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ORIGINAL ARTICLE

Genetic consultations in primary care: GPs' responses to three scenarios

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Abstract

Objective. This study investigated general practitioners' responses to three scenarios in which patients consulted regarding genetic conditions. *Design*. Self-completed postal study. *Setting.* Primary care in Northern Ireland. *Subjects*. Questionnaire were distributed to all the GPs in Northern Ireland (n = 1079). A total of 541 GPs participated (50%). *Main outcome measures*. Responses to three scenarios in which patients consulted regarding their family history and risk of bowel cancer, breast cancer, and cystic fibrosis. *Results.* Most GPs correctly identified the patients' risk of bowel cancer, recommended regular colonoscopy, advised lifestyle changes, and did not refer to the genetic clinic. GPs who were qualified for longer were more likely to recommend colonoscopy and less likely to advise lifestyle changes. With the breast cancer patient GPs adopted a cautious approach; most would refer to the genetic and mammography clinics. With the cystic fibrosis example, most correctly identified the patient's risk of carrying the gene, would refer to the genetic clinic, and would encourage the patient to discuss the risk with his partner. In general, doctors were unsure, but would pass on genetic information to insurance companies if requested. *Conclusion*. The study suggests that, in most cases, general practitioners correctly identify at-risk individuals but there may still be some uncertainty regarding referrals. The results suggest that ways of educating GPs should be explored. Educational interventions should be linked to a greater understanding of factors involved in referral (including the influence of gender and experience). The guidelines provided to GPs in relation to the provision of genetic information to insurance companies may need to be reviewed in some countries.

Key Words: *General practice, genetic testing, primary care, referral*

Scientific understanding of the influence of genetic factors on disease has advanced rapidly. Genetic testing is now available in a variety of conditions [1]. The prediction of risk of certain cancers and carrier screening for cystic fibrosis have become important elements of primary care [2]. Patients are also aware of these advances and regard primary care as a source of information, referral [3], and reassurance [4]. In such a rapidly expanding field, it may be difficult for general practitioners (GPs) to keep up to date. Insurance companies also see the value of this information and may request details about family history and genetic conditions.

The role of the GP in offering genetic counselling or referral is, as yet, undefined. GPs' perceptions of their roles include family history taking, referral, emotional support, discussing the need for testing, and explaining the relationship between genetics and disease [5,6]. By reassuring low-risk individuals and referring people who would most benefit, GPs can

GPs are a source of information and referral for patients with concerns about genetic conditions.

- Most correctly identified increased risk, would refer to the appropriate clinics and would offer screening and lifestyle guidance.
- Older GPs were more likely to advise screening and less likely to advise lifestyle changes in bowel cancer.
- Females were more cautious in breast cancer referrals.

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improve the cost effectiveness of specialist units [7]. Not all GPs have a good understanding of genetics and may lack confidence in these roles.

Research suggests that some patients are inappropriately referred, and that low-risk patients are referred and some high-risk individuals are not receiving referrals. In a study of UK genetics centres, 27% of consultations were with individuals at population-level risk [1]. In a study of 50 GPs' referral letters, GPs did not appear to know when a patient should be referred to geneticists and had unrealistic expectations of the clinic [8]. An examination of GPs' breast cancer referrals found considerable variation in risk estimates provided [8]. A US study found that physicians provided counselling and evaluation and were unlikely to refer patients unless the patient expressed an interest in genetic testing. They concluded that more education was required to ensure appropriate referrals [10].

Most research is from the specialist perspective, with few studies of GPs' consultation and referral behaviour. This study was undertaken to investigate the factors affecting GPs' responses to three scenarios in which patients consulted with queries regarding three common conditions where genetic risk is known and where it may have an important influence on patient well-being. The aim of the study was to explore GPs' understanding of genetic risk and how this translates into a decision to refer for specialist opinion.

Material and method

All the GPs in Northern Ireland (Central Services Agency GP list – used to process salaries) received an anonymous postal questionnaire and a pre-paid return envelope. The participants returned a coded postcard separately to identify non-respondents for a targeted second mailing. The questionnaire included questions on GPs' perceptions of their roles and confidence in implementing genetic technologies [12] and described three scenarios designed by the research team based on the most common genetic consultations. The questions were piloted and tested for face and content validity in a study of 21 GPs. The data were analysed using descriptive statistics and non-parametric tests (chi-squared, Mann–Whitney and Kruskal–Wallis ANOVA) in SPSS.

Results

Response rates

There were 541 replies from 1079 questionnaires, five were not working in primary care, one had left the practice, and one was on extended leave. The response rate was 50.5%.

The characteristics of the respondents are given in Table I. The majority were male (63%; 6% declined). 13% had qualified within the previous 10 years, and only 1% had qualified within the previous 5 years. Significantly more females had qualified recently ($p < 0.001$).

Scenario 1: Bowel cancer

A man aged 30 has just heard that his father has carcinoma of the bowel and a paternal uncle (aged 55) died recently from a similar condition.

Table II gives the responses to this scenario. The majority believed that his risk was much higher than the general population and 37% believed that his risk was a little higher. Most GPs (68%) would encourage regular colonoscopy. There was an association between years qualified and responses, with those responding "yes" being qualified for longer ($p < 0.005$). The vast majority of GPs (97% and 96%) would advise dietary changes. Around three-quarters (73% and 75%) would advise a reduction in alcohol consumption and increased exercise. GPs who were qualified for longer were more likely not to advise on intake of fruit and vegetables ($p < 0.05$), and fibre ($p < 0.05$), and physical activity ($p < 0.05$). The majority of respondents would refer this patient to a surgical clinic but not the genetic clinic.

Scenario 2: Breast cancer

A 20-year-old woman tells you that her mother died from breast cancer aged 40 and a maternal aunt died from breast cancer aged 35. She wondered if she should attend the breast clinic herself.

Responses to this scenario are given in Table III; the majority, 87%, believed that the risk was much

Table I. Characteristics of respondents.

Years qualified	Male% (95% CI)	Female% (95% CI)
0–5	0	3 (1.2–5.1)
6–10	10 (7.4–12.4)	16 (11.6–20.1)
11–15	19 (15.6–22.2)	29 (23.7–34.2)
16–20	23 (19.4–26.4)	26 (21–31.2)
21–25	24 (20.3–27.5)	15 (10.9–19.1)
26+	24 (20.3–27.5)	12 (8.4–15.9)
Total	66	34

Table II. Scenario 1: Bowel cancer.

What are the risks of him developing the disease himself?	
	% (95% CI)
Same as general population	1 (0.4–1.6)
A little higher than the general population	37 (33.8–40)
Much higher than the general population	60 (56.8–63.1)
Don't know	2 (1.2–3.0)

Would you encourage this patient to have regular colonoscopy?	
	% (95% CI)
Yes	68 (65–71)
No	16 (13.8–18.5)
Don't know	16 (13.8–18.5)

Would you encourage this patient to modify each of the following aspects of his lifestyle to reduce his risk?

	Yes% (95% CI)	No% (95% CI)
Intake of fruit and vegetables	97 (95.8–98)	3 (1.9–4.1)
Physical activity	73 (70.1–75.8)	28 (24.9–30.7)
Alcohol intake	75 (72.2–77.8)	25 (22.1–27.6)
Intake of fibre	96 (94.7–97.2)	4 (2.8–5.3)

Would you refer this patient to any of the following clinics?

	Yes% (95% CI)	No% (95% CI)
Surgical clinic	82 (79.4–84.4)	18 (15.5–20.5)
Genetic clinic	29 (26.2–32)	71 (68.1–73.9)

Would you declare this family history in a 'Patient's Medical Record' form from an insurance company?	
	% (95% CI)
Yes (%)	65 (61.9–68)
No (%)	12.3 (10.3–14.5)
Don't know	22.2 (19.6–25)

Table III. Scenario 2: Breast cancer.

What are the risks of her developing breast cancer herself?	
	% (95% CI)
Same as general population	0.2 (0.0–0.5)
A little higher than the general population	12 (9.9–14.1)
Much higher than the general population	87 (84.8–89.1)
Don't know	0.4 (0.1–0.9)
Would you encourage this patient to have an annual mammogram?	
	% (95% CI)
Yes	46 (42.8–49.1)
No	39 (36–42.3)
Don't know	16 (13.7–18.4)
Would you prescribe oral contraceptives for this patient?	
	% (95% CI)
Yes	53 (59.8–56.2)
No	26 (23.3–28.9)
Don't know	22 (19.4–24.8)

Would you refer this patient to any of the following clinics?

	Yes% (95% CI)	No% (95% CI)
Mammography clinic	69 (65.9–71.8)	31 (28.2–34.1)
Genetic clinic	73 (70.1–75.8)	27 (24.2–29.9)

Would you declare this family history in a 'Patient's Medical Record' form from an insurance company?	
	% (95% CI)
Yes	72 (69.1–74.8)
No	9 (7.2–10.9)
Don't know	19 (16.5–21.5)

higher than in the general population. Females estimated a higher risk than males ($p < 0.01$); 46% would advise the patient to have annual mammography, and 16% chose "don't know". Those who responded "yes" were qualified longer ($p < 0.05$). Over half (53%) would prescribe oral contraceptives for the patient and 22% did not know. The majority would refer this patient to the mammography clinic and the genetic clinic. A chi-squared test showed that females were more likely than males to refer this patient to the genetic clinic ($p < 0.001$). The majority claimed that they would provide family history information about bowel cancer and breast cancer to an insurance company.

Scenario 3: Cystic fibrosis

A 25-year-old man attends saying he and his partner wish to have children; he is concerned about the risks of cystic fibrosis as his brother has the condition. He is not aware of any history of cystic fibrosis in his partner's family.

Table IV gives the responses to this scenario. Over half (57%) believed that the risk of his child having cystic fibrosis was a little higher than in the general population; a quarter believed that it was much higher. Males were more likely to believe that the child had a higher risk than females ($p < 0.05$). Two-thirds would encourage the patient to take a test for the cystic fibrosis gene and 89% would encourage the patient to discuss the risk with his partner. The majority (97%) would not refer the patient to the gynaecological clinic and 90% would refer him to the genetic clinic.

Discussion

In this survey GPs' responses to three genetics consultation scenarios were assessed. Most correctly identified the patients' level of risk, recommended appropriate screening, and advised lifestyle changes. The majority would not refer to the genetic clinic in the case of bowel cancer but would refer to the genetic and mammography clinics for breast cancer. Most correctly identified the patient's risk of carrying the cystic fibrosis gene, would refer to the genetic clinic, and would encourage the discussion of risk with his partner. The majority would provide genetic information to insurance companies if requested. The limitations of the study will now be outlined and the implications of the findings discussed.

The strength of this study is in the large number of responses. A response rate of 50.5% is not ideal but is typical of GP studies [12]. The gender distribution is representative of GPs in Northern Ireland [13]; however, we have no details of the distribution with regards to years qualified or age. Less experienced males may therefore be under-represented. Responses from late responders to postal surveys may differ from immediate responders [14]. Whilst we redistributed the questionnaire to non-respondents and encouraged late responses we cannot assume that the results are generalizable to the non-respondents. The scenarios represent typical presentations of common conditions and are representative of what GPs might expect in a consultation. The scenarios were short and decisions were therefore based on limited information about the patient and relatives. Furthermore, scenarios only tell us what respondents say they would do and not what they would actually do in practice.

Table IV. Scenario 3: Cystic fibrosis.

What are the risks of his child being born with the condition?		
	% (95% CI)	
Same as general population	9 (7.2–10.9)	
A little higher than the general population	57 (53.7–60.1)	
Much higher than the general population	25 (22.3–27.9)	
Don't know	9 (7.1–10.9)	
Would you encourage this patient to take a test to determine whether he is a carrier of the cystic fibrosis gene?		
	% (95% CI)	
Yes	66 (62.9–69)	
No	12 (10–14.2)	
Don't know	22 (19.3–24.6)	
Would you encourage this patient to discuss the risk of cystic fibrosis with his partner?		
	% (95% CI)	
Yes	89 (86.9–90.9)	
No	4 (2.8–5.3)	
Don't know	7 (5.2–8.5)	
Would you refer this patient to any of the following clinics?		
	Yes% (95% CI)	No% (95% CI)
Gynaecological clinic	2 (1.1–2.9)	98 (97.1–98.9)
Genetic clinic	90 (88.1–91.9)	10 (8.1–11.9)

Scenario 1: Bowel cancer

The patient presented in scenario one has a 1 in 12 risk of developing bowel cancer [15]. This is twice the population risk but the patient does not meet the Amsterdam criteria for referral for genetic testing for hereditary non-polyposis colorectal cancer (HNPCC) [16]. Most respondents (60%) correctly identified that his risk was much higher than in the general population. Colonoscopy is recommended to detect and remove polyps in carriers of HNPCC-associated mutations [16]. Most (68%) indicated that they would encourage regular colonoscopy. Some authors note, however, that the benefits of colonoscopy should be weighed against the risks associated with the procedure [15] and, again, many GPs would refer the patient to the surgical clinic to make the decision.

The links between lifestyle and bowel cancer are well known [15,17] and the fact that most GPs would recommend increased consumption of fibre, fruit, and vegetables, a reduction in alcohol intake, and increased physical activity is therefore encouraging. A particularly interesting finding was that GPs who were qualified longer were more likely to recommend colonoscopy, and less likely to advise lifestyle changes.

Scenario 2: Breast cancer

According to referral guidelines [9], this patient should have been offered surveillance with annual mammography and monthly breast self-examination but should not have been referred to the genetic clinic. Most GPs (87%) correctly believed that the patient's risk was much higher than in the general population with females estimating a higher risk than males. Although only 46% would encourage annual mammography, the majority would adopt a cautious approach, referring this patient to both the mammography and genetic clinics. Whilst this may detect early tumours in high-risk patients, low-risk patients may be referred inappropriately raising anxiety unnecessarily. The gender differences are also interesting and one-fifth of GPs did not know whether it would be appropriate for GPs to prescribe oral contraceptives (guidelines suggest that that oral contraceptives lead to a small increase in risk but that their advantages usually outweigh their disadvantages) [18].

Scenario 3: Cystic fibrosis

In this scenario the patient has an affected sibling, both parents are carriers, thus he has a 2 in 3 chance of also being a carrier. Assuming his partner has population risk, the likelihood of them having an affected child is 1 in 150 [19]. Most GPs correctly identified this to be higher than the population risk. Genetic testing would determine whether the patient was a carrier; two-thirds of respondents would encourage the patient to do this and the majority of GPs would have referred him to the genetic clinic. His partner's family history would also affect the risk estimates and so, correctly, 89% would encourage the patient to discuss the risk with her.

One of the key roles of GPs is the referral of individuals and high-risk families to genetics services. Other studies on referral decisions [1,8,10] suggest uncertainty about who should be referred and the advice provided. Our study suggests that, in most cases, GPs correctly identified those at risk. There may still be some uncertainty about referrals but in such a rapidly expanding field some uncertainly may be expected. Genetic clinics have still to establish their precise role and, as technology advances, further opportunities for genetic testing in different conditions will arise.

The dissemination of written guidelines is a common means of educating GPs in many aspects of care, but there is now evidence that guidelines alone are ineffective. De Bock et al. [9] found that GPs only partly followed guidelines for breast cancer referrals and concluded that additional forms of education were required. GPs are, however, supportive of guidelines [20,21] and studies have found that guidelines led to fewer low-risk referrals [22] and improved description of risk in referral letters. GPs also support computer-based decision programs [24,25] and initial results suggest that computer systems may lead to more appropriate management decisions [25,26].

Training courses may also improve the appropriateness of referrals [27]. However, it can be difficult to differentiate between the effects of the intervention and the accompanying packs [2]. Recent studies have analysed the additional factors influencing referrals to gain a fuller understanding of these processes. For example Hapgood et al. [28] found that GPs who believed that counselling in primary care was beneficial and that referral was not useful were more likely to provide accurate breast cancer risk estimates. This suggests that beliefs about the roles of primary care and specialists may affect the information obtained and, subsequently, the accuracy of their risk estimates.

One of the interesting features of this study was the differential response with age and gender. That older GPs were more likely to refer for colonoscopy may be considered counterintuitive – we might have expected that younger GPs would be more aware of risk and refer. Similarly, females were more likely to refer for breast cancer. While this study cannot

explain this, we might speculate that personal experience and gender influences referral behaviour. Education or guidelines alone are unlikely to change this unless linked to a greater understanding of factors involved in referral [28].

Insurance companies

As the results of genetic testing may predict lifetime risk, insurance companies would find the results invaluable, although the public are generally unsupportive [29]. In some countries the insurance industry has applied a voluntary moratorium on the use of genetic results in insurance. However, family history information is widely used in insurance underwriting and this remains a form of genetic information [29]. Many of the GPs would pass on information about family history and referrals to insurance companies although some were uncertain about how they would respond. These results refer only to the UK situation. Those countries with no legislation in this area may wish to review their guidelines so that GPs have consistent guidance.

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