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Communication Study

Communicating cancer risk within an African context: Experiences, disclosure patterns and uptake rates following genetic testing for Lynch syndrome

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ABSTRACT

Objective: Data pertaining to Lynch syndrome within a developing country are sparse. This study explored the emotional reaction to a mutation-positive test result among a group of individuals from South Africa. As genetic information is not only limited to the individual but extends to the biological family, communication patterns and uptake of testing among at-risk family members was also investigated.
Methods: Eighty individuals participated in this qualitative interview study.
Results: Eight emotional reactions were observed, of which two were of particular concern: (1) secrecy due to disbelief and (2) interpretation of a mutation-positive result as a cancer diagnosis. Disclosure rates of personal genetic test results were high to family members, but low to general healthcare providers. Disclosing the test result was not always followed by a discussion of implications of the genetic information or availability of predictive testing for at-risk family members. The uptake rate of predictive testing among the participants' siblings and children was 97% and 73.6%, respectively.
Conclusion: Awareness of concerning emotional reactions following the delivery of a genetic test result and insight into disclosure patterns, especially the information that is not communicated, will prove beneficial in improving the effectiveness of counselling and management in Lynch syndrome families.
Practice implications: Implementation of these findings into the PT programme will have a positive effect on the genetic counseling process.

1. Introduction

Lynch syndrome (LS) is characterised by autosomal dominant inheritance, predisposing individuals to a high risk (80–85%) of early-onset colorectal cancer (CRC) as well as other extracolonic tumours [1,2]. The identification of several genes predisposing to LS has enabled predictive testing (PT) to be offered to South African (SA) families with a high-risk for the condition, since 1993/1994 [3]. Mutation-positive individuals are enrolled into surveillance services either at a tertiary centre or for those that are located in remote areas an outreach endoscopic programme runs on an annual basis (described in greater detail in Bruwer et al. [4]. The dichotic outcome of PT enables (1) the identification of individuals with a high-risk (mutation-positive result) to promote preventative strategies, thereby reducing the incidence of and mortality from CRC, and (2) for those with a low-risk (mutation-negative result), to alleviate unnecessary concern for themselves and their offspring and subsequent discharge from intensive screening [5,6].

However, once a disease causing mutation has been identified in an individual, the result not only affects the proband, but extends to his or her biological family. Informing relatives about the identification of a pathogenic mutation allows unaffected family members the opportunity to ascertain their cancer-risk through mutation-specific testing and to determine if they require high-risk cancer screening [7]. Consequently, the dissemination of cancer-risk information and subsequent access to genetic counselling and testing services among relatives depends, partly, on whether or not the proband discusses the test result with family members (and on how accurate the understanding of this information is). Timely disclosure of the health information is also vital to ensure that at-risk family members have access to genetic testing and counselling and, if required, endoscopic surveillance prior to the development of a preventable cancer.

The aim of the present study was to elucidate how mutation-positive individuals reacted to the news of their increased risk of developing CRC and how and when this information was communicated to their family. Furthermore, the subsequent uptake of PT among all siblings and children of the mutation-positive individual are provided.

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2. Methodology

2.1. Participants

Purposeful sampling was used to recruit individuals for qualitative interviews to uncover meaning and generate understanding of the emotional effect of receiving a mutation-positive test result and the subsequent communication of the familial implications of genetic testing in SA families with LS. The uptake rate of PT among the participant's siblings and eligible children (children over the age of 18 years) was also determined. A cross-sectional design was used and eligible participants were (a) 18 years or older and (b) mutation-positive. The colorectal cancer genetic co-ordinator, involved in the service for more than a decade, identified 'information-rich' cases from the list of 180 individuals meeting the inclusion criteria. For purposes of confidentiality and privacy, these individuals were first contacted by the colorectal cancer genetic co-ordinator and the purpose of the study was explained prior to enrollment. Participation was voluntary and the contact details of those individuals agreeing to participate in the study were then provided to the researcher (ZB). The researcher travelled to the participant's homes or venues of their choice to conduct the interviews. Ethical approval for the study was obtained from the Research Ethics Committee of the University of Cape Town (Rec/Ref: 213/2009) (see Fig. 1).

2.2. Data collection

Semi-structured interviewing including both open- and closed-ended questions was used as the measuring tool. Questions were based on those covered in validated questionnaires and from study-specific questions relating to family communication sourced from the literature [7–15]. Closed-ended questions were used to collect demographic data and 'yes/no' or pre-categorised scales of various items. Frequency tables were used to categorise responses into descriptive statistics such as percentages and means. For each open-ended question, neutral probes were listed to clarify responses if they were incomplete or inaccurately understood.

A detailed three-generation pedigree was drawn for each participant. The family history provided information about the names and number of siblings and children of the participant and were be used to calculate uptake rate among these family members. The names of each of these family members were searched against the records of the database/registry to confirm that genetic testing had occurred.

The questions were pilot-tested on a small sample (two individuals) and questions were investigated for ease of comprehension, avoidance of ambiguity and to ensure that answers elicited the type of information envisaged [16–18]. The researcher conducted all interviews personally in the preferred language of the participant during the months of June 2009 and April 2011. The interviews took approximately one hour to complete and were recorded to preserve the data on tape, allowing for the analysis to be completed subsequent to interviewing. All interviews were transcribed verbatim and written consent was obtained for interviewing, recording and transcription of the data.

The interview schedule appears in Appendix A.

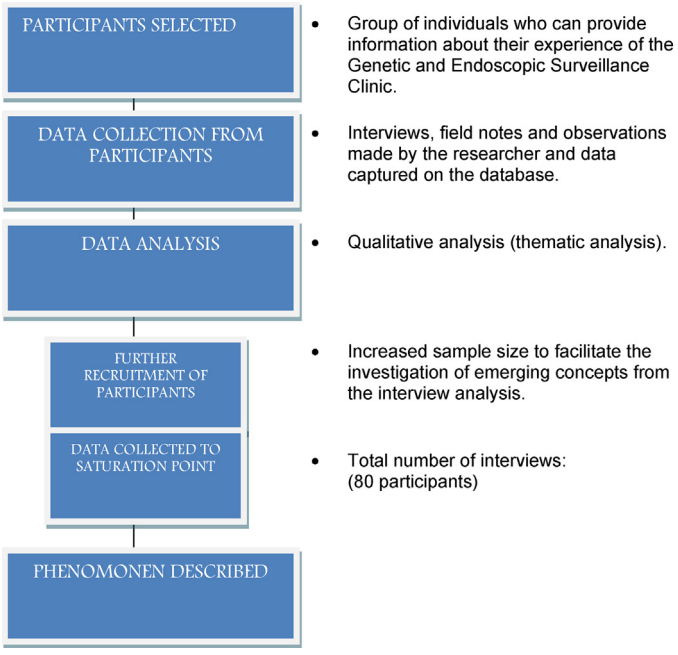

Fig. 1. An outline of the methodology used for the study.

2.3. Data analysis

A thematic analysis approach was undertaken to analyse the data. Transcripts were read through several times and then fragmented into separate categories. The initial categories for sorting the data were based on the literature and the researcher's background knowledge of the families at risk for LS. Codes were then assigned to each category, with the coded material used to explain the nature of the participant's experience. Constant comparing and contrasting of categories identified patterns of meaning and new information obtained from subsequent interviews was converted into new codes for further analysis. This facilitated sub-theme and theme development and a rich and exhaustive approach of the experience of receiving a mutation-positive test result as well as the subsequent communication of genetic information. Although several topics had been predetermined based on the structure of the interview schedule, themes within these topics and their relatedness to one another emerged from the interview rather than hypotheses being created, a priori. Saturation was declared for each category when it was found that new data did not contribute to further understanding. Coding and theme development was confirmed by two supervisors.

Quotations have been included to support theme development and have been translated by the researcher who is fluent in both Afrikaans and English, the languages in which the interviews were conducted.

3. Results

3.1. Participants

Eighty participants agreed to participate in the study conducted from 2009 to 2011. The participants were mostly female (55/80) and between the ages of 21 and 70 years (mean = 40.8 years). All participants were mutation-positive and between 1 and 12 years (mean = 8.6 years) had elapsed subsequent to the disclosure of the genetic test results. With regards to demographic characteristics, 64% (51/80) were married, and the majority classified themselves as Mixed Ancestry (Coloured population) and spoke Afrikaans as a home language (74/80; 93%). The remainder were Caucasian with English as a first language. More than half (43/80; 54%) of the participants were from a rural residential location and unemployed (46/80; 56%). The number of siblings and children per participant is displayed in Table 1.

Eighty-six percent (69/80) of the interviews were conducted in the participants' homes; the remainder were seen in a private room in the local clinic or private office near their place of employment or residence (these participants, for reasons of