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Impact of genetic risk assessment on nutrition-related lifestyle behaviours

Jacqueline A. Vernarelli¹

¹Department of Nutritional Sciences, The Pennsylvania State University, University Park, PA 16802, USA

Abstract

Genetic susceptibility testing for common complex disease is a practice that is currently in clinical use. There are two types of gene mutations, and therefore, two varieties of genotype testing: deterministic and susceptibility. As the term suggests, deterministic genes determine whether or not a person will develop a given trait in Mendelian fashion, such as Huntington's disease. Genotype screening for such deterministic mutations has existed for decades, and is commonly used in routine medical practice. In recent years, the sequencing of the human genome has identified several 'susceptibility genes' or genes with incomplete penetrance. Mutations in these genes may increase disease susceptibility, but are not causative for disease. Genetic susceptibility testing allows unaffected individuals to obtain risk information for a variety of common complex diseases and health conditions including Alzheimer's disease (AD), CVD, cancer and diabetes. The availability of genetic susceptibility testing has increased over the past decade, and several studies are now focusing on the impact that genetic testing has on health and other lifestyle behaviours related to nutrition. The aim of this paper is to review the literature and evaluate what, if any, impact genetic risk assessment has on behaviours related to nutrition and physical activity. This paper summarises seven clinical studies that evaluated the impact of disclosing genetic risk information for disease on nutrition-related health behaviour changes. Of these seven studies, only three studies reported that health behaviour change was influenced by genotype disclosure.

Introduction

Genetic susceptibility testing for common complex disease is a practice that is currently in clinical use. There are two types of gene mutations, and therefore, two varieties of genotype testing: deterministic and susceptibility. As the term suggests, deterministic genes determine whether or not a person will develop a given trait in Mendelian fashion, such as Huntington's disease. Genotype screening for such deterministic mutations has existed for decades, and is commonly used in routine medical practice. In recent years, the sequencing of the human genome has identified several 'susceptibility genes' or genes with incomplete penetrance. Mutations in these genes may increase disease susceptibility, but are not causative for disease. Genetic susceptibility testing allows unaffected individuals to obtain risk information for a variety of common complex diseases and health conditions including Alzheimer's disease (AD), CVD, cancer and diabetes. The availability of genetic susceptibility testing has increased over the past decade, and individuals are now able to purchase direct-to-consumer genetic tests to obtain risk estimates for a variety of diseases and conditions. As access to genetic information increases, it is important to understand the potential consequences of genetic susceptibility testing on lifestyle, particularly those changes related to nutrition or exercise behaviours, some of which have been summarised previously(1). It is important to note that the studies reviewed in this paper evaluating health behaviour change following genetic risk assessment for disease are not association studies, and do not summarize how the general population will react to genetic risk assessment.

Instead, the focus of this paper provides insight as to how at-risk individuals behave in response to genetic susceptibility testing for disease. In the growing field of personalized nutrition, it is important to evaluate the behavior of at-risk individuals, as they may provide insight as to what types of health behavior changes may be expected in individuals who learn of their risk for a disease. Examining nutritional behaviors in particular is of interest given the lack of currently published information in the context of the influence of nutrition behavior on disease prevention. By understanding the impact of genetic testing on nutrition behaviors, the scientific community may be better able to determine the most effective methods of promoting disease prevention.

A literature search of the PubMed database of the United States National Library of Medicine was conducted to find studies of adult, human subjects that evaluated impact of genetic risk assessment for common complex diseases on lifestyle behaviors specifically related to nutrition. For the purpose of this paper, qualifying lifestyle behaviors included: changes in diet, use of nutritional supplements or changes in physical activity. Only full-text articles written in English were included in this review. Studies were excluded if (a) study participants did not receive the results of genetic testing; (b) prenatal or childhood genetic testing was conducted; (c) studies assessed single-gene disorders (i.e., Huntington's disease). On the basis of these search criteria, seven clinical studies that included evaluation of health behaviour changes following genetic risk assessment were identified. The seven identified studies, along with related references, are summarised in Table 1 and later in the text.

Genetic susceptibility for common complex diseases

Following the sequencing of the human genome, genetic susceptibility testing for several complex common diseases has been identified. Genetic variants that increase the risk for various cancers (including breast, ovarian, oesophageal and colorectal), obesity, diabetes and AD are all known. Mutations resulting in a predisposition for various types of cancer development were among the first to be identified. One of the first genetic susceptibility tests to be widely used in both research and clinical practice is screening for mutations in the BRCA1/2 tumour suppressor genes to confer increased risk of breast and ovarian cancer. Mutations in the BRCA1 or BRCA2 gene confer a 10–87% lifetime risk for development of breast or ovarian cancer(9). Hereditary non-polyposis colorectal carcinoma is associated with multiple gene mutations, some of which confer a lifetime risk of disease development over 80%(10,11). Mutations in the mitochondrial aldehyde dehydrogenase 2 gene are associated with risk for development of alcohol-related cancers. In particular, individuals who are heterozygous for the ALDH2*2 allele have an increased risk for development of oesophageal cancer(12,13). ApoE is an apoprotein that has an essential role in lipid transport. Mutations in the apoE gene have been implicated in the causal pathway for several diseases, including AD, metabolic syndrome, cognitive disorders, immunoregulation and atherosclerosis(14). The presence of the e4 allele increases the risk for developing AD 5–15-fold, depending on the number of copies present(15). Extensive research conducted at Boston University has led to the development of risk curves for development of AD based on mutations of the apoE gene. The risk curves allow for individualised lifetime risk estimates for AD that are based on not only genotype but also age, sex and race(15). Genetic risk assessment for AD is currently available only for research use, and currently not available for clinical practice. More recent research has identified genetic variations that result in disease risk for other complex syndromes, including obesity. Recent studies have focused on variation in the fat mass and an obesity-associated gene (FTO) as a genetic determinant of obesity(16,17), although risk varies greatly based on ethnicity(18).

Genetic risk assessment and behavior change

The influence of genetic risk assessment and genotype disclosure on behavior change has been extensively reviewed by Marteau and co-workers(19,20). The most recent systematic review provides substantial information regarding the influence of genetic tests on a wide variety of health behaviour changes, and provides useful information for further reading. Of the studies that have evaluated behaviour change following genetic risk assessment, very few studies have evaluated the impact of genetic risk assessment on diet and exercise behaviours. Instead, the majority of studies have focused on other lifestyle behaviours, such as smoking cessation or cancer screening uptake. A summary table of studies included in this review is presented in Table 1.

Cancer

A multitude of studies have been conducted to assess health behaviour changes following risk assessment for breast cancer, with the majority of studies addressing behaviour changes related to update of prophylactic surgery, medication or screening(21–28). In the case of colorectal cancer, the study of Ceballos et al.(29) evaluated the motivation of participants to increase screening uptake following genetic risk assessment. Other studies evaluating changes in lifestyle behaviours following risk assessment for cancer evaluated changes in smoking behaviours. McBride et al.(30) conducted several studies to evaluate the influence of genetic risk assessment for cancer predisposition on smoking behaviours. In a study of 557 African American smokers attending an urban health clinic in the United States, smokers who were provided with genetic risk information were more likely to report smoking cessation attempts at 6 months post genetic disclosure than those who were not provided with genetic risk information. However, by 12 months, no differences between groups were observed. In the study by Carpenter and co-workers, the study personnel provided a1 antitrypsin deficiency genetic testing to healthy smokers; where positive a1 antitrypsin gene mutation confers increased risk of emphysema following cigarette exposure(31,32). The study found that smokers, who were told that they had a mutation associated with severe a1 antitrypsin deficiency, were significantly more likely to report smoking quit attempts immediately after disclosure than non-carriers or unaffected carriers in the short term. Three months postgenotype disclosure, however, there were no differences in quit attempts between groups. This may reflect in part the difficult nature of making a health behaviour change in regards to smoking cessation(33).

Only two studies focused on changes in diet and physical activity behaviours following BRCA1/2 screening and genotype disclosure. In the results reported by O'Neill et al.(2), 115 women with a family or personal history of breast cancer were screened for BRCA1/2 mutations and subsequently evaluated for post-genotype disclosure behaviour changes in diet and exercise; see Table 1. Of the women screened, 40% were positive for the mutation; 40% had an ambiguous test result (owing to the lack of comparison with an affected relative) and 20% were negative. Lifestyle habits pertaining to diet (including fat intake) and exercise behaviours were surveyed at baseline, 3 and 6 months post-genotype disclosure. Repeated measures analysis was used to evaluate differences across time. The authors did not find any differences in diet or exercise behaviors at any time point among the genotype groups.

Similarly, Quach et al.(3) evaluated the nutrition behaviors of 120 Ashkenazi Jewish individuals with a family or personal history for breast/ovarian cancer following genotype testing for BRCA1/2 mutations, presented in Table 1. Participants were evaluated at baseline and 6 months post-genotype disclosure. The authors did not observe any differences in diet or exercise behaviours related to BRCA1/2 testing during the follow-up visit.

A third study, although not a clinical trial, evaluated changes in dietary behaviors following risk assessment for 'alcohol-related cancers (e.g. oesophageal cancer)(7). This study randomised 200 Asian college students to receive information regarding cancer risk with or without genotype disclosure. Data on dietary behaviours (specifically alcohol consumption) were evaluated at baseline and at 1-month post-risk disclosure. The authors found that participants who learned that they were positive for a genotype that confers increased cancer risk reported a significant decrease in alcohol consumption between baseline and the 30 d follow-up period.

CVD

Marteau *et al.*(4) conducted a study involving adults with a family history of familial hypercholesterolaemia were randomized to receive risk assessment by clinical diagnosis alone or clinical diagnosis plus genetic testing. During subsequent follow-up visits, participants provided information about risk perceptions as well as lifestyle behaviors. The study found that there are no differences between the disclosure groups in terms of changes in diet (assessed as fat and saturated fat intake) or exercise behaviors 6 months after receiving the information. In another study of ninety-eight African-American women randomized to receive genetic information about hypertension risk, Taylor & Wu(8) evaluated post-disclosure changes in diet and physical activity. The authors found that women who learned that they were at an increased risk for hypertension reported increased physical activity between baseline and the 6-month follow-up period; however, the difference in physical activity were not statistically significant.

Diabetes

Presently, very little evidence has been published regarding the relationship between genetic risk assessment for diabetes and subsequent health behavior changes. One study evaluates the potential impact of such testing by surveying twenty-two adults at a risk for development of Type II diabetes (risk defined by the presence of one or more of the following: abnormal fasting blood glucose levels, history of hypertension or dyslipidaemia, elevated body weight status) on the potential impact of additional genetic risk assessment. The authors found that subjects responded that disclosure of genetic risk information would motivate them to make risk-reducing health behaviors. However, given the hypothetical nature of this study, it is impossible to speculate whether or not genetic risk assessment would have resulted in any lifestyle behavior changes(34).

Alzheimer's Disease

The Risk Evaluation and Education for Alzheimer's disease (REVEAL) study is an ongoing series of multi-centre randomized clinical trials that examine the impact of genetic risk assessment (including apoE genotype disclosure) on cognitively normal adults(35,36). The REVEAL trial was the first to evaluate the disclosure of genetic risk information for a complex, progressive disease with no known method of treatment or cure. Several data collection methods were used, including validated survey tools to collect information about health behaviors, nutritional choices and exercise habits. Additional psychological data were collected using various validated tools to collect information about cognitive status, stress and depression(37). High response rates and detailed follow-up visits allowed for excellent data collection and provided researchers with a robust dataset. Details of the overall REVEAL study, including the clinical trial rationale, study design, survey tools and other results have been previously described elsewhere(6,36). The REVEAL study provides an instructive context to examine health behavior changes following genetic risk assessment. In the first REVEAL trial, Chao *et al.*(5) determined that participants who learned that they were at an increased genetic risk for AD were approximately three times as likely to report

making an AD-related health behavior change than those who are apoE e4 negative; see Table 1. In this study, health behavior change was assessed using a composite variable that encompassed changes in diet, exercise, supplements and medications, and was derived from a relatively small, homogeneous study sample. Subsequent results from the second REVEAL trial(6) involving 253 cognitively normal adults with a family history of AD were provided with apoE genotype disclosure. The study indicated that adults who were e4+ were over twice as likely to report making an AD-related change in nutrition behaviour following genotype disclosure than those who were e4-. Subsequent analysis determined that this change was driven by a change in supplement use among e4+ participants, where those testing positive for the risk-increasing allele had 4.75 times the odds of reporting a change in dietary supplement use (95% CI 2.2 10.1; P<0.0001). Interestingly, there were no significant differences in reported changes in overall dietary habits or exercise behaviors between e4- and e4+ participants. The authors speculated that part of the behavior changes observed in this trial may have been due to the popularity of supplements marketed for 'brain health' at the time of data collection, despite the lack of scientifically valid studies indicating that supplements were useful in the prevention of AD. Data from both REVEAL trials have been the first to demonstrate a robust change in nutrition-related behaviors following genetic risk assessment. Currently, the fourth REVEAL trial is underway (NCT01434667) and will evaluate the impact of genotype disclosure on additional health and lifestyle behaviors.

Ongoing and future studies

A search of the Clinical Trials Registry (<http://www.clinicaltrials.gov>), a multi-national registry of over 128000 clinical trials from over 180 countries (at the time of publication) was conducted to identify current trials evaluating the impact of genetic risk assessment on health behavior changes. Several trials were identified, and involved a variety of complex conditions, such as AD (NCT01434667), obesity (NCT01355224), heart disease (NCT01577719), several types of cancer (NCT00782366, NCT00150917 and NCT00287898) and diabetes (NCT01186354, NCT01060540 and NCT01034319). Complete descriptions of two trials (22013171 and 3280160), in which researchers are evaluating whether genetic risk disclosure for Type 2 diabetes leads to changes in dietary intake, diet patterns or exercise behaviors, have recently been published. At the time of this publication, all of the aforementioned studies were ongoing, and no results have been published.

Summary

Genetic risk assessment is a relatively new development in the medical world. The influence that test results have on health behavior changes is relatively unknown. Health behaviour outcomes are of particular importance to public health research, as communication of disease risk may prompt specific health behavior changes. In this review of the literature, it was observed that the influence of genetic risk assessment on health behavior changes differed by disease model and disclosure type. The majority of studies reviewed for this paper indicate that genetic risk assessment for disease is unlikely to lead to health behavior change, a finding similar to those in a review of health behavior changes following direct-to-consumer genetic test results(38). There are several reasons for these negative findings, some of which may be explained in part by various theories of health behavior, including the health belief model(39) and Leventhal's common sense model(40). Making changes to dietary behaviors, physical activity or smoking practices generally require changes to behavioral capacity and self-efficacy. Using the health belief model theories, one could suggest that the perceived susceptibility and/or severity for disease development were not

enough to overcome the perceived barriers to behavior change in disease models with known mechanisms for treatment.

Only one group of studies summarized in this paper demonstrated a robust response in nutrition-related lifestyle behavior changes related to nutrition following genetic risk assessment(5,6). The response of adults who learned that they were genetically susceptible to develop AD differed from the response of adults in any other study of any other disease model. Unlike other disease models, the presence of the e4 allele of apoE gene confers a clearly documented susceptibility for AD, a severe disease with no known treatment or cure. It is possible that the perceived severity of AD is greater than that of other diseases, such as familial hypercholesterolemia or even various cancers, because there are no known treatments for AD. The four perceptions of the health belief model (perceived severity, susceptibility, barriers and benefits) that influence health behavior changes are readily identifiable in the context of these studies. When looking at the perceived severity of AD, it is not unreasonable to extrapolate that for first-degree relatives of patients with AD, the disease itself seems exceptionally severe for participants in the REVEAL II study. The magnitude of perceived severity may be even greater among first-degree relatives who also served as primary caregivers for the AD patient. It is also possible that there is an age-related effect pertaining to nutrition-related behavior changes; middle-aged relatives of AD sufferers may be more likely to report behavior changes as an attempt to prevent the onset of disease in later life. It is important to note that there is no scientific evidence that supports the use of dietary supplements as a method of treating or preventing AD. Surprisingly, in the findings published by Alamian involving women who underwent genetic testing for breast cancer, it was women who received inconclusive results, as opposed to those testing positive or negative for the BRCA mutation who were more likely to report supplement use, potentially suggesting that women with perceived susceptibility for disease development based on family history alone may make health behavior changes regardless of genotype status(41). Nicher and Thompson(42) observed this same concept in a study conducted to survey supplement users. The authors found that one participant reported the use of over five dietary supplements as a method for preventing breast cancer; her risk perception was based on family history alone, without ever having genetic testing performed. The authors point out that current societal beliefs suggest that supplementing the diet may help aid in the promotion of wellness as well as the prevention of illness, especially since surveys have demonstrated that adults generally perceive dietary supplements and medical foods to be safe(43). Additionally, changes to dietary supplement use require fewer psychological and behavioural barriers than making changes to dietary intake or smoking behaviours, and, therefore, it may be more likely that changes in supplement use would be observed in any studies conferring increased risk for disease, not just studies that disclose genotype information.

Conclusions

Despite the recent surge in popularity of personalized medicine, research regarding the impact of genetic testing on health behaviors is very limited. Application of the health belief model to predicting health behavior changes in these types of situations is difficult: little is known how the public (let alone individuals) perceive genetic test results. The majority of studies evaluating health behavior changes have had negative results, with very few studies evaluating health behavior changes related to nutrition. The current state of the literature demonstrates a clear need for future research to evaluate health behavior changes following genetic risk assessment, particularly as the availability of direct-to-consumer genetic testing continues to increase.

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Table 1
Summary of studies evaluating nutrition-related changes following genetic risk assessment

Reference	Condition	Study population	Lifestyle behaviours assessed	Assessment time points	Findings
O'Neill <i>et al.</i> ⁽²⁾	BC	115 women with family or personal history of BC receiving BRCA1/2 screening	Diet; Physical activity	Baseline*, 3 months and 6 months	No differences in diet, physical activity in BRCA 1/2+ v. BRCA1/2- women
Quach <i>et al.</i> ⁽³⁾	BC	120 Ashkenazi Jewish individuals with family or personal history of BC receiving BRCA1/2 screening	Diet; Physical activity;	Baseline, 6 months	No difference in diet or exercise behaviours
Martean <i>et al.</i> ⁽⁴⁾	FH	341 families with a history of FH randomised to receive risk information for FH with or without genetic assessment	Diet; Physical activity; Medications; Smoking	Baseline, 1 week, 6 months	No difference in diet, physical activity, smoking, or medication use between groups at 6 month follow up
Chao <i>et al.</i> ⁽⁵⁾	AD	162 adults with a family history of AD receiving risk assessment for AD (with and without apoE disclosure)	Composite variable that included any change in diet, exercise, vitamins and medications	Baseline, 12 months	Adults who were underwent genotype disclosure and learned they had the high-risk allele were more likely to report AD-related lifestyle behaviour changes than those who were apoE4 negative
Vernarelli <i>et al.</i> ⁽⁶⁾	AD	272 adults with a family history of AD receiving apoE genotype disclosure	Diet; Physical activity Dietary supplement use; Medication use	Baseline, 6 weeks	Adults who learned they had the high-risk allele were more likely to report changes in dietary supplement use than those who were apoE4 negative
Hendershot ⁽⁷⁾	Cancer (alcohol-related, e.g. oesophageal)	200 Asian college students randomised to receive risk information for increased cancer risk with and without genotype disclosure	Alcohol consumption	Baseline, 1 month	Subjects who learned they were at increased genetic risk for alcohol-related cancers reported significant decrease in alcohol consumption from baseline to 1 month
Taylor & Wu ⁽⁸⁾	Hypertension	98 African-American women undergoing genetic risk assessment for hypertension	Diet (sodium, potassium intake); Physical activity; BMI	Baseline, 6 months	Women at risk for hypertension reported increased physical activity, though finding was NS.

FH, familial hypercholesterolemia; AD, Alzheimer's disease; BC breast cancer.

* For each study, 'baseline' refers to a time point pre-genotype disclosure.