

## Short Report

# An exploratory comparison of genetic counselling protocols for HNPCC predictive testing

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Most UK genetics centres offering predictive testing for hereditary non-polyposis colorectal cancer (HNPCC) use an extended counselling protocol originally developed for Huntington's disease. Shortened counselling may be more appropriate in the context of treatable genetic conditions such as HNPCC. Twenty-six high-risk individuals were randomized to extended genetic counselling (two sessions of education and reflection held 1 month apart) or shortened genetic counselling (a single educational session) prior to HNPCC testing. Prospective questionnaires, interviews and transcripts of counselling sessions were analysed. Participants were unsure what to expect prior to genetic counselling and had already decided to undergo genetic testing. There was no evidence of psychological harm caused by shortened genetic counselling, with a high level of satisfaction with the counselling received in both groups. Reflective counselling occurred in both groups but was framed in terms of practical action and information. Participants expressed differing preferences for the level of information received. This exploratory study indicates that shortened genetic counselling may be an appropriate means of supporting decisions already made by individuals about HNPCC testing. However, participants would benefit from preparatory information to help them reflect on issues not previously considered, which can then be explored more fully as part of a tailored counselling approach.

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Colorectal cancer (CRC) is one of the most common cancers in the West (1). Around 1–5% of cases are linked to hereditary non-polyposis colorectal cancer (HNPCC), an autosomal dominant cancer syndrome conferring a 50–80% lifetime risk of CRC in gene carriers (2). Predictive genetic testing enables carriers to be targeted for regular screening to detect CRC at an early and potentially curable stage (3, 4). Testing is typically offered within an extended counselling protocol, originally developed in the context of Huntington's disease (HD) to facilitate reflection about the consequences of testing in the absence of therapeutic options. This consists of at least two counselling sessions involving education and reflection, separated by a

1-month contemplation period (5). Reflection is considered to be a major component which aims to promote informed decision-making and facilitate psychological adjustment through intense engagement with risk in hypothetical scenarios (6). This approach represents rational models of decision-making in which patients systematically weigh up information to reach an 'optimal' decision (7).

Individuals at high risk of inherited cancer syndromes may not require the extensive discussion and contemplation time prescribed by the HD model (8, 9). Prospective studies confirm that there are no long-term adverse psychological effects of HNPCC testing (10, 11) and that most participants are satisfied with a single pre-test

counselling session (12). However, it is unclear from these non-randomized studies whether favourable psychological outcomes were attributable to the use of particular forms of counselling. The only randomized trial of pre-test counselling approaches found that a counselling intervention did not confer advantages over a single session of education prior to BRCA1 gene testing (13). A shortened protocol may therefore be appropriate in the context of treatable genetic conditions such as HNPCC, and many centres are already reducing their protocols for such testing (14, 15). This small study aimed to explore the process of extended *vs* shortened genetic counselling protocols and compare their impact on psychological and decision-making outcomes in individuals eligible for HNPCC predictive testing.

## Patients and methods

Ethical approval was obtained. Currently unaffected individuals were recruited from families in Wales in which an HNPCC gene mutation had been identified. Fifty-one individuals were given a standard introductory letter and reply slip by the proband following consultation with the genetic nurse specialist (GNS). Thirty-four of 51 family members returned the reply slip indicating permission to be telephoned by the GNS. Individuals were then sent a study information sheet, consent form and anonymized baseline questionnaire which was returned by 31 individuals (four subsequently declined). Twenty-seven participants were randomized (see Fig. 1). One participant withdrew from the extended protocol for practical reasons associated with attending further counselling. The final sample consisted of 26 participants.

### Study design

Participants received extended or shortened genetic counselling prior to HNPCC testing (see Fig. 1). A randomization schedule was generated

by an independent researcher, incorporating stratification by clinical team. Random allocations were revealed to participants and clinicians at the first or single counselling session using sealed, opaque envelopes. Clinical staff were trained to use a standard counselling protocol for each group (see Table 1). All counselling sessions were timed, indicating a similar mean duration of shortened counselling single sessions and extended counselling first sessions.

### Extended genetic counselling

Participants randomized to extended counselling received two sessions of education combined with reflective counselling, held 4 weeks apart. Educational and reflective components of extended counselling are summarized in Table 1 (items 1–10).

### Shortened genetic counselling

Participants received a single educational session covering core educational components (items 1–5). Participants who felt unready to make a decision had the option of further counselling, although no such crossover occurred.

Test results were disclosed at a clinic session 2 weeks after genetic counselling in both groups. Gene carriers were referred for appropriate cancer screening. Participants in both groups were contacted by a GNS 1 week after test results, with the offer of a further 1-month follow-up appointment (12).

### Sample characteristics

Fourteen women and 12 men participated, with an average age of 41 years. Participants had been aware of their increased risk status for 9 years on average, and 10 had prior contact with the genetics service. Fourteen were part of a regular bowel screening programme. Study groups were equivalent on all baseline demographic, clinical and psychological variables.

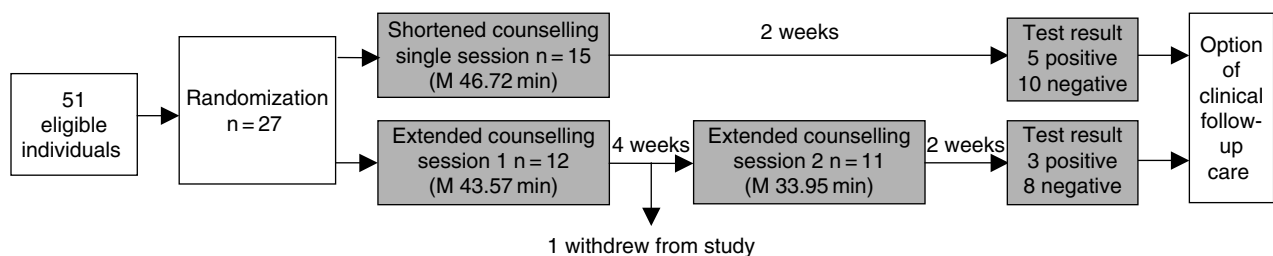


Fig. 1. Study design.

Table 1. Summary of counselling content

Counselling content	Extended counselling protocol	Shortened counselling protocol
Education	(1) HNPCC and colorectal/other cancer risks (2) Factual details of HNPCC gene testing (3) Prevention and surveillance options after a positive/negative test result (4) Potential genetic implications for family members (5) General discussion of reasons for/against testing	Items 1–5
Reflection	(6) Experiential issues relating to risk (7) Personalized discussion of reasons for/against testing (8) Result scenarios – potential consequences for individual and family (9) Rehearsal of ways of coping	None

Measurement

*Questionnaires*

Questionnaires were administered prior to and immediately after genetic counselling, with a subset administered 3 months post-result. Two ‘in-house’ measures included counselling preferences and the extent to which these were fulfilled, and intention to undergo HNPCC testing. Validated measures included systematic decision-making process (16), satisfaction with the decision to undergo testing (17), psychological distress (18) and knowledge about HNPCC (19). All measures demonstrated satisfactory internal consistency.

*Qualitative interviews*

Semistructured interviews were performed with eight participants (four shortened and four extended) an average 16 months post-testing. Interviews were stratified by test result and included five women and three men (age range = 25–51). Topics covered information sources, decision-making process, genetic counselling process, quality of interaction in genetic counselling and impact of the test result. Permission was obtained from participants to audiorecord the interviews.

*Transcripts of genetic counselling sessions*

Eighteen counselling sessions involving 12 consenting participants (i.e. six shortened and six extended) were audiorecorded and transcribed for comparative analysis of reflective elements in shortened and extended counselling.

Analysis

Using SPSS version 11.5, non-parametric statistics were performed to compare psychological and decision-making outcomes of genetic counselling protocols (Table 2). Due to very small numbers when these data were stratified by test result,

outcomes at 3 months were observed descriptively (Table 3). Transcripts of interview data and counselling sessions were analysed using NVivo qualitative analysis software. Thematic analysis of the interviews identified recurrent themes through constant comparative analysis undertaken by three researchers (LH, KB and KB). Discourse analysis of communicative frames within the counselling sessions identified proportions and sequencing of educational and reflective frames (20). This part of the analysis has been reported in more detail elsewhere (21).

**Results**

This article presents an overview of the key themes drawn from combined data sources. Statistical comparisons (Table 2) are supported by qualitative comments from participants.

Expectations of genetic counselling

At baseline, counselling preferences were significantly more likely to be for information than support ( $Z = -2.98, p = 0.003$ ): ‘It did take me by surprise. I thought it would be more medical ... more the calculated kind of thing, rather than an emotions thing’ (extended counselling, negative result). Interviewees in both groups were unsure what to expect: for example, three had expected to simply ‘give blood’ without preparatory counselling; two others were shocked that a blood sample would be taken, although they welcomed the chance to be tested. However, participants were very positive in their evaluations of genetic counselling, reporting high levels of satisfaction with the information and support received in both groups ( $p = 0.13$ ): ‘There was never any question unanswered, and there was

Table 2. Psychological and decision-making outcomes of extended vs shortened genetic counselling

Questionnaire measure <sup>a</sup>	Study group	Pre-counselling Median (IQ-R)	Post-counselling Median (IQ-R)	Mann – Whitney <i>U</i> test <sup>b</sup>	
				Z	p
Counselling preference – information (score range = 0–6)	Extended	6.00 (0.00)	6.00 <sup>c</sup>	–1.53	0.13
	Shortened	6.00 (1.00)	6.00 (0.25)		
Counselling preference – support (range = 0–6)	Extended	3.00 (5.00)	6.00 (0.50)	–0.23	0.82
	Shortened	4.00 (5.00)	6.00 (1.00)		
Intention to undergo testing (range = 0–8)	Extended	8.00 (0.00)	8.00 <sup>c</sup>	0.00	1.00
	Shortened	8.00 (0.00)	8.00 <sup>c</sup>		
Systematic decision-making process (range = 0–26)	Extended	9.00 (5.00)	8.00 (5.00)	–0.72	0.47
	Shortened	5.00 (6.00)	7.50 (2.25)		
Satisfaction with test decision (range = 0–30)	Extended	–	30.00 (0.00)	–0.85	0.40
	Shortened	–	30.00 (0.50)		
Psychological distress (range = 0–36)	Extended	10.00 (5.36)	10.00 (4.00)	–0.03	0.98
	Shortened	8.00 (4.00)	9.00 (6.00)		
Knowledge about HNPCC (range = 0–8)	Extended	4.00 (2.00)	5.00 (1.00)	0.00	1.00
	Shortened	4.00 (2.00)	5.00 (1.00)		

<sup>a</sup>Higher scores indicate higher levels of each measure.

<sup>b</sup>Statistical comparisons were performed on the difference between post-counselling scores.

<sup>c</sup>Interquartile range (IQ-R) could not be calculated due to the absence of variation in scores.

never any concern that was washed over' (shortened counselling, positive result).

#### Decision-making about genetic testing

Prior to genetic counselling, there was a definite intention to undergo testing, which did not differ after counselling in both groups ( $p = 1.00$ ). Similarly, neither counselling approach led to a more systematic decision-making process ( $p = 0.47$ ). All those interviewed said that making the decision had been easy: 'They said I should go away and think about it, but I didn't need to ... The answer was "yes"' (extended

counselling, positive result). All had decided to undergo testing as soon as they heard that it had become available, often several years ago: 'The first time I went down, if they would have offered me the test there I would have had it. I didn't go just to have counselling, I went to have the test ... I knew all about it anyway for the past 20 years then. It wasn't such a big deal for me' (shortened counselling, positive result). However, participants in both groups regarded counselling as important in supporting their decisions, and high levels of decision satisfaction were shown regardless of study group ( $p = 0.40$ ).

Table 3. Descriptive comparison of psychological and decision-making outcomes at 3-month follow-up

Questionnaire measure <sup>a</sup>	Study group	Test result	Three-month post-result <sup>b</sup> Median (IQ-R)
Psychological distress (score range = 0–36)	Extended	Positive	14.00 <sup>c</sup>
		Negative	8.00 (9.25)
	Shortened	Positive	6.50 (2.50)
		Negative	9.00 (4.00)
Satisfaction with test decision (range = 0–30)	Extended	Positive	25.00 <sup>c</sup>
		Negative	30.00 (6.00)
	Shortened	Positive	30.00 (0.00)
		Negative	30.00 (6.00)
Knowledge about HNPCC (range = 0–8)	Extended	Positive	5.00 <sup>c</sup>
		Negative	5.50 (2.25)
	Shortened	Positive	5.50 (1.75)
		Negative	5.00 (1.00)

<sup>a</sup>Higher scores indicate higher levels of each measure.

<sup>b</sup>Statistical tests of significance were not performed.

<sup>c</sup>Interquartile range (IQ-R) could not be calculated due to very small numbers.

## Psychological distress

Levels of psychological distress were not significantly different immediately after extended counselling or shortened counselling ( $p = 0.98$ ). However, trends at 3-month follow-up suggested that gene carriers in the shortened group experienced the least distress along with a high level of decision satisfaction, in contrast to carriers in the extended group who reported the most distress and least decision satisfaction (see Table 3).

## Knowledge about HNPCC

The effect of counselling protocol on knowledge was not significant ( $p = 1.00$ ), although in both groups there appeared to be a small improvement which was sustained in the longer term (see Table 3). All interviewees described genetic counselling as highly informative and learnt fundamental facts relating to inheritance of which they were previously unaware. However, different preferences were expressed for the level of detail provided. A participant in the extended group found the repetition of facts slightly tedious, while recognizing that what she required would not necessarily be suited to everyone: 'It was always the same thing, the percentages, the predictive tests, all of it really ... I suppose it didn't help me but I think it might help some people' (extended counselling, negative result). One female participant (extended counselling, positive result) remained convinced that only men would be affected by colon cancer and was shocked to receive a positive test result. Long-standing beliefs may not simply be corrected through the provision of facts alone, and a deeper dialogical exploration may be required for some individuals.

## The role of reflection in HNPCC counselling

Analysis of counselling sessions indicated that counsellors' reflective questions occurred equally in both protocols. Reflective talk produced by participants was minimal in both protocols, but more likely to occur in the first or only session. When addressing emotional issues, counsellors tended to supply participants with possible themes for introspection within an informational frame rather than to encourage reflective talk: 'some people will say "not gonna think about this ..." and other people will say "if there's information there I want to know it ..."'. Hypothetical scenarios were formulated primarily by counsellors and in terms of practical action: 'what you could do' (e.g. participation in

screening programmes). Prompts for participants to engage in reflection were less likely to succeed after extensive discussion of hypothetical scenarios focusing on practical actions. Interviewees in both groups said that the reflective elements had taken them by surprise, though in retrospect they could see the benefit of exploring feelings: 'It made you confront them. I didn't like it, but it was a good idea' (extended counselling, negative result).

## Discussion

This exploratory study found no evidence of harm caused by shortened genetic counselling. Neither counselling protocol seemed to influence individuals' decisions, rather serving to confirm decisions already made (16, 22). This supports naturalistic models of decision-making which acknowledge the role of intuition in making complex decisions (23). However, care should be taken to avoid overgeneralizing from this small and selective sample. Those undergoing predictive testing are a highly select sample and likely to have been aware of their increased risk for some time (24–26). For such individuals, shortened genetic counselling may be an appropriate way of supporting decisions already made about HNPCC testing.

Genetic counselling should be tailored to individual need with the option of further counselling sessions – for example, in cases where individuals have only recently learnt of their increased risk and/or show a high level of distress. Variation in preferences for level of information highlights the importance of tailoring information to clients' agendas, which may vary according to information-seeking preference (27), engagement with risk (28) and pre-existing distress (29). All participants would benefit from information about what to expect – for example, a preparatory leaflet or telephone call outlining the genetic counselling and testing process (30). Such an approach may be helpful in encouraging patients to reflect on issues not previously considered, which can then be explored more fully during shortened genetic counselling.

To our knowledge, this is the first study to examine the process of HNPCC genetic counselling. Participants in both groups appeared highly satisfied with the amount and type of pre-test counselling. Reflective elements occurred in both protocols but tended to be counsellor-led and framed around practical action and information, an approach which seems appropriate given the therapeutic options that exist for HNPCC in

contrast to HD. In the context of this controlled study, clinicians were required to adhere to a standardized counselling protocol. However, in practice, the distinction between educational and reflective components may become blurred, and a more flexible approach may be adopted. For example, counsellors could engage participants in reflection during one comprehensive counselling session by responding empathically to spontaneous cues, rather than following a pre-established agenda involving multiple switches between educational and reflective frames.

Despite the limitations of sample size, this exploratory study forms an important foundation for an adequately powered multicentre trial of different approaches to genetic counselling for HNPCC and other treatable inherited conditions. A large trial would have clear implications for health service planning, not only in terms of allocating resources effectively but also in tailoring genetic counselling to individual needs.

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