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Competing interests statement

The author declares that he has no competing financial interests.

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SCIENCE AND SOCIETY

Genetic testing for cancer susceptibility: the promise and the pitfalls

Caryn Lerman and Alexandra E. Shields

Genetic testing for hereditary cancer risk is now available and has the potential to reduce cancer mortality through the targeting of preventive therapies and by motivating behavioural change. However, generating and communicating genetic information can have psychological and social consequences. As testing extends from identifying rare hereditary cancers to testing for common genetic variants that are associated with cancer risk, how do we address these complex problems to maximize the benefits of genetic testing?

The number of known genetic mutations that are associated with cancer susceptibility is growing at an exponential rate¹, and the use of genetic testing for cancer susceptibility is becoming more widespread. Genetic testing is now available for the main cancer susceptibility genes, in which rare mutations

predispose to uncommon inherited cancer syndromes, such as hereditary **breast and ovarian cancer (HBOC)**, **hereditary non-polyposis colon cancer (HNPCC)** and **familial adenomatous polyposis (FAP)**. The list of available genetic tests for hereditary cancer syndromes is shown in TABLE 1, and a detailed review of cancer syndromes and laboratories that are performing research and clinical genetic testing can be found at the **GeneTests web site** (see online links box).

The specific processes and outcomes of genetic testing for cancer susceptibility are shown in FIG. 1, which uses genetic testing for HBOC as an example. As with other forms of genetic testing for disease susceptibility², following a detailed family- and personal-history assessment, the genetic counsellor, medical geneticist or other health professional provides an individual with a genetic risk assessment, based on the information collected, and

reviews the benefits, limitations and risks of genetic testing³. The individual must then decide whether to proceed with testing and the appropriate biological samples are collected and analysed. Once the results are obtained, the individual must provide written informed consent to participate in an in-person counselling visit, during which the results are disclosed and interpreted and options for medical management are addressed.

For hereditary cancer syndromes, such as HBOC, a positive test result for a known disease-conferring mutation indicates that an individual has an increased risk of developing cancer; however, it is by no means certain that such individuals will develop cancer. By contrast, a negative test result in a family with a known deleterious mutation provides definitive information that the individual has only an average or 'general population' risk of developing HBOC. Cancer could still develop in such a person though, as a result of other genetic and/or environmental factors. In families in which the specific disease-conferring mutation has not yet been identified, a positive result for a novel mutation or a negative genetic test result would not be informative. It should be noted, however, that there are important differences in the counselling process, implications and outcomes of genetic testing for the different hereditary cancer syndromes. This review focuses primarily on HBOC and HNPCC as examples.

Recent research has begun to identify common genetic variants that augment the effects of risk-factor exposure, such as genes that affect the metabolism of hormones or that predispose individuals to behaviours that are associated with cancer risk ('cancer-risk behaviours'); for example, genes that predispose an individual to tobacco addiction. However, risk factors/behaviours that are associated with cancer, such as tobacco use and obesity, are traits that are influenced by a complex interplay of numerous genetic, psychosocial and environmental factors. Therefore, future testing for such traits would provide less information than tests that are used for hereditary cancers, as the test results would have a much higher level of uncertainty, even if positive. A comparison of the features of the main cancer-predisposing genes compared with genes that are associated with complex traits is shown in TABLE 2.

Although the full impact of genetics on clinical care is yet to be realized and remains uncertain, it is anticipated that genetic testing for cancer susceptibility could eventually allow physicians to identify individuals who are susceptible to certain types of cancer, and thereby allow them to tailor preventive and

therapeutic modalities based on that individual's genotype. Despite this potential, the clinical integration of genetic testing for cancer susceptibility poses complex psychological, social and ethical concerns. So, how is genetic testing used to determine cancer susceptibility? Also, what are the associated psychological and

social outcomes of genetic testing and what are the emerging ethical and health-care policy issues? Finally, how might we improve our knowledge of cancer genetics and apply this to genetic-testing research, giving consideration to the implications of detecting more common genetic variants?

Uptake of genetic testing
Initially, linkage analysis (FIG. 2) was used to identify genetic markers that were associated with disease susceptibility in families with a preponderance of breast, ovarian and/or colon cancer. Among the first test results to be disclosed in the clinical-research setting were those for members of families with HBOC, in which linkage was confirmed between breast and/or ovarian cancer and specific genetic markers in proximity to *BRCA1* (REF. 4). This work was soon extended to hereditary colon cancer syndromes, such as HNPCC and FAP⁵. In anticipation of the future clinical availability and use of such testing, research was conducted to examine whether individuals would be interested in genetic testing and to identify the possible factors that influence such decisions. The results of these studies showed that over 70% of individuals in the general population^{6,7} and at least 80% of respondents with family histories of cancer were highly interested in undergoing testing^{8,9}.

Unexpectedly, once testing was made available, empirical studies documented that only about 50% of the individuals at high risk for HBOC or HNPCC opted to be tested^{10–14} (FIG. 3). Rates for clinic-based high-risk populations are much more variable, ranging from 38% in a European study¹⁵ to 82% in a study carried out in the United States¹⁶ (FIG. 3).

There are many reasons why potential candidates might decline genetic testing for cancer susceptibility. These include the uncertainty that is associated with positive test results, psychological distress, concerns about family stress, lack of health insurance and concerns regarding potential discrimination^{10,11,14}. Surprisingly, relatively few individuals who received genetic testing for breast cancer susceptibility reported that they sought testing as a result of a physician recommendation¹⁷. This might be due to the fact that many physicians are not adequately prepared to recognize familial cancer syndromes or to make appropriate referrals¹⁸.

Psychosocial outcomes of testing
Although psychological and social concerns might deter some high-risk individuals from genetic testing, empirical data from controlled outcome studies in the United States¹⁴, Europe¹⁹ and Australia²⁰ have not provided evidence for significant or prevalent adverse psychological sequelae of testing. In fact, research data from families with hereditary cancer¹⁴ and from individuals who were tested in clinical settings²¹ indicate that there are positive psychological benefits

Table 1 | Hereditary cancer syndromes

Syndrome	Associated genes	Predominant tumour types or abnormalities
Hereditary breast and ovarian cancer	<i>BRCA1</i> <i>BRCA2</i>	Breast carcinomas, ovarian carcinomas
Carney complex	<i>PRKAR1A</i>	Skin pigment abnormalities, endocrine tumours, schwannomas
Cowden	<i>PTEN</i>	Breast carcinomas, thyroid carcinomas, endometrial carcinomas
Familial adenomatous polyposis	<i>APC</i>	Adenomatous polyps of the colon/rectum, gastrointestinal cancers, papillary thyroid carcinomas
Familial melanoma	<i>CDKN2A</i> <i>CDK4</i>	Cutaneous malignant melanoma, pancreatic cancers
Hereditary papillary renal carcinoma	<i>MET</i>	Papillary renal-cell carcinomas
Hereditary non-polyposis colorectal cancer	<i>MSH2</i> <i>MSH6</i> <i>MLH1</i> <i>PMS1</i> <i>PMS2</i>	Colorectal and endometrial adenocarcinomas
Hereditary diffuse gastric cancer	<i>CDH1</i>	Diffuse adenocarcinomas of the stomach wall
Juvenile polyposis coli	<i>MADH4</i>	Multiple juvenile polyps in the gastrointestinal tract, colorectal and gastrointestinal malignancies
Li–Fraumeni brain	<i>TP53</i>	Breast cancers, soft-tissue sarcomas, tumours, adrenocortical tumours, leukaemia
Multiple endocrine neoplasia type 1	<i>MEN1</i>	Primary hyperparathyroidism, pancreatic islet-cell tumours, anterior pituitary tumours
Multiple endocrine neoplasia type 2	<i>RET</i>	Medullary thyroid carcinomas, pheochromocytomas, mucosal neuromas
Nevoid basal-cell carcinoma	<i>PTCH</i>	Basal-cell carcinomas
Neurofibromatosis type 1	<i>NF1</i>	Neurofibrosarcomas, astrocytomas, melanomas, rhabdomyosarcomas, chronic myeloid leukaemia
Neurofibromatosis type 2	<i>NF2</i>	Bilateral vestibular schwannomas, meningiomas, spinal tumours, skin tumours
Peutz–Jeghers	<i>STK11</i>	Gastrointestinal-tract carcinomas, breast carcinomas, testicular cancers, gynaecological malignancies
Pheochromocytoma	<i>SDHB</i> , <i>SDHC</i> , <i>SDHD</i>	Pheochromocytomas, glomus tumours
Retinoblastoma	<i>RB</i>	Paediatric retinal tumours
Tuberous sclerosis complex	<i>TSC1</i> <i>TSC2</i>	Multiple hamartomas, renal-cell carcinoma, astrocytomas
von Hippel–Lindau	<i>VHL</i>	Renal-cell carcinomas, retinal and central nervous system haemangioblastomas, pheochromocytomas

APC, adenomatosis polyposis coli; *CDH1*, cadherin 1 (E-cadherin); *CDK4*, cyclin-dependent kinase 4; *CDKN2A*, cyclin-dependent kinase inhibitor 2A; *MEN1*, multiple endocrine neoplasia 1; *RB*, retinoblastoma; *STK11*, serine/threonine kinase 11; *TSC*, tuberous sclerosis.

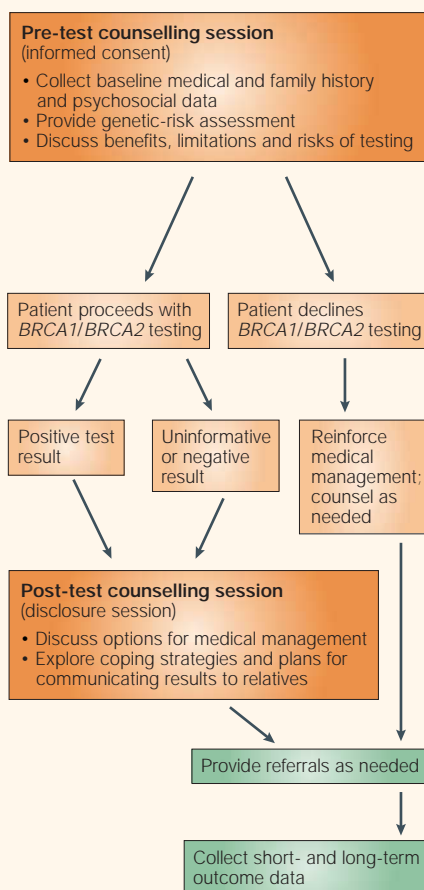


Figure 1 | Process of genetic testing and counselling for hereditary breast and ovarian cancer. The flow diagram represents the procedures for genetic testing, using hereditary breast and ovarian cancer (HBOC) as an example. Before testing an individual's DNA for the *BRCA1* and *BRCA2* mutations that are associated with HBOC, the individual participates in a pre-test counselling session to allow the counsellor to collect relevant information about the patient. At the same time, the counsellor provides information about HBOC and the impact that genetic testing could have on the individual's life. If the individual decides to proceed with the testing, a blood sample will be taken for analysis. The counsellor discloses the results of the test at a post-test counselling session, in which medical-management options and coping strategies are discussed. The counsellor will provide referrals to oncologists, surgeons and other specialists if necessary.

for members of high-risk families who test negative, such as a decrease in psychological distress. Surprisingly, there is little evidence of negative psychological effects in identified mutation carriers, possibly because many high-risk individuals already assume that they are mutation carriers, and consequently learning their mutation status is no more distressing than fearing the outcome²¹. By contrast, in one study of *BRCA1/BRCA2*-linked families,

individuals who experienced high levels of cancer-related distress during pre-counselling and then declined to be tested were at the greatest risk of increased depression²². So, the decision to defer or decline testing could actually promote, rather than alleviate, distress for some individuals.

Although the available outcome data indicate that we should be optimistic about the psychological consequences of receiving the results of cancer-susceptibility testing, there are caveats. First, more subtle effects caused by genetic testing — including anxiety, cancer-related worries, family stresses and difficulty with medical decision making — have been reported among individuals who have tested positive^{20,23}. Such effects might be sufficiently severe to compromise quality of life for some individuals²³. Second, although psychological reactions to genetic testing might be favorable overall, there are clearly a small subset of individuals who could be at risk of adverse psychological consequences such as those previously described²⁴. The development and evaluation of more intensive counselling programmes for such individuals presents another important challenge in the field.

Behaviour change following testing

A key rationale for genetic testing for pre-disposition to cancer is that a positive result might motivate high-risk individuals to alter their behaviour to reduce cancer risk. This might include increasing the frequency of cancer-screening tests and making healthy lifestyle changes, such as nutritional or dietary changes, stopping smoking, or increasing physical activity. Indeed, being informed about the need to increase cancer surveillance and prevention practices is cited frequently as a motivation for seeking genetic testing¹⁴. Unfortunately, however, available empirical data indicate that simply informing people of their genetic risk does not produce changes in cancer-related behaviours²⁵.

Most empirical studies of cancer detection and management practices following genetic testing have focused on participants in programmes that test for mutations in *BRCA1/BRCA2*, because such genetic tests were the first to be widely available (FIG. 4). Irrespective of where the testing was carried out, about 30–40% of mutation carriers do not receive the recommended mammography screening within the 12 months following testing^{12,26,27}. Rates of ovarian cancer screening among carriers with intact ovaries are even lower^{27,28} and the reasons for suboptimal rates of cancer screening among mutation carriers are largely unknown. In addition, although

prophylactic oophorectomy — the surgical removal of one or both ovaries — can reduce the risk of breast and other *BRCA1/BRCA2*-related cancers by at least 50%^{29,30}, only a small proportion of women choose this option, and rates are highly variable across studies (FIG. 4). One possible reason for the low rates of prophylactic oophorectomy is a lack of awareness of the limitations of ovarian cancer screening³¹. Although relatively few unaffected mutation carriers opt for prophylactic mastectomy^{12,27}, a positive *BRCA1/BRCA2* test following a new diagnosis of breast cancer increases the likelihood of choosing prophylactic bilateral mastectomy instead of breast-conserving surgery³². Among women with an increased risk of developing breast cancer, anxiety and cancer worries can also prompt this decision^{33,34}. Future research is needed to improve our understanding of the factors that influence these decisions among carriers of cancer-susceptibility mutations.

Less research has been conducted to examine screening practices following testing for colon cancer susceptibility, despite the fact that colonoscopy is an effective form of colon cancer prevention. Using this procedure, pre-malignant polyps can be identified and removed before the cancer becomes life threatening. Available data indicate that at least three-quarters of unaffected mutation carriers obtain recommended bowel screening³⁵. By contrast, some individuals who receive a negative genetic test for hereditary colon cancer continue to obtain unnecessarily intensive bowel screening³⁶. So, although a negative test result for a known risk-confirming mutation in a high-risk family member is informative and indicates a reduction to average risk status, some non-carriers might not feel sufficiently confident in the test results to alter their screening practices. On the other hand, some individuals who receive a negative test result might have a false sense of reassurance that could impede adherence to screening recommendations for the general or average risk population.

Ethical and social considerations

Any ethical analysis of genetic testing for cancer susceptibility must always balance the potential benefits of undergoing testing with the possible harms³⁷. Despite the potential medical benefits, there are several sources of potential harms, beyond those of psychological stress at the individual and family levels (described above). These ethical and social considerations include: breaches of privacy and genetic discrimination; racial discrimination

Table 2 | Cancer-predisposing genes versus common genetic variants

Characteristic	Genetic mutations in key cancer-susceptibility genes (such as <i>BRCA1</i> and <i>APC</i>)	Genetic variants associated with cancer-risk behaviours/complex traits
Prevalence	Rare	Common
Relative risk (penetrance)	High	Low
Attributable (population) risk	Small	Moderate to large
Aetiological heterogeneity*	Sometimes	Always
Pleiotropy†	Rare	Often
Gene–gene interactions	Possible	Likely
Gene–environment interactions	Possible	Likely

*Refers to multiple causal factors in disease aetiology. †Refers to multiple effects of a particular susceptibility mutation. *APC*, adenomatosis polyposis coli.

based on differences in the frequency of risk-conferring alleles; and limitations in the capacity of health-care providers to offer informed consent and deliver genetic services effectively.

Privacy and genetic discrimination. One of the primary sources of potential harm to patients undergoing genetic testing stems from inadequate protection of privacy and potential discrimination following disclosure of genetic information to third parties such as insurance companies and employers. Nearly two-thirds of Americans would refuse a genetic test if employers or health insurers could access the results³⁸ and many have opted not to seek medical care or file an insurance claim because they did not want to harm their job prospects³⁹. Similar privacy concerns have been documented in surveys in Canada and Europe⁴⁰.

These concerns are not unfounded. Approximately 15% of Americans who are at risk of inheriting a condition reported that they had been asked questions about genetic diseases on job applications; 13% reported that they or a family member had been fired or denied a job because of a genetic condition in the family. Also, 22% of those with a known genetic condition reported that they had been refused insurance coverage, even if asymptomatic⁴¹. In Australia, a review of the policies of life-insurance underwriters indicated that they all required that the results of genetic testing be revealed, if known by the applicant⁴². Although similar privacy concerns have been documented in surveys in Canada and Europe, the potential impact of genetic testing on access to affordable health insurance is particularly troublesome in the United States, where there is no government-sponsored health-care system.

Privacy laws must provide a system that is strong enough to reassure patients that information generated by genetic testing will not be used to discriminate against

them. In the United States, the Health Insurance Portability and Accountability Act of 1996 (HIPAA) prohibits insurers from excluding an individual from group coverage, or increasing premiums for an individual in a group plan, as a result of past or present medical conditions including those revealed through genetic testing. However, it fails to protect individuals from being denied coverage or being charged higher premiums based on genetic information when they are purchasing coverage in the individual market. HIPAA also does not include limits on insurers collecting genetic information or requiring genetic testing of those applying for health insurance of any kind. Several nations, including the United Kingdom and Canada, have recently strengthened privacy protection for health information, but few contain sufficient protections to allay patients' concerns⁴³.

Once privacy has been breached, patients must depend on anti-discrimination legislation to protect them from possible abuses. The 1990 Americans with Disabilities Act prohibits certain uses of genetic information and a 2000 Executive Order prohibits discrimination of federal employees based on genetic information. At present, no federal law in the United States bans genetic discrimination in the general population. State laws remain the primary source of protection, yet, at present, only 41 states ban genetic discrimination in group health insurance and only 31 states have passed laws that ban the misuse of genetic information by employers⁴⁴.

Although the potential for genetic discrimination in health care is considerably less in nations with a government-sponsored health-care system, such as the United Kingdom or Canada, public concern has prompted government action in other areas. The British government, for example, has issued a moratorium prohibiting insurers

from using genetic test results in setting premiums for certain life insurance, long-term care and income-protection policies until 2006 (REF 45). Failure to address patients' privacy concerns will seriously undermine our ability to integrate genetic testing into cancer prevention and treatment.

Discrimination based on race or ethnic ancestry. As an increasing number of cancer-predisposing mutations are identified, differences in the prevalence of highly penetrant risk-conferring mutations are emerging. For example, Caucasian women have been found to have a higher frequency of disease-related *BRCA1* and *BRCA2* mutations than African-American women⁴⁶. Genetic testing in American and European Jewish populations has revealed a higher prevalence of distinct founder mutations in *BRCA1*, *BRCA2* (REFS 47,48) and *APC*⁴⁹, which predisposes to FAP compared with non-Jewish individuals.

The appropriate use of racial/ethnic categories in genetic research has been intensely debated^{50–52}, particularly with respect to using socially defined categories, such as 'African American' in the context of the United States, as a proxy for ancestry. Once a particular group is identified as having a higher prevalence of risk-conferring genotypes, there are increased concerns about discrimination and stigmatization against individuals and communities⁵³. For example, mandatory screening for sickle haemoglobin initially resulted in racial stigmatization and discrimination in insurance and employment against African Americans, regardless of whether or not they had sickle-cell anaemia^{54,55}. Moreover, African Americans were targeted, despite the high frequency of sickle haemoglobin in many other communities⁵⁵. The same could occur in the context of genetic testing for cancer; for example, in Jewish individuals who have a higher probability of carrying cancer-susceptibility mutations^{47–49}.

Limitations in provider capacity to offer appropriate genetic services. As our understanding of cancer genetics increases and is used to devise strategies for managing the disease, demand for trained medical geneticists will increase. Initially, genetic counselling was developed in the context of reproductive decision making or testing for untreatable conditions, in which access to detailed counselling to facilitate informed decision making was emphasized³. As genetic testing becomes more useful in directing clinical treatment for complex conditions such as cancer, the appropriate content of

genetic counselling is likely to change. However, it will remain essential that genetic counsellors, or physicians, are able to communicate complex information to patients related to the meaning of test results, the implications for treatment choices and the social risks that are associated with testing.

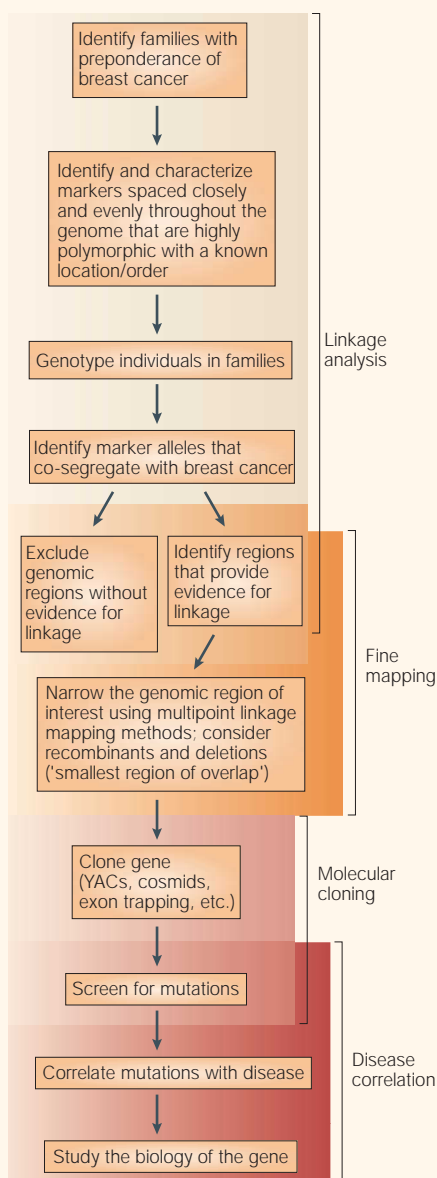


Figure 2 | Identifying genes associated with cancer. The flow diagram represents the process involved in identifying genes that are associated with familial breast cancer. Initially, linkage analysis is used to identify regions of the genome that are associated with breast cancer in individuals from families who are susceptible to the disease. These regions of the genome are examined by fine mapping to identify the associated genes, which are then cloned, sequenced and analysed for mutations. Any mutations that are identified, such as those in the *BRCA1* and *BRCA2* genes, are investigated further to establish if they are associated with breast cancer. YACs, yeast artificial chromosomes.

Despite the growing need for genetics services, there are insufficient numbers of trained medical geneticists⁵⁶; only 1,800 certified genetic counsellors⁵⁷ are trained to deal with the impact of genetic testing in the United States. Therefore, primary-care physicians, oncologists and other non-geneticists must have a greater role in providing genetic services⁵⁸. Unfortunately, however, most physicians have little formal training and limited knowledge of clinical genetics⁵⁹. In a recent survey in the United States, fewer than 30% of physicians overall and only 50% of oncologists felt qualified to provide genetic counselling. In the first study in the United Kingdom of general practitioners' attitudes towards genetic testing — including for breast cancer — 50% of respondents reported counselling patients about genetic testing in the past year, although only 21% felt sufficiently prepared to do so⁶⁰. Moreover, nearly a third of physicians in a United States study were unaware that a negative genetic test result for FAP could be a false-negative result if an *APC* mutation had not already been identified in an affected relative. This situation, which can have detrimental effects on patients, should not be allowed to continue.

Time constraints and pressures associated with managed care exacerbate the problems that arise from inadequate education of those providing the care. Although recent guidelines emphasize that genetic testing for cancer susceptibility should take place only in the context of pre- and post-test counselling², there is some evidence that providers respond to resource constraints by shortening existing protocols to a single counselling session⁶¹. Despite the lack of time and preparedness to provide genetic counselling, rates of referral to genetic specialists are low. In a national review of cases in the United Kingdom, fewer than half of patients with known high risks of genetic disorders were referred to medical geneticists⁵⁶. In a population in the United States, only 7% of patients at a heightened risk of developing cancer based on family history were referred by oncologists for genetic consultation. There is some evidence, however, that rates of referral to cancer-genetics services are increasing in some countries⁶². Clearly, further efforts to educate and support health-care providers will be essential to realize potential future benefits of genetic research in reduced morbidity and mortality.

Future directions and challenges
With the rapid emergence of cancer molecular-epidemiology research, scientists are identifying common genetic variants that predispose to cancer by promoting the carcinogenic

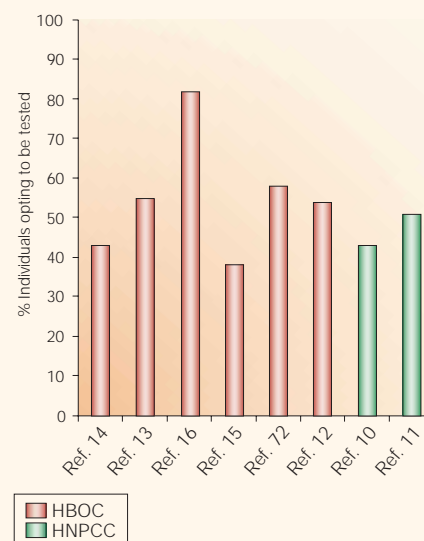


Figure 3 | Comparing uptake rates of genetic testing in families with hereditary breast and ovarian cancer or hereditary non-polyposis colon cancer. Numerous studies have been carried out to investigate the uptake rate of genetic testing for cancer susceptibility by individuals with a strong family history of disease. Although testing is widely available, studies show that, on average, only 50% of individuals with a family history of hereditary breast and ovarian cancer (HBOC) or hereditary non-polyposis colon cancer (HNPCC) choose to be tested.

effects of factors that are associated with cancer or by increasing tendencies towards cancer-risk behaviours. For example, genetic variants that alter oestrogen metabolism might influence both the efficacy and level of harm from use of hormone-replacement therapy and, therefore, have the potential to influence cancer risk⁶³. Furthermore, there is emerging evidence that genetic variation in neurotransmitter receptors, reuptake proteins, and metabolizing enzymes might contribute to the propensity to cigarette smoking⁶⁴ and obesity⁶⁵ — two of the main causes of preventable cancer mortality.

It is possible that research on genetics and cancer-risk behaviours could ultimately lead to more effective forms of individualized cancer-prevention strategies; for example, diet, exercise, pharmacological interventions and frequency of screening can be tailored to each individual based on their genotype. However, there are additional complexities inherent in the genetics of complex behaviours that might exacerbate current ethical concerns (TABLE 2). For example, genetic variants that can predispose to these cancer-risk behaviours have also been related to various other socially sensitive behaviours, including cocaine and alcohol addiction, gambling, sexual activity

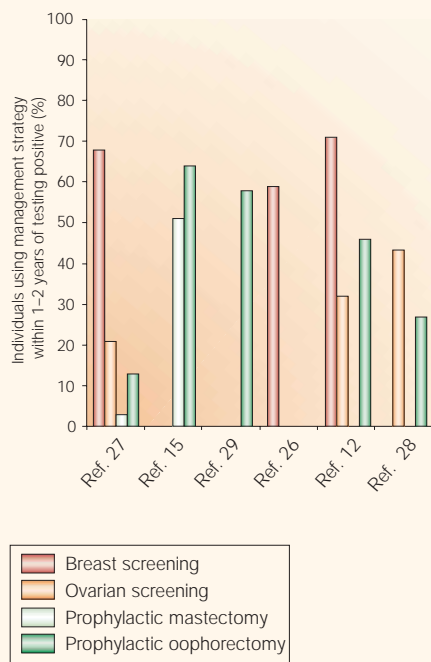


Figure 4 | Use of medical-management strategies by patients who tested positive for hereditary breast and ovarian cancer mutations. Breast screening, ovarian screening, prophylactic mastectomy and prophylactic oophorectomy are the four medical-management strategies that are available for hereditary breast and ovarian cancer. However, studies indicate that simply informing people of their genetic risk does not produce changes in cancer-related behaviours or increases in the uptake of these management strategies. If where a medical-management strategy is not shown for a particular reference in the figure, it is because the strategy was not investigated; except in Ref. 12, in which prophylactic mastectomy was also investigated and 0% of individuals opted for the strategy.

and various psychiatric conditions^{66,67}. The potential for stigmatization and discrimination could be especially pronounced for racial or ethnic subgroups in which genetic risk variants are more prevalent⁶⁸. As cancer prevention and treatment effects expand to incorporate genetic information, ethical assessments will need to address this added layer of complexity.

Despite the potential medical and psychosocial benefits of genetic testing for cancer susceptibility, the widespread application of this technology in practice faces several barriers. Concerns about privacy and genetic discrimination, particularly with respect to affordable health insurance, remain a deterrent for both patients and providers. The communication of cancer genetics research that identifies certain sub-populations must also be addressed with care, lest this information lead to sub-optimal care or increase the

risk of discrimination for members of identified communities. Further development of provider guidelines for cancer-susceptibility testing⁶⁹, as well as improved provider education⁷⁰, are needed. Finally, as the medical use of genetic testing for cancer susceptibility and these related behaviours increases, the appropriateness of population-based genetic screening will need to be evaluated⁷¹. At present, genetic testing for cancer susceptibility does not meet the criteria that have been proposed for implementing population screening, such as the existence of established data on the cost-effectiveness of screening, access to screening and preventive interventions, and adequate safeguards to ensure privacy⁷¹. It is essential that research and analysis of these ethical and policy issues be conducted simultaneously with the scientific research now underway, to ensure that ongoing genetic research is translated into improved cancer treatment and prevention efforts.

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Competing interests statement

The authors declare that they have no competing financial interests.

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