Appendix 53: Characteristics of Included Studies (Question 7)

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Aktan-Collan 2011, Finland[196]	1) To systematically investigate whether LS carriers inform their offspring about the mutation and possibilities of predictive genetic testing, and the outcome of this information; 2) to explore the challenges in the disclosure process, wish for professional support, and the gender impact on communication.	Survey	Adult (> 40) LS carriers who have children	248	Questionnaire
Aktan-Collan 2000, Finland[172]	To investigate acceptance of an independent predictive genetic test, satisfaction with taking the test and reasons for and against taking the test.	Survey	Adults (≥ 18) without cancer diagnosis and at 50% risk of HNPCC	Baseline: 446 1-month follow-up: 299 1-year follow-up: 271	Questionnaire
Aktan-Collan 2013[179]	1) To examine the long-term psychosocial consequences of genetic testing; 2) to determine how the results of the testing related to satisfaction with the decision to undergo testing; 3) to examine behaviour of attending post-testing colonoscopy surveillance among both carriers and non-carriers and the relation to psychosocial factors.	Survey	Adult members of family with verified LS mutation, without cancer diagnosis and at 50% risk of HNPCC	208	Questionnaire
Aktan-Collan 2001[224]	To study the possible association of the result with emotional consequences such as general anxiety, fear of cancer and death, satisfaction with life, and attitude to the future in those who have undergone HNPCC testing.	Survey	Adults without cancer diagnosis, and at 50% risk of HNPCC	271	Questionnaire
Aktan-Collan 2001[248]	1) To evaluate the number of insurance policies purchased in the course of the predictive genetic testing programme; 2) to describe the number of existing insurances, the actual purchase of insurance policies in pre- and post-test periods and the planned purchase of insurance policies after testing.	Survey	Adults without cancer diagnosis, and at 50% risk of HNPCC	271	Questionnaire

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Aktan-Collan 2001[229]	To determine how the members of HNPCC families comprehended their predictive test results in terms of their risk of developing CRC and discuss what may have influenced this.	Survey	Adults (≥ 18) without cancer diagnosis, and at 50% risk of HNPCC in LS family with verified *MLH1* mutation	271	Questionnaire
Arver 2004, Sweden[188]	To prospectively evaluate the psychosocial consequences during the first year following pre-symptomatic testing with respect to anxiety, depression and quality of life in self-referred individuals tested for breast/ovarian or CRC genes known in their families.	Survey	Adult (> 18) female members of a family with verified mutation in *BRCA1*, *BRCA2*, *MLH1*, *MSH2*	21	Questionnaire
Balmana 2004, United States[243]	1) To explore potential differences in motivations and concerns about genetic testing among individuals at risk for HNPCC, FAP, and HBOC syndromes; and 2) to evaluate the influence of several clinical and demographic factors on the decision-making for undergoing genetic testing.	Survey	Eligible for genetic testing; at risk or affected by HNPCC, FAP, or HBOC	130 (HNPCC = 37)	Questionnaire
Barrow 2015, United Kingdom[187]	To assess the uptake of predictive testing and colorectal screening in FDRs of MMR mutation carriers and to elicit reasons for non-uptake and non-engagement.	Cross-sectional	Adults (> 18) FDR of MMR mutation carriers	591	Chart review
Broderson 2004, United Kingdom[244]	To investigate the anticipated emotional and behavioural reactions of patients at familial risk of CRC who are undergoing surveillance, to the offer of a genetic test for HNPCC.	Survey	Individuals with family history suggestive of hereditary cancer	437	Questionnaire
Bruwer 2013, South Africa[163]	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.	Qualitative description	Adults (≥ 18) who are mutation positive	80	Interview, observation

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Burton-Chase 2014, United States[222]	To evaluate the prevalence of gynecologic cancer screening among women with LS, their knowledge of LS risk and screening recommendations, and their perceptions regarding communication about LS with providers.	Qualitative description	Adult (≥ 25) females without a diagnosis of gynecologic cancer, who were LS mutation-positive or met Amsterdam II criteria	74	Interview, questionnaire
Carlsson 2007, Sweden[205]	To explore experiences from and perceived impact on life after genetic testing for HNPCC.	Qualitative description	Members of families with verified HNPCC mutation	19	Interview
Ceballos 2008, United States[237]	1) To assess willingness of CRC cases and relatives to receive genetic information that may indicate an increased risk for cancer; 2) to whom they would disclose genetic information; and 3) whether receiving genetic test results may influence future prevention behaviours.	Survey	Cases: Adults with CRC; FDR of cases	Cases = 45 Relatives = 102	Questionnaire
Claes 2004, Belgium[173]	To assess motivation, recall of cancer risks, and illness representations of individuals who had a predictive test for HNPCC as well as the short-term impact of predictive testing.	Survey	Members of families with verified dMMR mutation, without cancer diagnosis	40	Questionnaire, interview
Claes 2005, Belgium[192]	(1) To evaluate distress, illness representations, and health-related behaviour 1 year after disclosure of a predictive test result for HNPCC; (2) to delineate pre-test variables that would be associated with post-test distress and health-related behaviour.	Survey	Members of families with verified dMMR mutation, without cancer diagnosis	72	Questionnaire, interview
Codori 1999, United States[250]	To explore predictors of genetic testing for HNPCC in terms of psychological well-being and cancer prevention and early detection behaviours.	Survey	Adults (> 18) without cancer diagnosis, and member of a family with verified	258 from 95 families	Questionnaire, interview

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
			HNPCC mutation or with a family history suggestive of hereditary cancer		
Cragun 2012, United States[184]	To explore differences between CRC survivors who did and did not express interest in having genetic testing for hereditary CRC if it were made available to them and to determine which factors most strongly correlate with interest in genetic testing.	Survey, secondary analysis	Patients with CRC	91	Questionnaire
de Leon 2004, Italy[189]	1) To evaluate how many high-risk individuals in each family underwent genetic testing for the search of constitutional mutations; 2) to ascertain whether mutation-positive unaffected individuals made proper use of the test (i.e., accepted endoscopic surveillance); and 3) to investigate the main findings of endoscopic surveillance in gene carriers.	Cross-sectional	Members of families with verified HNPCC germline mutation	164 from 32 families	Chart review
Dewanwala 2011, United States[210]	To examine the attitudes toward child-bearing and prenatal genetic testing among individuals undergoing genetic evaluation for LS.	Survey	Adults (≥ 18) with personal or family history suggestive of LS	161	Questionnaire
Esplen 2001, Canada and United States[174]	To systematically examine the attitudes and motivations associated with genetic testing for HNPCC, the current levels of psychosocial functioning of individuals engaged in the genetic testing process, and patterns of disclosure for receiving those results.	Survey	Adults (≥ 18) with personal or family history suggestive of HNPCC, and eligible for genetic testing	50	Questionnaire

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Esplen 2003, Canada[203]	To describe current distress levels in a sample of high- and intermediate-risk CRC patients who have provided a blood sample for genetic testing, and explore the relationship between variables associated with current pre-test distress, and post-test distress.	Survey	Cases: Adults (> 20) with high or intermediate risk of CRC and cancer of the large bowel; FDR of cases and family history suggestive of hereditary cancer	220	Questionnaire
Esplen 2007, Canada[177]	To assess the psychosocial impact of genetic counselling and testing among individuals undergoing HNPCC genetic testing.	Survey	Adults (> 18) with CRC	314	Questionnaire
Esplen 2015, Canada[211]	To examine the longer-term psychosocial and behavioural impact on individuals undergoing genetic testing for LS in two Canadian familial CRC registries.	Survey	Adults (≥ 18) who were confirmed either mutation positive or negative	155	Questionnaire
Fantini 2007, France[206]	To characterize people who participate in a screening program for hereditary CRC, and to determine whether people at risk who test for inherited CRC are likely to develop a higher level of psychological distress than the norm.	Survey	Adults at risk for HNPCC, with or without cancer diagnosis	77	Questionnaire
Glanz 1999, United States[164]	To identify the sociodemographic, psychosocial, and social influence factors related to intentions to participate in genetic testing and genetic counselling for CRC risk among persons from 3 ethnic groups who are at increased family risk.	Survey	Cases: Adults (> 20) with cancer of the large bowel; FDR (> 18) of cases (excluding parents) without cancer diagnosis	426	Questionnaire
Graves 2014, United States[197]	To evaluate a telephone protocol for returning research results of MMR gene testing to identify LS.	Survey	Members of families with verified *hMSH2*, *hMLH1*, *hMSH6*, *hPMS2* mutation	Baseline: 107 6-month follow-up: 85	Questionnaire, interview

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Gritz 1999, United States[226]	To examine the association between baseline measures of attitudes toward genetic testing and counselling for HNPCC and the affective state of patients who had given blood for a HNPCC genetic testing study.	Survey	Adults (≥ 18) HNPCC patients	269	Questionnaire
Palmquist 2010[165]	To explore the familial context of risk perception formation in 3 families with a documented genetic susceptibility to LS.	Qualitative description	Adults FDR (> 18) at 25% or 50% risk of dMMR mutation; member of a family with verified dMMR mutation or currently undergoing LS testing (including spouses)	26 from 3 families	Interview
Gritz 2005[225]	To examine the impact of HNPCC genetic test results on psychological outcomes among cancer-affected and -unaffected participants up to 1 year after results disclosure.	Survey	Cases: Diagnosed with CRC with a family history suggestive of hereditary cancer; relatives of HNPCC mutation-positive patients at 25% or 50% risk	155	Questionnaire
Pentz 2005[231]	To describe HNPCC family members' perceptions about who has a right to know about a genetic mutation in the family and who should disclose this information to family members, with a focus on the role that should be played by health professionals in disseminating this information.	Qualitative description	Members of families with verified HNPCC mutation, or undergoing testing, or family with at least 5 members at 50% risk	80 from 16 families	Interview

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Koehly 2003[221]	1) To describe the composition of familial networks; 2) to characterize the patterns of family functioning and genetic counselling and testing discussions within the families; 3) to examine how the patterns, or the familial culture, were related to discussing genetic counselling and testing among family members; 4) to identify the characteristics of individuals with the most influence in the family.	Qualitative design, using Social Network methodology	Member of family with verified HNPCC mutation (including spouses)	36 from 5 families	Interview
Peterson 2003[166]	To describe how information about the identification of an HNPCC gene mutation was disseminated in 5 families, when and under what circumstances this information was shared, and how family members reacted to and acted on this information.	Qualitative description	Adults (≥ 18) from a family with a verified HNPCC mutation and at least 5 members at 50% risk	39 from 5 families	Interview
Vernon 1999[195]	To assess the association between intention to learn genetic test results and sociodemographic factors, medical history, psychosocial factors, attitudes, beliefs, and decisional considerations related to genetic testing.	Survey	Patients with CRC, and unknown mutation status	269	Questionnaire
Hadley 2003, United States[170]	To assess CRC screening behaviours of index cases with indeterminate HNPCC genetic test results and their at-risk FDRs, and what relational factors affect CRC screening in families in which the index case received indeterminate genetic test results.	Survey	Member of family with verified HNPCC mutation	104	Questionnaire
Eliezer 2014[230]	To explore how personal genetic test results and immediate and extended family members' test results for LS shape subsequent cancer distress, cancer worry, and depression.	Survey	Member of family with verified mutation	179 from 26 families	Questionnaire

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Morris 2013[232]	To investigate relationships between spirituality (consisting of religious and existential well-being) and: 1) psychological factors (perceived cancer risk and worry, depressive symptoms, and cancer-related distress); 2) family network characteristics (social support and family communication regarding risk and genetic testing); and 3) screening practices for CRC.	Survey	Adults from families with verified HNPCC mutations	123 from 34 families	Questionnaire
Ersig 2011[257]	To examine the association of selected individual and relational characteristics with communication about HNPCC, and compare these associations between families with and without identified mutations.	Cross-sectional	Probands: mutation-positive status; FDR of probands	Index cases: 20 FDRs: 31	Interview
Ersig 2010[251]	To explore thoughts about and response to risk for HNPCC in the context of indeterminate genetic test results.	Qualitative description	Probands: indeterminate mutation status with HNPCC-associated cancer or met eligibility criteria for testing; FDR of probands	Index cases: 10 FDRs: 16 from 11 families	Interview
Hadley 2010[234]	To examine whether previous family experiences with genetic services for the inherited cancer susceptibility syndrome known as LS were associated with a decline or increase in baseline levels of depressive symptoms, disease worry, and genetic test–related distress among family members who receive genetic services at more distant time intervals.	Survey	Member of family with verified mutation	297 from 38 families	Questionnaire

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Ashida 2009[236]	1) To investigate the associations between perceived family relationships before genetic testing as well as changes in perceptions 6 months after results disclosure and the changes in depression scores over a 12-month period after disclosure; and (2) To evaluate the moderating effect of family norms and discrepancy in worry about cancer on these associations.	Survey	Families with verified LS or at risk for inheriting a mutation	178 from 24 families	Questionnaire
Ersig 2009[214]	To examine CRC screening by endoscopy and disclosure of genetic test results among index cases at risk for HNPCC in the year after genetic test results were received. Of particular interest were the effects of mutation status (mutation-positive vs. inconclusive) and disclosure of the genetic test result to others on endoscopy completion.	Survey	Adults (≥ 18) with at least one HNPCC-associated cancer diagnosis, and a personal or family history suggestive of hereditary cancer	69	Questionnaire
Ersig 2009[209]	To compare colonoscopy screening, and the effect of relational factors on screening, between families with indeterminate and mutation-positive HNPCC genetic test results.	Survey	Individuals who underwent genetic testing and their adult children and siblings	46	Questionnaire, interview
Palmer 2005[245]	To describe how signal detection methods could be used to evaluate the impact of counselling and testing for susceptibility genes.	Survey	Members of a family with verified HNPCC mutation, without cancer diagnosis	56	Questionnaire
Hadley 2004[215]	To assess endoscopy use and predictors of adherence to endoscopy screening guidelines after the receipt of positive- and true negative HNPCC mutation results among asymptomatic individuals in families with known HNPCC mutation.	Survey	Adults (≥ 18) without a cancer diagnosis, at 50% risk of HNPCC mutation	56	Questionnaire
Halbert 2004, United States[212]	1) To assess whether genetic testing for HNPCC mutations and receipt of positive test results have an effect on the use of	Survey	Members of families with verified HNPCC	98	Interview, questionnaire

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
	colonoscopy; 2) to identify factors associated with adherence to identify potential barriers to patient compliance.		mutation at 25% risk of inheriting mutation		
Ho 2003, China[182]	1) To study the decisional consideration process of Hong Kong Chinese toward genetic testing of CRC; and (b) to examine factors affecting decisional consideration of CRC genetic screening testing.	Survey	Individuals belonging to a Hereditary Gastrointestinal Cancer Registry	62 from 35 families	Questionnaire
Johnson 2002, United States[213]	To evaluate, in a clinic population, the impact of both genetic counselling and testing on subsequent CRC screening behaviour.	Cross-sectional	Patients without a cancer diagnosis, at risk of HNPCC	65	Chart review
Keller 2004, Germany[167]	To evaluate the uptake of genetic counselling in a nonselected sample of patients at risk for HNPCC.	Survey	Cases: adults (≥ 18), had surgery for CRC- or HNPCC-associated cancers with a family history suggestive of hereditary cancer; relatives of probands	25	Questionnaire
Keogh 2009, Australia[198]	To assess whether knowledge of insurance implications influenced uptake of genetic testing by participants in a research study of the causes of CRC.	Pre-post	Cases: CRC diagnosis before age of 45 years; FDR and SDR of probands	106 (47 from the original protocol; 59 in the modified protocol)	Chart review
Kidambi 2015, United States[199]	To examine 2 different IHC-based LS screening protocols at an urban, university hospital: selective screening based on criteria and universal screening of all CRC patients — in terms of number of LS cases identified.	Chart review	Patients with surgically resected CRC	392 (107 selective; 285 universal)	Chart review
Kinney 2000, United States[181]	To examine the level of interest in genetic testing for heritable CRC in African-American and white patients, and assess	Survey	Adults (≥ 18) with CRC	98	Structured Interview

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
	psychological factors and knowledge of the availability of genetic testing for CRC.				
Kinney 2000, United States[202]	To determine colorectal cancer screening behaviours, risk perceptions, and willingness to receive genetic testing to determine colorectal cancer susceptibility in FDRs of CRC patients.	Survey	FDR of persons with CRC	95	Structured interview
Kohut 2007, Canada[217]	To survey participants of the OFCCR regarding their duty to warn family members about a HNPCC-causing mutation.	Survey	High-risk individuals in patient registry; intermediate-risk individuals with personal or family history suggestive of hereditary cancer	105	Questionnaire
Kupperman 2013, United States[216]	To measure patient preferences (utilities) for scenarios entailing differing decisions regarding test use and risk-reducing surgeries and their associated outcomes among a group of individuals with a wide range of familiarity and experience with LS testing.	Cross-sectional	Individuals having undergone genetic testing and counselling for LS; NR for patients unknowledgeable about LS	70	Questionnaire, preference elicitation (time trade off)
Landsbergen 2011, Netherlands[223]	To investigate whether high levels of overall psychological distress are present during MSI testing and whether these levels are correlated with time since CRC diagnosis.	Survey	Patients with CRC, or eligible for MSI testing	89	Questionnaire
Landsbergen 2012[233]	To investigate general distress and cancer-specific distress in CRC patients during their treatment phase who meet criteria for MSI testing indicated by a pathologist.	Survey	Patients with CRC diagnosed before age 50 years, or second CRC diagnosed before 70 years (including partners)	CRC patients: 81 Partners: 50	Questionnaire
Landsbergen 2009, Netherlands[194]	To explore the reactions of patients to the offer of genetic testing in the period directly after surgical removal of the tumour.	Qualitative description	Patient with CRC and MSI-positive tumour	8	Interview, chart review

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Leenan 2016, Netherlands[183]	1) To evaluate experiences and attitudes toward a family-mediated approach in an LS cohort; 2) to compare tested (mutation carriers and non-carriers) and non-tested individuals on demographic characteristics, anxiety, cancer worry, medical history, family communication, experiences and attitudes toward the family-mediated approach; and 3) to explore the motivations for uptake or decline of genetic testing for LS.	Survey	Individuals with a personal or family history of LS	129 from 33 families	Questionnaire
Lerman 1996, United States[175]	To explore how patients might react both to the availability of genetic testing for CRC susceptibility and to disclosure of genetic test results.	Survey	FDR of persons with CRC	45	Structured interview
Lindor 2004, United States[180]	To explore which patients opted to learn their results, to determine what reasons they had for deciding to learn results, to determine the effect of detail (an in-depth explanation of testing versus a brief overview), and to assess global psychological reactions to being offered and receiving these complex test results via a written communication.	Survey	Patients with CRC diagnosed before age 50 years; individuals with a family history suggestive of hereditary cancer	414	Questionnaire
Loader 2005, United States[169]	To assess the impact of a genetic evaluation for CRC genetic susceptibility including whether counselled individuals remember the information provided, change their behaviour, and alert their relatives to their potential risk.	Survey	Patients with CRC diagnosed before age 60 years with a FDR or SDR with CRC	37	Questionnaire, interview
Lynch 1999, United States[171]	To describe experiences with DNA-based genetic counselling with 7 HNPCC families, 5 of which showed *hMLH1* and two of which manifested *hMSH2* germline mutations.	Unclear	NR	199 from 7 families	Interviews, during counselling sessions

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Manne 2007, United States[191]	1) To characterize the level of knowledge about the purpose and results of the MSI test among individuals considering this test, and to describe the level of previous exposure to information about the MSI test; 2) to characterize motivations for (perceived benefits) and against (perceived barriers) having the MSI test; 3) to examine the contribution of attitudinal and non-attitudinal variables to perceived benefits and barriers of MSI testing.	Cross-sectional data collected as part of an RCT	Individuals eligible for genetic testing (rBG criteria)	125	Questionnaire
McCann 2009, United States[186]	To explore the factors influencing family communication about genetic risk of CRC and colonoscopy among people who had a strong family history of CRC who were attending a genetic clinic with a view to having a genetic test for HNPCC.	Qualitative Description	Individuals with a strong family history of CRC	30 from 17 families	Interview
Meiser 2004, Australia[200]	To assess psychological impact and screening behaviours in both carriers and non-carriers of mutations pre-disposing to HNPCC, at baseline, 1 year, and 3 years after genetic testing.	Survey	Members of a family with verified HNPCC mutation	114	Questionnaire
Collins 2007[246]		Survey	Individuals undergoing predictive genetic testing, without a personal history of HNPCC-associated cancers or CRC	73	Questionnaire
Collins 2005[247]		Survey	Individuals undergoing predictive genetic testing, without a personal history of HNPCC-associated	98	Questionnaire

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
			cancers or CRC		
Mesters 2005, Netherlands[219]	To investigate people's perspective regarding informing one's biological family on the hereditary predisposition for HNPCC.	Qualitative description	Individuals with a family history suggestive of hereditary cancer	30	Interview
Murakami 2004, Japan[228]	To identify the prevalence and predictive factors of major and minor depression, acute stress disorder, PTSD, and post-traumatic stress symptoms after the disclosure of genetic test results for HNPCC in Japanese individuals, both in probands affected with cancer and in unaffected relatives.	Pre-post	Members of family (≥ 20) with verified *hMLH1*/*hMSH2* mutation	42	Interview, questionnaire
Petersen 1999, United States[207]	To examine the relationship between family history, risk perception, and interest in CRC gene testing.	Survey	Adults (> 18) without a cancer diagnosis, and at least one FDR with CRC	Surveys: 1,217 Interviews: 156	Questionnaire
Ramsey 2010, United States[235]	1) To determine how a hypothetical test for gene variants associated with a moderately increased CRC risk might influence individuals' health-related quality of life, cancer worry, health habits, and screening behavior; 2) to determine whether an individual's family history of CRC (and thus his/her current estimate of cancer risk) modified these issues.	Survey	Cases: Adults (> 18) with CRC; FDR of cases; population-based controls	Population-based controls: 170 FDRs: 310	Questionnaire
Ramsey 2003, United States[190]	1) To examine beliefs about and key issues related to testing for CRC susceptibility genes; and 2) to compare the relative importance of these issues and how they influence willingness to accept testing among the 3 groups (CRC patients, FDRs of CRC patients, and people with no personal or family history of CRC).	Qualitative description	Patients from registry, and their FDRs; population controls	CRC patients: 6 FDRs: 4 FDRs Controls: 5	Focus group
Reeve 2000, New Zealand[193]	To examine the impact of genetic testing for HNPCC by intensive study of the only group	Qualitative description	Individuals tested for HNPCC	7	Interview

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
	in New Zealand to have undergone genetic testing for HNPCC.				
Roygnan 2008, France[204]	To observe how probands deal with the transmission of the genetic information to their families; to let them talk freely about how they felt and how they told their relatives.	Qualitative description	Mutation-positive individuals	7	Interview
Shiloh 2008, United States[238]	1) To examine the distress and depression levels of individuals testing for HNPCC mutations prospectively from baseline (before testing) through 6 and 12 months after disclosure of test results; 2) to explore the modifying effects of mutation status and monitoring coping styles on distress and depression; and 3) to explore interactions between mutation status and monitoring coping style.	Survey	Cases: patients with CRC or endometrial cancer diagnosed before age 40 with tumours consistent with HNPCC, or with family history suggestive of HNPCC; family members of cases with identified mutations	253 (67 index cases and 186 family members at risk)	Questionnaire
Shipman 2013, United Kingdom[176]	To gain insight from those who have undergone MMR tumour testing and to assess the significance that testing holds for them. Of particular interest are the ways in which respondents “account” for testing via displays of knowledge and responsibility.	Qualitative description	Individuals consenting to MSI testing without prior genetic counselling	11	Interview
Stoffel 2008, United States[218]	To examine how genetic testing information is communicated in families at risk for LS, and to identify factors associated with disclosure of genetic test results to close and distant family members.	Survey	Adults (> 18) with a personal or family history suggestive of hereditary cancer (BG)	174	Questionnaire

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Tomiak 2014, Canada[178]	1) To investigate the different factors affecting uptake of genetic counselling and testing in newly diagnosed CRC patients being seen in a Canadian academic hospital Cancer Assessment Clinic; 2) to examine the experience of individuals facing a choice about genetic counselling and/or testing in the context of newly diagnosed CRC, focusing on motivations and barriers encountered.	Qualitative description	Patients with CRC eligible for genetic testing	19	Interview
van Oostrom 2007, Netherlands[240]	To explore predictors for hereditary cancer distress 6 months after genetic susceptibility testing for a known familial *BRCA1/2* or HNPCC-related mutation, in order to gain insight into aspects relevant for the identification of individuals needing additional psychosocial support.	Survey	Adults at 50, 25, or 12.5% risk for known familial BRCA1/2 or HNPCC mutations	HNPCC: 96	Questionnaire
van Oostrom 2007[227]	To study differences between individuals opting for genetic cancer susceptibility testing of a known familial *BRCA1/2* and HNPCC-related germline mutation.	Survey	Adults at 50%, 25%, or 12.5% risk for known familial *BRCA1/2* or HNPCC mutations	HNPCC: 96	Questionnaire
van Oostrom 2007[241]	To examine prospectively the contribution of family functioning, differentiation to parents, family communication and support from relatives to psychological distress in individuals undergoing genetic susceptibility testing for a known familial pathogenic *BRCA1/2* or HNPCC-related mutation.	Survey	Adults (≥ 18) from a family with a verified HNPCC mutation or *BRCA1/2* mutation, with or without a personal history of cancer	HNPCC: 96	Questionnaire
van Oostrom 2006[242]	To explore the effect of age at the time of parental cancer diagnosis or death on psychological distress and cancer risk perception in individuals undergoing genetic testing for a specific cancer susceptibility.	Survey	Adults at 50%, 25%, or 12.5% risk for known familial *BRCA1/2* or HNPCC mutations	HNPCC: 96	Questionnaire

First Author Publication Year, Country of Origin[a]	Study Objectives	Study Design	Inclusion Criteria	Sample Size	Data Collection Methods
Vernon 1997, United States[249]	To describe the demographic and psychosocial correlates of 2 measures of psychologic distress (depression and anxiety) among 200 CRC patients undergoing genetic testing for HNPCC.	Cross-sectional	Patients with CRC with unknown mutation status	200	Questionnaire
Wagner 2005, Netherlands[201]	To evaluate the use of colonoscopy by proven HNPCC mutation carriers, and satisfaction with the counselling and screening procedures in the long term.	Survey	Individuals with known MMR gene mutations	70	Questionnaire
Wakefield 2007, Australia[168]	To describe the perceived pros and cons reported by individuals who have undergone at least one genetic counselling consultation, as well as having completed a genetic testing decision aid, before considering genetic testing for HBOC or HNPCC risk.	Qualitative description	Adults eligible for HBOC or HNPCC testing	22	Questionnaire, chart review
Walsh 2012, United States[185]	To establish key characteristics that patients, consumers, and health professionals consider in decision-making about being tested for hereditary LS.	Qualitative description	Patients with CRC, or at high risk for CRC; average-risk individuals; physicians; genetic counsellors	8	Focus group
Watkins 2011, Canada[208]	To explore how confirmed carriers experience disease management and view the quality of interactions with health care providers and the overall health care system.	Grounded theory	Member of family with known *MSH2* mutation	23	Interview
Yamashita 2008, Japan[239]	To investigate the psychological impact of disclosure of genetic test results and the factors associated with it, especially focusing on memory function, in participants who underwent genetic testing for HNPCC and were informed of the results.	Survey	Adults (> 20) with family history suggestive of HNPCC	46	Questionnaire

BG = Bethesda Guidelines; CRC = colorectal cancer; dMMR = deficient mismatch repair; FAP = familial adenomatous polyposis; FDR = first-degree relative; HBOC = hereditary breast–ovarian cancer syndrome; HNPCC = hereditary non-polyposis colorectal cancer; LS = Lynch syndrome; MMR = mismatch repair; MSI = microsatellite instability; NR = not reported; OFCCR = Ontario Familial Colon Cancer Registry; PTSD = post-traumatic stress disorder; rBG = revised Bethesda Guidelines; SDR = second-degree relative.
[a] Studies with a common (sub) sample of patients are grouped together, with the primary study left-justified in the cell and related studies right-justified in subsequent rows.