ignorance could have influenced his non-adherence to surveillance guidelines after his surgery.

For the two participants who knew that they could develop another cancer, one described that he would be upset and the other participant felt she would not handle it if, indeed, another cancer were found. This compared to those participants in whom a cancer had not been detected, and who all felt that they would accept and handle it if CRC were to be found during a colonoscopy. Their inexperience of surgery might have caused their indifference about developing CRC, compared to those who had experienced the reality of an operation.

Two participants (adherent) were scared of developing CRC compared to four who were not (three adherent participants and one non-adherent). The remaining non-adherent participants chose not to answer the question. This compares to the study by Bleiker, where fear of detection of CRC during a colonoscopy was a factor contributing to non-adherence (Bleiker et al. 2005).

One non-adhering participant reasoned that because her mother was older than she was that this caused her to worry only about her mother, and not herself, developing cancer.

4.4. SOCIAL STIGMA OF CANCER

The researcher had not anticipated the social stigma of cancer to surface during the interviews. This issue must have been of particular importance to those who mentioned it, since it came unprompted by the researcher. However, it was therefore unknown whether stigma of cancer was apparent in the community of the participants who did not mention it in their interviews. For this reason, the researcher was only able to discuss this issue for the five participants who did.

Stigma means that the individual has an unwanted abnormality (in this case cancer) and is therefore disqualified from being fully socially accepted (Koller et al. 1996). The effects of stigma are emphasised during social interactions between stigmatised and unstigmatised people. This is due to the emotional responses that stigma induces in the unstigmatised individual. These responses are generally negative (anxiety, disgust, sadness, anger, or helplessness), but may also have positive aspects like empathy or overconcern. Both these types of responses reflect the attitude that the stigmatised person is unfavourably different from 'normal' people (Koller et al. 1996).

In this study, two attitudes towards stigma were apparent for the participants who mentioned it in their interviews. The first mind-set was that stigma was a negative trait of the community in which these individuals resided and was a result of the ignorance of people around cancer. According to one participant, the community believed that cancer was contagious. A pilot study was done on 900 respondents of the population of West Bengal to assess their level of awareness regarding cancer. A small proportion (21%) of the respondents expressed the vague idea that cancer was an infectious disease that was creating a problem of isolation from the family or society for cancer patients (Ray and Mandal 2004). As shown by Ray and Mandal's study, ignorance of cancer can lead to discrimination within the community against those with it.

The other dimension that was mentioned during the interviews and follow-up group discussion was how the stigma of cancer within the community created an awareness that was used to distinguish families with cancer from families without cancer. This phenomenon was clearly illustrated when some participants *proudly* informed the researcher that their specific clan name distinguished those with their surname who had the '*familie siekte*' – the familial colorectal cancer, from those who did not. The medical staff in the clinics were also aware of this. One participant recalled that doctors in Kimberly only referred him to GSH for specialised treatment of his symptoms when they realised he was a member of the clan, and this recognition probably saved his life.

One participant, however, found that there was no stigma in his community. He mentioned that he did not feel ashamed to tell people that he had had cancer and had his colon removed.

CHAPTER 5:
CONCLUDING REMARKS

CHAPTER 5

CONCLUDING REMARKS

5.1. SUMMARY

From this qualitative research project, it was determined that all participants were from socioeconomically disadvantaged backgrounds. Half of the group was unemployed and none had achieved a tertiary education. For one non-adhering participant such issues of poverty and family and social problems dominated her interview and there is a possibility that such issues are of more importance in her life than CRC screening, and thus could play a role in her non-adherence.

The findings indicate that among the non-adherent participants the preparation, the discomfort during the colonoscopy procedure, a family members' painful experience during a colonoscopy and the transport to the colonoscopy were the self-reported reasons for non-adherence. The groups' objective reasons for non-adherence were their misunderstanding of the reason for regular surveillance, not realising they were at risk of CRC and being unaware that they had received their genetic test result.

When adhering participants were asked directly why they had followed the recommended schedule for screening the most frequently stated reasons were to maintain their health and know their cancer status. The option of treatment or surgery and several deaths in the family from CRC were the other reasons given for adherence. All adherent participants understood the reason for attending regular surveillance and used their knowledge, whether on a basic or more advanced level, to rationalise their reasons for adherence.

Participants may be encouraged to go for regular screening by using stronger sedatives and acknowledging the discomfort experienced during the procedure.

5.2. CONCLUSION

The rapid development of our understanding of molecular genetics has created new possibilities not only to diagnose, but also to determine susceptibility to familial cancers. The transition from genetic research to clinical service opens up unique opportunities as well as unique psychosocial situations for the affected families (Chapman and Burn 1999). In a developing country like South Africa, a genetic service will not only be influenced by

psychosocial issues within a family, but the problems of poverty, low education level, unemployment and inaccessibility to services are also of primary concern.

Since 1994 the Division of Human Genetics at UCT has transformed research into a genetic service to families affected with HNPCC living in the Northern and Western Cape Province. The service has included offering predictive genetic testing to at risk family members and providing CRC screening in the form of colonoscopies and flexible sigmoidoscopies to individuals at risk. Taking such a service into the rural, impoverished areas of the Northern Cape has allowed these families to benefit from a service that would otherwise be unaffordable and inaccessible to them. However, offering a genetic service in a developing country where the reality of life is poverty, unemployment and low education levels adds complexity to the challenge of such intervention.

Providing these families the opportunity of genetic testing and surveillance comes with a responsibility to the genetics team and the families to whom these services are being offered. The genetics team must educate individuals at their level of understanding to empower them to take responsibility of their new-found knowledge and use it to maintain their health by attending CRC surveillance. It is only by this teamwork of the genetics team and the individuals it is offered to, that adherence rates can increase.

From this research project, it was evident that certain aspects of the colonoscopy procedure and family members' painful experience affected participants' adherence. However, the factors affecting adherence go far beyond the physicality of the screening. A complex mixture of psychological, attitudinal, cultural and socioeconomic aspects could also influence adherence behaviour. Thus a strategy to maximise surveillance will need to take the individual within the context of their family, community and culture into consideration. As previously stated, the ultimate goal of this research project together with a more comprehensive investigation of the whole HNPCC registry, would be to improve genetic counselling services in motivating, educating and supporting individuals who do not adhere to surveillance and thus to maximise screening adherence for those at high risk for CRC. However, as stated in a study by Bleiker: "Although 100% compliance with screening recommendations is probably not a realistic goal, any incremental increase in compliance rates will hopefully translate into reduced morbidity and mortality in this vulnerable population." (Bleiker et al. 2005). The same challenge, but compounded by the cultural diversity and socioeconomic issues of this developing country, is faced by the CRC team at the Southern-most tip of Africa.

5.3. RECOMMENDATIONS FOR THE GENETIC SERVICE

During the course of this research project the participants suggested ways in which to improve the screening for them and the researcher uncovered areas in the service that can be improved on and developed. The following are recommendations to improve the genetic service:

- Privacy after the procedure and stronger sedation if needed
- Six monthly follow-up with a genetic counsellor after the post-test result-giving session
- Constant follow-up for individuals not coping or showing misunderstanding
- Initiate a support group within the community where people are given the opportunity to talk about their concerns of CRC

5.4. RECOMMENDATIONS FOR FURTHER RESEARCH

One of the aims of the present study was to use selected information gained from this research to provide recommendations for a comprehensive investigation of mutation carriers' perception of factors affecting non-adherence to surveillance guidelines, to either prove or disprove the tentative factors of influence proposed by this study. This knowledge can then be generalised to a wider spectrum of non-adherent individuals. From the findings of this research project, the following areas are recommended for further research:

- To interview all non-adherent individuals in the Northern Cape who have received a positive predictive genetic result to determine the factors affecting non-adherence.

- To evaluate further the influence of social stigma of cancer on screening behaviour.

- To evaluate the participants' coping style. A study by Miller (1995) found that patients coped better (psychologically, behaviourally, and physiologically) when the information they received about their cancer diagnosis was individualised to their coping styles. It would be worthwhile to determine the effect coping style has on adherence to surveillance in a developing country like South Africa, where there is extensive poverty issues and cultural diversity.

- To explore the preventative health beliefs that either motivate or provide barriers to CRC screening for individuals at increased risk for developing CRC using the Health Belief

Model (HBM). The HBM suggests that the chance an individual will take a health-related action is determined by both the *individual's willingness* to take action and by their *perceived benefit* of the action compared to the *perceived barriers* involved in the proposed action. Willingness is controlled by both *perceived susceptibility* to a particular disease and *perceived severity* of the consequences of developing the disease. The action does not occur unless the individual believes in both the personal susceptibility and the serious repercussions of the disease (Jacobs 2002; Keller et al. 2002). Thus the following aspects should be taken into consideration for participants' belief for colonoscopies:

- psychological readiness to attend colonoscopies
- perceived benefit of colonoscopies (e.g. cancer control, treatment options)
- perceived barriers of colonoscopies (e.g. prep, discomfort)
- perceived susceptibility (e.g. low or high risk of CRC)
- perceived severity of CRC (e.g. surgery or death).

APPENDICES

APPENDIX A

ETHICAL CLEARANCE

ETHICAL CLEARANCE

UNIVERSITY OF CAPE TOWN

Research Ethics Committee
E53 Room 44.1,Old Main Building Groote
Schuur Hospital, Observatory, 7925
Queries : Xolile Fula
Tel : (021) 406-6492 Fax: 406-6411
E-mail : Xfula@curie.uct.ac.za

16 March 2005

REC REF: 393/2004

Prof RS Ramesar
Human Genetics

Dear Prof Ramesar

HEREDITARY NONPOLYPOSIS COLORECTAL CANCER: FACTORS CONTRIBUTING TO ADHERENCE AND NON-ADHERENCE TO SURVEILLANCE FOR MUTATION CARRIERS

Thank you for submitting your study to the Research Ethics Committee for review.

It is a pleasure to inform you that the Ethics Committee has formally approved the above-mentioned study on the 08 March 2005.

Please see attached comments and references provided by a reviewer which may be of assistance in your study.

Your comments to the queries are noted with thanks.

Please quote the REC. REF in all your correspondence

Yours sincerely

PROF T. ZABOW
CHAIRPERSON

Response to second review: 393/2004

Thank you for resubmitting this protocol.

I am satisfied with the changes made but wish to point out that the issue of reliability and validity in the qualitative paradigm has not really been sufficiently addressed. While this shouldn't hold up formal approval, I do wish to draw the researcher's attention to the manner in which these issues are handled in this qualitative paradigm.

Particularly when asking open-ended questions – the traditional concepts of reliability are hard to apply. The qualitative tradition refers to trustworthiness [this demonstrates the rigor of the study].

Secondly – a decision trail or audit trail must be evident, so that the researcher's action, influences on them, and events that occurred are documented (Holloway & Wheeler, 1996)

Guba and Lincoln refer to four alternatives to evaluate trustworthiness: credibility, transferability, dependability and confirmability.

Some options for further reading:

Guba, E. G. and Lincoln, Y (1989). Fourth generation evaluation. Newbury Park, Sage Publications.

Holloway, I. (1997). Basic Concepts for Qualitative Research. Oxford, Blackwell Science Ltd.

Holloway , I. and Wheeler, S. (1996) *Qualitative Research for Nurses*. Oxford Oxford, Blackwell Science.

Lincoln, Y and Guba, E. (1985). Understanding and doing naturalistic inquiry. Beverly Hills, Sage Publications.

APPENDIX B

INFORMATION SHEET

AFRIKAANS VERSION OF INFORMATION SHEET FOR PARTICIPANTS INVOLVED ONLY IN INTERVIEWS

DIE GENETIKA VAN OORERFLIKE NIE-POLIPOSE KOLOREKTALE KANKER IN SUID AFRIKA
Afdeling Mensgenetika, Fakulteit Gesondheidswetenskappe
Universiteit van Kaapstad

HOOFNAVORSER: Profesor Raj Ramesar Ph.D. (rr@cormack.uct.ac.za)
NAVORSINGSKÖORDINEERDER: Sr. Ursula Algar (ursula@curie.uct.ac.za)
Tel: 021 404 5499

M.Sc in Genetiese Raadgewing Navorsingsprojek
Oorerflike Nie-Polipose Kolorektale Kanker: Faktore wat Bydra tot Volhouding en Nie-Volhouding met die Waakprogram vir Mutasiedraers in Plattelandse Gebiede in die Noord- en Wes-Kaap

Inligtingsvorm

Hierdie studie word gedoen om te bepaal wat beïnvloed mense, wat positief getoets word vir die geen wat dikderm kanker veroorsaak, om vir of nie vir siftingsprogramme (e.g. kolonoskopie, sigmoidoskopie) te gaan nie. Deur aan hierdie studie deel te neem help u ons om party van hierdie redes te verstaan. Die kennis wat met hierdie studie verkry word sal ons help in die ondersteuning, voorligting en aanmoediging van persone om siftingsprogramme gereeld by te woon.

Gedurende die studie sal een onderhoud van omtrent 60-90 minute lank in April 2005 gedoen word. Die onderhoud sal op band opgeneem word sodat die navorser nie hoef te skryf gedurende die onderhoud nie.

Alle inligting wat gedurende die studie verkry word sal vertroulik bly. Die naam van die deelnemer sal nie op die onderhoudvorm wees nie, want 'n kode sal gebruik word om elke deelnemer te identifiseer. Slegs die hoofnavorser (Prof. Ramesar) sal toegang tot die kodes hê. Die onderhoud wat op band opgeneem word, sal binne een jaar vernietig word.

Deelname aan hierdie studie is heeltemal vrywillig. U kan enige tyd aan hierdie studie onttrek. Wat ook al u besluit is, sal nie u mediese behandeling of u verhouding met die mediese personeel beïnvloed nie.

Indien u enige vrae het oor die studie, voel asseblief vry om my (navorser) of Prof. Ramesar te kontak.

As u enige vrae het in verbanding met u reg as 'n deelnemer, voel asseblief vry om Prof. Tovia Zabow, die Voorsitter van Universiteit van Kaapstad Etiese Hersiening Komitee by 021 406 6492, te kontak.

Dankie vir u deelname.

Brenda Kruger (bkruger@cormack.uct.ac.za)
M.Sc. Genetiese Raadgewingstudent
Afdeling Mensgenetika
Universiteit van Kaapstad
021 406 6425

ENGLISH VERSION OF INFORMATION SHEET FOR PARTICIPANTS INVOLVED ONLY IN INTERVIEWS

THE GENETICS OF HEREDITARY NONPOLYPOSIS COLORECTAL CANCER IN SOUTH AFRICA
Division of Human Genetics, Faculty of Health Sciences
University of Cape Town

PRINCIPLE INVESTIGATOR: Professor Raj Ramesar Ph.D. (rr@cormack.uct.ac.za)
RESEARCH CO-ORDINATOR: Sr. Ursula Algar (ursula@curie.uct.ac.za)
Tel: 021 404 5499

M.Sc in Genetic Counselling Research Project

Hereditary Nonpolyposis Colorectal Cancer: Factors Contributing to Adherence and Non-adherence to Surveillance for Mutation Carriers in Rural Areas of the Northern and Western Cape

Information Sheet

This study is being done to find out why people, who are positive for the gene that causes colon cancer, go or do not go for screening (e.g. colonoscopy, sigmoidoscopy). By being in this study you are helping to understand some of these reasons. The knowledge we get from this study will help us in supporting, educating and encouraging people to go for regular screening.

During this study there will be one interview of about 60-90 minutes in April 2005. The interview will be tape recorded so that the researcher does not have to write during the interview.

All the information obtained during the study will remain confidential. The name of the participant will not appear on the interview sheet as a coding method will be used to identify each participant. Only the principal investigator (Prof. Ramesar) for this study will have access to the code. Within one year, the tapes will be destroyed.

Taking part in this study is entirely voluntary. You can withdraw from the study at any time. Whatever decision you make will not affect your medical treatment or your relationship with the medical staff.

Should you have any questions about the study, please feel free to contact me (researcher) or Prof Ramesar.

If you have any questions about your rights as a participant, please contact Prof. Tovia Zabow, the Chair of UCT Ethics Review Committee at 021 406 6492.

Thank you for your participation.

Brenda Kruger (bkruger@cormack.uct.ac.za)
M.Sc. Genetic Counselling Student
Division of Human Genetics
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AFRIKAANS VERSION OF INFORMATION SHEET FOR PARTICIPANTS IN INTERVIEW AND FOLLOW-UP GROUP DISCUSSION

DIE GENETIKA VAN OORERFLIKE NIE-POLIPOSE KOLOREKTALE KANKER IN SUID AFRIKA
Afdeling Mensgenetika, Fakulteit Gesondheidswetenskappe
Universiteit van Kaapstad

HOOFNAVORSER: Profesor Raj Ramesar Ph.D. (rr@cormack.uct.ac.za)
NAVORSINGSKÖORDINEERDER: Sr. Ursula Algar (ursula@curie.uct.ac.za)
Tel: 021 404 5499

M.Sc in Genetiese Raadgewing Navorsingsprojek

Oorerflike Nie-Polipose Kolorektale Kanker: Faktore wat Bydra tot Volhouding en Nie-Volhouding met die Waakprogram vir Mutasiedraers in Plattelandse Gebiede in die Noord- en Wes-Kaap

Inligtingsvorm

Hierdie studie word gedoen om te bepaal wat beïnvloed mense, wat positief getoets word vir die geen wat dikderm kanker veroorsaak, om vir of nie vir siftingsprogramme (e.g. kolonoskopie, sigmoidoskopie) te gaan nie. Deur aan hierdie studie deel te neem help u ons om party van hierdie redes te verstaan. Die kennis wat met hierdie studie verkry word sal ons help in die ondersteuning, voorligting en aanmoediging van persone om siftingsprogramme gereeld by te woon.

Gedurende die studie sal een onderhoud van omtrent 60-90 minute lank in April 2005 gedoen en 'n bespreking-groep in Julie 2005 gehou word. Die onderhoud en bespreking-groep sal op band opgeneem word sodat die navorser nie hoef te skryf gedurende die onderhoud nie.

Alle inligting wat gedurende die studie verkry word sal vertroulik bly. Die naam van die deelnemer sal nie op die onderhoudvorm wees nie, want 'n kode sal gebruik word om elke deelnemer te identifiseer. Slegs die hoofnavorser (Prof. Ramesar) sal toegang tot die kodes hê. Die onderhoud en bespreking-groep wat op band opgeneem word, sal binne een jaar vernietig word.

Deelname aan hierdie studie is heeltemal vrywillig. U kan enige tyd aan hierdie studie onttrek. Wat ook al u besluit is, sal nie u mediese behandeling of u verhouding met die mediese personeel beïnvloed nie.

Indien u enige vrae het oor die studie, voel asseblief vry om my (navorser) of Prof. Ramesar te kontak.

As u enige vrae het in verbanding met u reg as 'n deelnemer, voel asseblief vry om Prof. Tovia Zabow, die Voorsitter van Universiteit van Kaapstad Etiese Hersiening Komitee by 021 406 6492, te kontak.

Dankie vir u deelname.

Brenda Kruger (bkruger@cormack.uct.ac.za)
M.Sc. Genetiese Raadgewingstudent
Afdeling Mensgenetika
Universiteit van Kaapstad
021 406 6425

ENGLISH VERSION OF INFORMATION SHEET FOR PARTICIPANTS IN INTERVIEW AND FOLLOW-UP GROUP DISCUSSION

THE GENETICS OF HEREDITARY NONPOLYPOSIS COLORECTAL CANCER IN SOUTH AFRICA
Division of Human Genetics, Faculty of Health Sciences
University of Cape Town

PRINCIPLE INVESTIGATOR: Professor Raj Ramesar Ph.D. (rr@cormack.uct.ac.za)
RESEARCH CO-ORDINATOR: Sr. Ursula Algar (ursula@curie.uct.ac.za)
Tel: 021 404 5499

M.Sc in Genetic Counselling Research Project

Hereditary Nonpolyposis Colorectal Cancer: Factors Contributing to Adherence and Non-adherence to Surveillance for Mutation Carriers in Rural Areas of the Northern and Western Cape

Information Sheet

This study is being done to find out why people, who are positive for the gene that causes colon cancer, go or do not to go for screening (e.g. colonoscopy, sigmoidoscopy). By being in this study you are helping to understand some of these reasons. The knowledge we get from this study will help us in supporting, educating and encouraging people to go for regular screening.

During this study there will be one interview of about 60-90 minutes in April 2005 and a group discussion in July 2005. The interview and discussion-group will be tape recorded so that the researcher does not have to write during the interview.

All the information obtained during the study will remain confidential. The name of the participant will not appear on the interview sheet as a coding method will be used to identify each participant. Only the principal investigator (Prof. Ramesar) for this study will have access to the code. Within one year, the tapes will be destroyed.

Taking part in this study is entirely voluntary. You can withdraw from the study at any time. Whatever decision you make will not affect your medical treatment or your relationship with the medical staff.

Should you have any questions about the study, please feel free to contact me (researcher) or Prof. Ramesar.

If you have any questions about your rights as a participant, please contact Prof. Tovia Zabow, the Chair of UCT Ethics Review Committee at 021 406 6492.

Thank you for your participation.

Brenda Kruger (bkruger@cormack.uct.ac.za)
M.Sc. Genetic Counselling Student
Division of Human Genetics
University of Cape Town
021 406 6425

APPENDIX C

INFORMED CONSENT FORM

AFRIKAANS VERSION OF INFORMED CONSENT FORM FOR PARTICIPANTS INVOLVED ONLY IN THE INTERVIEWS

DIE GENETIKA VAN OORERFLIKE NIE-POLIPOSE KOLOREKTALE KANKER IN SUID AFRIKA
Afdeling Mensgenetika, Fakulteit Gesondheidswetenskappe
Universiteit van Kaapstad

HOOFNAVORSER: Professor Raj Ramesar Ph.D. (rr@cormack.uct.ac.za)
NAVORSINGSKÖORDINEERDER: Sr. Ursula Algar (ursula@curie.uct.ac.za)
Tel: 021 404 5499
ETIESE KOMITEE: Prof. Tovia Zabow (021 406 6492)

M.Sc in Genetiese Raadgewing: Navorsingsprojek

Oorerflike Nie-Polipose Kolorektale Kanker: Faktore wat Bydra tot Volhouding en Nie-Volhouding met die Waakprogram vir Mutasiedraers in Plattelandse Gebiede in die Noord- en Wes-Kaap

Toestemmingsvorm

Naam van Deelnemer:.....
Adres:.....
.....
.....
Tel. Nr:.....
Ondernemer Nr:.....

Ek, ..., het die aangehegte deelnemer inligtingsvorm gelees en ek verstaan die doel en aard van die studie.

Ek is bewus dat die navorser en 'n vertaler gedurende die onderhoud teenwoordig sal wees. Ek is bewus daarvan dat my privaatheid gehandhaaf sal word en dat alle inligting wat verkry word vertroulik sal bly.

Ek verstaan dat my deelname aan die studie vrywillig is en dat ek enige tyd aan hierdie studie mag onttrek. Wat ook al my besluite is, dit sal nie my mediese behandeling of my verhouding met die mediese personeel beïnvloed nie.

Handtekening: _____

Datum: _____

Ek verstaan dat die onderhoud op band opgeneem sal word.

Handtekening: _____

Datum: _____

Brenda Kruger (bkruger@cormack.uct.ac.za)
M.Sc. Genetiese Raadgewingstudent
Afdeling Mensgenetika
Universiteit van Kaapstad (021 406 6425)

ENGLISH VERSION OF INFORMED CONSENT FORM FOR PARTICIPANTS INVOLVED ONLY IN THE INTERVIEWS

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RESEARCH CO-ORDINATOR: Sr. Ursula Algar (ursula@curie.uct.ac.za)
Tel: 021 404 5499
ETHICS COMMITTEE: Mr. Xolile Fula (021 406 6492)

M.Sc in Genetic Counselling Research Project

Hereditary Nonpolyposis Colorectal Cancer: Factors Contributing to Adherence and Non-adherence to Surveillance for Mutation Carriers in Rural Areas of the Northern and Western Cape

Consent Form

Participant Name:.....
Address:.....
.....
.....
Tel. No:.....
Participant No:.....

I, ..., have read the attached participant information sheet and I understand the purpose and nature of this study.

I am aware that the researcher and a translator will be present during the interview. I am aware that my privacy will be maintained and that all information obtained will remain confidential.

I understand that my participation in this study is voluntary and that I may withdraw at any stage. Whatever my decision is will not affect my medical treatment or your relationship with the medical staff in any way.

Signature: _____

Date: _____

I understand that the interview will be tape recorded so that the researcher is able to recall the experience accurately.

Signature: _____

Date: _____

Brenda Kruger (bkruger@cormack.uct.ac.za)
MSc. Genetic Counselling Student
Division of Human Genetics
University of Cape Town (021 406 6425)

AFRIKAANS VERSION OF INFORMED CONSENT FORM FOR PARTICIPANTS IN INTERVIEW AND FOLLOW-UP GROUP DISCUSSION

DIE GENETIKA VAN OORERFLIKE NIE-POLIPOSE KOLOREKTALE KANKER IN SUID AFRIKA
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ETIESE KOMITEE: Prof. Tovia Zabow (021 406 6492)

M.Sc in Genetiese Raadgewing: Navorsingsprojek

Oorerflike Nie-Polipose Kolorektale Kanker: Faktore wat Bydra tot Volhouding en Nie-Volhouding met die Waakprogram vir Mutasiedraers in Plattelandse Gebiede in die Noord- en Wes-Kaap

Toestemmingsvorm

Naam van Deelnemer:.....
Adres:.....
.....
.....
Tel. Nr:.....
Ondernemer Nr:.....

Ek, ..., het die aangehegte deelnemer inligtingsvorm gelees en ek verstaan die doel en aard van die studie.

Ek is bewus dat die navorser en 'n vertaler gedurende die onderhoud en bespreking-groep teenwoordig sal wees. Ek is bewus daarvan dat my privaatheid gehandhaaf sal word en dat alle inligting wat verkry word vertroulik sal bly.

Ek verstaan dat my deelname aan die studie vrywillig is, en dat ek enige tyd aan hierdie studie mag onttrek. Wat ook al my besluite is, dit sal nie my mediese behandeling of my verhouding met die mediese personeel sal beïnvloed nie.

Handtekening: _____

Datum: _____

Ek verstaan dat die onderhoud en bespreking-groep op band opgeneem sal word.

Handtekening: _____

Datum: _____

Brenda Kruger (bkruger@cormack.uct.ac.za)
M.Sc. Genetiese Raadgewingstudent
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021 406 6425

ENGLISH VERSION OF INFORMED CONSENT FORM FOR PARTICIPANTS IN INTERVIEW AND FOLLOW-UP GROUP DISCUSSION

THE GENETICS OF HEREDITARY NONPOLYPOSIS COLORECTAL CANCER IN SOUTH AFRICA
Division of Human Genetics, Faculty of Health Sciences
University of Cape Town

PRINCIPLE INVESTIGATOR: Professor Raj Ramesar Ph.D. (rr@cormack.uct.ac.za)
RESEARCH CO-ORDINATOR: Sr. Ursula Algar (ursula@curie.uct.ac.za)
Tel: 021 404 5499
ETHICS COMMITTEE: Mr. Xolile Fula (021 406 6492)

M.Sc in Genetic Counselling Research Project

Hereditary Nonpolyposis Colorectal Cancer: Factors Contributing to Adherence and Non-adherence to Surveillance for Mutation Carriers in Rural Areas of the Northern and Western Cape

Consent Form

Participant Name:.....
Address:.....
.....
.....
Tel. No:.....
Participant No:.....

I, ..., have read the attached participant information sheet and I understand the purpose and nature of this study.

I am aware that the researcher and a translator will be present during the interview discussion-group. I am aware that my privacy will be maintained and that all information obtained will remain confidential.

I understand that my participation in this study is voluntary and that I may withdraw at any stage. Whatever my decision is will not affect my medical treatment or your relationship with the medical staff in any way.

Signature: _____

Date: _____

I understand that the interview and discussion-group will be tape recorded so that the researcher is able to recall the experience accurately.

Signature: _____

Date: _____

Brenda Kruger (bkruger@cormack.uct.ac.za)
MSc. Genetic Counselling Student
Division of Human Genetics
University of Cape Town (021 406 6425)

APPENDIX D

QUALITATIVE INTERVIEW GUIDE

AFRIKAANS VERSION OF THE INTERVIEW GUIDE

Ondernemer Nr:.....

A. Sosiodemografiese en Sosioekonomiese Inligting

1. Wat is u geboortedatum?

2. Is u getroud?

- Enkellopend
- Weduwee/wewenaar
- Geskei
- Vervreemd maar nie geskei nie
- Getroud
- Woon saam met vriend/vriendin/kêrel

3. Het u kinders?

- Ja
- Nee

4. INDIEN JA VIR VRAAG 3: Hoeveel kinders het u?

5. Wat is die geboortedatums van u kinders?

6. Hoeveel persone woon saam met u?

7. Wat is u verwantskap met die persone en hoe oud is hulle?

8. Wat is die hoogste graad/standerd wat u op skool voltooi het?

- Graad 12 (matriek, St. 10)
- Graad 11 (St. 9)
- Graad 10 (St. 8)
- Graad 9 (St. 7)
- Graad 8 (St 6)
- Graad 7 (St 5)
- Ander

9. Wat is die hoogste kwalifikasie wat u na skool gekry het?

- Geen
- Ambag / Vakleerling
- Kollege sertifikaat
- Diploma (na Graad 12)
- Universiteitsgraad
- Na-graadse diploma / -graad
- Ander

10. Werk u?

- Werk voltyds
- Werk deeltyds Watter soort werk?.....
- Eie besigheid
- Werkloos
- Huisvrou
- Voltydse student
- Deeltydse student
- Permanent onbevoeg om te werk
- Afgetree or pensionaris
- Ander (spesifiseer)

11. Wie in die huis werk en watter soort werk doen hulle?

12. Word die inkomste van alle persone wat in die huis woon saamgevoeg om te betaal vir lewenskoste?

- Ja
- Nee

13. Vir hoeveel mense versorg u met die inkomste?

14. Wat is u huidige inkomste per maand (en van alle persone wat saam met u in die huis woon)?

- Geen inkomste
- Onbevoegd vir werk / Ongeskiktheids toelae (Watter tipe = , hoeveel R)
- R1 - R400
- R401 - R800
- R801 - R1600
- R1601 - R3200
- R3201 - R6400
- R6401 - R12 800
- R12 801 - R25 600
- R25 601 - R51 200
- R52 201 - R102 400
- R102 401 - R204 800
- Meer as R204 801

15. Watter dag-hospitaal gaan u na toe wanneer u siek word?

16. Hoe gereeld gaan u na die kliniek?

17. Hoe ver is dit van u huis af?

18. Hoe lank neem dit u om daar te kom?

19. Hoe kom u by die kliniek? (vervoer)

20. Hoeveel kos u vervoer na en van die kliniek?

21. Het u 'n mediesefonds?

- Ja
- Nee – Hoe betaal u vir kolonoskopies?

22. Betaal jou mediesefonds vir kolonoskopies?

- Ja
- Nee – Hoe betaal u vir kolonoskopies?

23. Hoeveel kos 'n kolonoskopie vir u?

24. Waar gaan u vir kolonoskopies?

25. Hoe lank neem dit u om daar te kom?

26. Hoeveel kos u vervoer na en van die hospital vir die doen van 'n kolonoskopie?

B. Begrip van dikderm kanker

27. Is u bloed reeds getoets vir die geen wat dikderm kanker in u familie veroorsaak?

- Ja
- Nee (gaan na vraag 30)
- Ek weet nie (gaan na vraag 30)

28. INDIEN **JA** VIR VRAAG 27: Wanneer het u die resultate gekry?

29. Het u die geen (oorerflike materiaal) wat dikderm kanker in u familie versoorsaak?

- Ja
- Nee (gaan na vraag 31)
- Ek weet nie (gaan na vraag 31)

30. INDIEN **NEE OF EK WEET NIE** VIR VRAAG 27: Die navorser sal verduidelik dat die deelnemer reeds getoets is en sal die redes verduidelik waarom hy/sy dit nie verstaan nie. Dit sal op 'n ondersteunende wyse gedoen word. Die navorser sal die deelnemer verder op 'n ondersteunende wyse inlig oor sy/haar genetiese status.

31. INDIEN **NEE OF EK WEET NIE** VIR VRAAG 29: Die navorser sal die rede verduidelik waarom die deelnemer die antwoord gegee het. Die navorser sal die deelnemer verder op 'n ondersteunende wyse inlig oor sy/haar genetiese status.

32. Is u gelukkig om met die onderhoud voort te gaan?
(Dit sal die deelnemer in staat stel om toestemming te gee om voort te gaan met die onderhoud of om dit tot later uit te stel.)

33. Hoe voel u oor die inligting waaroor ons pas gesels het?

34. Aangesien u die geen het wat dikderm kanker veroorsaak, wat moet u doen om seker te maak dat u gesond bly?

35. Beskryf hoe u voel oor die feit dat u die geen vir dikderm kanker het?

36. Verduidelik hoe die resultaat u lewe beïnvloed het op emosionele en mediese vlak?

37. Het u u resultaat met enige iemand gedeel?

- Ja (Wie?)
- Nee

38. Wat was u rede?

39. Hoe het hulle gereageer op u resultaat?

40. Wie gee aan u die meeste ondersteuning in verband met dikderm kanker risiko?

41. Wat doen hulle om u te ondersteun?

C. Familiegeskiedenis van dikderm kanker

42. Wanneer het u vir die eerste keer bewus geword daarvan dat daar dikderm kanker in u familie is?

43. Hoe het dit u laat voel?

44. Wanneer het u vir die eerste keer bewus geword daarvan dat u 'n risiko het om dikderm kanker te kry?

45. Hoe het dit u laat voel?

46. Wie van u familielede het:

- 'n Hoe risiko om dikderm kanker te kry
- Dikderm kanker of het dit gehad
- Gesterf as gevolg van dikderm kanker

47. Hoe het u hierdie verliese in u lewe hanteer?

D. Kolonoskopie ervaring

48. Het u al ooit 'n kolonoskopie gehad?

- Ja (Hoeveel?)
- Nee
- Ek weet nie

49. Wanneer het u die laaste kolonoskopie gehad?

Maand..... Jaar.....

50. Wat is die redes waarom u vir die kolonoskopie(s) gegaan het? *(for adherent participants only)*

OR

50. (a) Wanneer het u NIE vir die koloskopie(s) gegaan NIE?

(b) Wat is die redes waarom u NIE vir die kolonoskopie(s) gegaan het NIE? *(for non-adherent participants)*

51. Dink u dit is nodig om vir kolonoskopies te gaan?

52. Hoe gereeld moet u vir 'n kolonoskopie gaan?

53. Vind u dit is nodig om gereeld vir kolonoskopies te gaan?

54. Wie het gesê dat u vir 'n kolonoskopie moet gaan?

55. Wat was hulle rede waarom u vir 'n kolonoskopie moet gaan?

56. Laat ons praat oor wat gebeur en hoe u voel gedurende 'n kolonoskopie, van die voorbereiding tot na die kolonoskopie?

57. Gaan jou familie lede met dikderm kanker of met 'n hoe risiko vir dikderm kanker gereeld vir kolonoskopies?

58. Het dit u beïnvloed om vir kolonoskopies te gaan?

59. Was daar al 'n poliep tydens 'n kolonoskopie gevind?

- Ja
- Nee
- Ek weet nie

60. Was daar al 'n kankerweefsel tydens 'n kolonoskopie gevind?

- Ja
- Nee
- Ek weet nie

61. INDIEN **JA** VIR VRAAG 59 EN/OF 60: Wat het gebeur na die poliep of kankerweefsel gevind is?

INDIEN **NEE** VIR VRAAG 59 EN/OF 60:

62. Hoe dink u sou u gevoel het indien 'n poliep of kanker tydens 'n kolonoskopie gevind is?

63. Hoe dink u sou u gevoel het indien 'n poliep of kanker NIE tydens 'n kolonoskopie gevind is NIE?

64. Hoe dink u sal u dit hanteer as u kanker (weer) ontwikkel?

65. Hoe bang is u om dikderm kanker (weer) te kry:

66. Hoekom?

E. Bekommernisse oor kolonoskopie

67. Is daar goed wat u bekommerd maak as u vir 'n kolonoskopie gaan? (bespreek)

68. Beskryf hoe u voel voor u vir 'n kolonoskopie ondersoek gaan?

69. Hoe angstig voel u VOOR, GEDURENDE en NA 'n kolonoskopie?

70. Vind u 'n kolonoskopie:

- Ongemaklik
- Pynlik
- 'n Indringende prosedure
- Skaam
- Maak jou bang

(en Hoekom?)

71. Wat is die positiewe redes om vir 'n kolonoskopie te gaan?

72. Wat is die slegste dinge om vir 'n kolonoskopie te gaan?

73. Wat sal die kolonoskopie beter/makliker vir u maak?

F. Opinie oor dikderm kanker diens

74. Hoe behulpsaam was die ondersteuningsdiens van die dikderm kanker-span?

75. Wat was van hulp gewees?

76. Wat was nie van hulp nie?

77. Is daar enigiets wat u sal help wanneer u vir kolonoskopies gaan?

G. Terugvoer

78. Het u enige vrae wat u wil vra?

79. Hoe voel u nou na die onderhoud?

H. Mening oor die Onderhoud

80. Hoe het u die onderhoud ervaar (u gevoelens, meer inligting nodig)?

81. Sal u daarvan hou om inligting oor die navorsingsresultate te ontvang?

82. INDIEN JA VIR VRAAG 81: Verkies u dat ek die navorsingsresultate aan u pos of moet ek u daaroor bel?

- Die navorser het die deelnemer bedank vir die tyd wat dit geneem het om die onderhoud te voer en sal kontak besonderhede aan die deelnemer verskaf indien verdere kontak benodig word.

I. Inligtingsessie

- Die navorser het ondersteunende en opvoedkundige insette gelewer na die onderhoud om emosionele bekommernisse en feite rondom dikderm kanker en kolonoskopie op te klaar. Dit is met behulp van visuelehulpmiddels verduidelik.

- Die navorser het die deelnemer vir genetiese raadgewing verwys indien nodig.

ENGLISH VERSION OF THE INTERVIEW GUIDE

Participant No:.....

A. Sociodemographic and Socioeconomic Information

1. What is your date of birth?

2. Are you married?

- Single
- Widowed
- Divorced
- Separated but not divorced
- Married
- Living with partner

3. Do you have any children?

- Yes
- No

4. IF **YES** TO QUESTION 3: How many children do you have?

5. What are the dates of birth of your children?

6. How many people do you live with?

7. What is your relationship to them and how old are they?

8. Until which grade/standard did you complete at school?

- Grade 12 (matric, Std 10)
- Grade 11
- Grade 10
- Grade 9
- Grade 8
- Grade 7
- Other

9. What is the highest qualification you have obtained since leaving school?

- No post-school
- Trade / apprentice
- Certificate from college
- Diploma (beyond Grade 12)
- Bachelors degree
- Postgraduate diploma / degree
- Other

10. Do you work?

- Full-time employed
- Part-time employed What sort of work?.....
- Self-employed
- Unemployed
- Housewife
- Full-time student
- Part-time student
- Permanently unable to work
- Retired or pensioner
- Other

11. Who in the house works and what do they do?

12. Is the household income pooled together to pay for living?

- Yes
- No

13. How many people does the income support?

14. What is your current (and household) income per month?

- No income
- Disability grant (what type = how much = R)
- R1 - R400
- R401 - R800
- R801 - R1600
- R1601 - R3200
- R3201 - R6400
- R6401 - R12 800
- R12 801 - R25 600
- R25 601 - R51 200
- R52 201 - R102 400
- R102 401 - R204 800
- More than R204 801

15. Which day hospital do you go to when you are sick?

16. How often do you go to the clinic?

17. How far is it from your house?

18. How long does it take to get there?

19. How do you get to the clinic?

20. How much does it cost to get there and back?

21. Do you have a medical aid?

- Yes
- No – How do you pay for colonoscopies?

22. Does your medical aid cover for colonoscopies?

- Yes
- No – How do you pay for colonoscopies?

23. How much does a colonoscopy cost for you?

24. Where do you have colonoscopies?

25. How long does it take for you to get there?

26. How much does your transport cost to get to and from the hospital for a colonoscopy?

B. Understanding of colon cancer status

27. Has your blood been tested for the gene that causes colon cancer in your family?

- Yes
- No (go to question 30)
- I don't know (go to question 30)

28. IF **YES** TO QUESTION 27: When did you receive your result?

29. Do you have the gene that causes colon cancer in your family?

- Yes
- No (go to question 31)
- I don't know (go to question 31)

30. IF **NO OR I DON'T KNOW** TO QUESTION 27: The researcher will explain that they did have the test at a certain date and explore their reasons for not understanding this in a supportive way. The researcher will go on to inform them of their genetic status in a supportive manner

31. IF **NO OR I DON'T KNOW** TO QUESTION 29: The researcher will explore with the participant their reason for their answer. The researcher will go on to inform them of their genetic status in a supportive manner.

32. Are you happy to go on with the interview?
(This will allow the participant to give consent to carry on with the interview or delay it to another day.)

33. How are you feeling about what we have just talked about?

34. Since you have the gene that cause colon cancer, what must you do to make sure you stay healthy?

35. Describe how you feel about your result for testing positive for colon cancer?

36. Explain how the result has affected your life (emotionally and medically)?

37. Have you shared this result with anyone?

- Yes (Whom?)
- No

38. What was your reason for doing so?

39. How have they responded to your result?

40. Who gives you the most support around colon cancer risk?

41. What do they do for you that is supportive?

C. Family History of colon cancer

42. When did you first realise colon cancer was in your family?

43. How did this make you feel?

44. When did you first realise that you were at risk of getting colon cancer?

45. How did this make you feel?

46. Which family members have:

- A high risk for getting colon cancer
- Colon cancer
- Have died of colon cancer

47. Explain how you have dealt or coped with these losses?

D. Colonoscopy experience

48. Have you ever had a colonoscopy?

- Yes (How many?)
- No
- I don't know

49. When was your last colonoscopy? Month..... Year.....

50. What are your reasons for going for the colonoscopy (ies)? *(for adherent participants)*

OR

50. (a) When did you NOT go for the colonoscopy (ies)?
(b) What are the reasons why you did NOT go for the colonoscopy (ies)
(for non-adherent participants)

51. Do you think it is necessary to go for a colonoscopy?

52. How often should you go for a colonoscopy?

53. Do you think it is necessary to go often for colonoscopies?

54. Who recommended you go for a colonoscopy?

55. What was their reason for sending you for a colonoscopy?

56. Let's talk about what happens and how you feel during a colonoscopy, from the prep till after the colonoscopy?

57. Do other family members with colon cancer or at high risk go for colonoscopies regularly?

58. Has this influenced you going for colonoscopies?

59. Has a polyp been found during a colonoscopy?

- Yes
- No
- I don't know

60. Has a cancer been found during a colonoscopy?

- Yes
- No
- I don't know

61. IF **YES** TO QUESTION 59 and/or 60: What happened after the polyp or cancer was found?

IF **NO** TO QUESTION 59 and/or 60:

62. How do you think you would you feel if a polyp or cancer was found during a colonoscopy?

63. How do you think you would you feel if a polyp or cancer was NOT found during a colonoscopy?

64. How do you think you would cope if you got cancer? OR How have you coped with having cancer?

65. How fearful are you of getting colon cancer (again):

66. Why?

E. Concerns about colonoscopy

67. Are there things that worry you about going for a colonoscopy?

68. Describe how you feel before going for a colonoscopy?

69. How anxious do you feel BEFORE, DURING and AFTER a colonoscopy?

70. Do you find a colonoscopy:

- Uncomfortable
- Painful
- An invasive procedure Why?
- Embarrassing
- Makes you scared

71. What are the positive things about going for a colonoscopy?

72. What are the worst things about going for a colonoscopy?

73. What would make a colonoscopy better or easier for you?

F. Opinion about colon cancer service

74. How helpful has the support service from our colon cancer team been?

75. What has been helpful?

76. What has not been helpful?

77. Is there anything that would help you when you go for colonoscopies?

G. Debrief

78. Do you have any further comments you would like to share or ask any questions?

79. How are you feeling after the interview?

H. Opinion of Interview

80. How has the interview been for you (feelings, more information)?

81. Would you like to be informed of the research findings?

82. IF **YES** TO QUESTION 81: Would you prefer me to post or phone you with the research findings?

- The researcher thanked the participant for their time in allowing them to do the interview and provided contact details for participants who required further contact.

I. Information Session

- The researcher provided supportive and educational input for the participant after the interview to revisit relevant emotional concerns and clarify facts on colon cancer and colonoscopy using visual aids.

- The researcher referred the participant for genetic counselling if needed.

APPENDIX E

FOLLOW-UP GROUP DISCUSSION GUIDE

AFRIKAANS VERSION OF THE FOLLOW-UP GROUP DISCUSSION GUIDE

1. Wat is die redes waarom jy nie vir die kolonoskopies wil gaan nie?

2. Wat moet verander sodat jy vir die kolonoskopies sal gaan?

3. Wat is die redes waarom jy vir die kolonoskopies wil gaan?

4. Hoekom het die dokters julle aanbeveel om vir die kolonoskopies te gaan?

5. In almal se eie woorde, wil ek weet wat julle verstaan van die bloedtoets wat julle gehad het wat kyk of julle die geen het wat die dikderm kanker in die familie veroorsaak?

ENGLISH VERSION OF THE FOLLOW-UP GROUP DISCUSSION GUIDE

1. What are the reasons why you do not want to go for colonoscopies?

2. What must change for you to go for colonoscopies?

3. What are the reasons why you will go for colonoscopies?

4. Why did the doctors recommend that you go for colonoscopies?

5. In you own words, what do you understand of the blood test that you had to see if you had the gene that causes the colon cancer in you family?

APPENDIX F

SOCIODEMOGRAPHIC AND SOCIOECONOMIC INFORMATION FOR EACH PARTICIPANT

Table 11. Sociodemographic information for each participant in the study (P1 – P8).

	P1	**P2**	**P3**	**P4**	**P5**	**P6**	**P7**	**P8**
Sex	Female	Female	Male	Female	Female	Male	Female	Male
Age (years)	34	41	38	33	45	34	46	38
Town address	Okiep	Port Nolloth	Komaggas (in jail)	Nababeep	Port Nolloth	Komaggas	Komaggas	Komaggas
Marital status	Divorced	Divorced	Divorced	Married	Separated	Married	Married	Married
Have children	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
- No. of children	Two	One	Two	Two	One	One	Three	Three
- Children's age (years)	11, 17	19	5, 13	8, 12	16	Did not mention	18, 23, 25	3, 5, did not mention
Education								
- School grade	8	7	9	6	10	12	8	5
- Highest qualification after school	None	None	None	None	None	None	None	None
Employment status	Unemployed	Unemployed	In jail	Unemployed	Church work	Plant operator (De Beers)	Housewife	Plant operator (De Beers)
Current household income per month	R1 460	Did not know	In jail	R5 000	Did not know	R8 000	R2 000	R10 000

Table 12. Socioeconomic information of each participant in the study (P1 – P8)[a].

	P1	**P2**	**P4**	**P5**	**P6**	**P7**	**P8**
No. people who work in the house	2	3	1	3	2	2	2
Relationship to people who work in house	Mom, sister	Niece, nephew	Husband	Niece, nephew	Wife	Husband, son	Wife
Household income pooled together	Yes	Yes	N/A	Yes	Yes	Yes	Yes
No. of people income supports	6	7	4	7	3	6	4

[a]Not applicable to P3 who was in jail.

APPENDIX G

COST AND TRANSPORT TO COLONOSCOPIES FOR EACH PARTICIPANT

Table 13. Location and costs of colonoscopies and transport for each participant (P1 – P8).

	P1	**P2**	**P3**	**P4**	**P5**	**P6**	**P7**	**P8**
Cost of colonoscopies	None	None	None	None	None	None	None	None
Hospital it takes place at	Okiep/ Springbok	Kleinsee	Nababeep/ Springbok	Nababeep/ Springbok Nababeep	Kleinsee/ Springbok/	Kleinsee	Kleinsee	Kleinsee
Time to get there	5-10 minutes	1-1 ½ hours	Did not mention	Unsure	1-1 ½ hours	25 minutes	1 hour	45 minutes
Transport	Walks/ ambulance	Kombi	Bus	Walks/ bus	Ambulance/ bus	Ambulance	Bus	Bus
Transport costs	None	None	Yes	None	None	None	None	Unsure

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REFERENCES

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