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## Patient responses to genetic information: studies of patients with hereditary cancer syndromes identify issues for use of genetic testing in nephrology practice

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### Abstract

Advances in the genetic basis of kidney disease may mean that genetic testing is increasingly important in reducing disease morbidity and mortality among patients. However, there is little research examining patient responses to genetic information for Mendelian and common kidney diseases. Existing research on kidney and other hereditary cancer syndromes can inform three major issues relevant to the nephrology context: (1) how patients understand their risk of disease following genetic counseling and testing; (2) their emotional responses to the information; and (3) their uptake of recommended risk-reducing strategies. Prior research suggests that genetic counseling and testing may improve patient understanding of genetics, but patients still might not fully understand the meaning of their results for disease risk. Genetic counseling and testing does not appear to result in long-term negative emotional effects among patients who carry mutations or those who do not. Finally, while genetic counseling and testing may improve adherence to recommended screening strategies, adherence varies substantially across different risk-reduction options. Previous research also suggests that computer-based interventions might be a useful adjunct to genetic counseling approaches. Examining whether and how these prior findings relate to the context of hereditary kidney disease is an important area for future research.

### Keywords

hereditary cancer syndromes; genetic counseling; risk perception; screening behaviors; health literacy

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## Introduction

The completion of the human genome project, and associated advances in genetic research, has allowed the identification of gene mutations that are associated with hundreds of disease outcomes, making it possible to determine whether unaffected individuals are at increased risk of disease.<sup>1</sup> Currently, over 1500 genetic or inherited diseases have a clinically based molecular test available.<sup>2</sup> Predictive genetic testing can potentially allow for focused prevention and screening programs, as well as personalized treatment.

Among the human disease genes that have been identified are genes involved in disorders that disrupt the structure, function, or developmental patterning of the kidney and urogenital tract.<sup>3</sup> Autosomal dominant polycystic kidney disease can result from mutations in the genes *PKD1* and *PKD2*; 300,000 individuals in the U.S. have or are at risk for developing this condition.<sup>4,5</sup> A number of hereditary kidney cancer syndromes have been identified with known gene mutations, including von Hippel-Lindau disease, hereditary papillary renal cancer, Birt-Hogg-Dubé syndrome, and hereditary leiomyomatosis renal cell carcinoma syndrome.<sup>6,7</sup> Patients affected by these syndromes generally have a younger age of presentation and multifocal and bilateral renal lesions.<sup>8</sup> While hereditary cancer syndromes account for less than 5% of kidney cancers, identification of these patients is critical, as early screening and surgical treatment may improve disease-related morbidity and mortality.<sup>6,9</sup> Screening may lead to earlier detection of renal tumors at a stage when treatment may be more effective and a greater number of surgical options can be considered.<sup>10,11</sup> However, patients from families with hereditary kidney cancer syndromes and their physicians often face difficult decisions regarding cancer control and quality of life.<sup>12</sup> Despite this, little research has examined how patients respond to genetic testing for hereditary kidney cancers or other kidney-related diseases.

Discussions of how genetic information might be used with patients in nephrology contexts can be informed by research conducted with patients affected by hereditary cancer syndromes. This review will focus on patient responses to genetic counseling and testing for hereditary breast and ovarian cancer (HBOC) and hereditary nonpolyposis colorectal cancer (HNPCC), also called Lynch syndrome, as the patient outcomes of predictive genetic testing have been extensively studied for these diseases.<sup>13</sup> More specifically, the review will first briefly summarize the implications of HBOC and HNPCC genetic testing for patient care. Then, existing research on patients' cognitive (e.g., risk perception, recall), emotional (e.g., distress, worry, anxiety) and behavioral responses to this genetic information will be discussed. The review will then present evidence concerning how the educational content and delivery of genetic information to patients affect these outcomes. The increasing importance of considering how patients' health literacy, or their health-related skills and knowledge, might affect their responses to genetic information will also be addressed, as over one-third of U.S. adults have limited health literacy.<sup>14</sup> The implications of existing research findings for the context of hereditary kidney diseases will be discussed.

## Implications of HBOC and HNPCC genetic information for patient care

The hereditary cancer syndromes HBOC and HNPCC have been extensively described elsewhere.<sup>15</sup> Briefly, for HBOC, mutations in the genes *BRCA1* and *BRCA2* can lead to greatly increased risk for both breast and ovarian cancer.<sup>15</sup> *BRCA* genetic testing is currently recommended for those individuals with a family history suggestive of the presence of a gene mutation.<sup>16</sup> Following genetic testing, patients carrying a mutation and their physicians must make decisions regarding various risk-reduction options, including prophylactic surgery, which can greatly decrease cancer risk,<sup>15,17</sup> screening (e.g., mammography, ovarian cancer screening), and chemoprevention (e.g., tamoxifen use). Similarly, HNPCC, which results from

a germline mutation in one of the identified DNA mismatch repair genes, substantially increases cancer risk, most notably for colorectal, endometrial, gastric, and ovarian cancers.<sup>15</sup> Mutation carriers are advised to screen more frequently, while, for both HBOC and HNPCC, individuals found not to carry a familial mutation are advised to follow general population screening recommendations.<sup>18</sup>

Currently, genetic testing for hereditary cancer syndromes is generally in conjunction with comprehensive pre- and post-test genetic counseling by a trained genetic counselor.<sup>19</sup> Briefly, pre-test genetic counseling is a process of informed consent that includes discussion of possible outcomes of testing, as well as benefits, risks and limitations of testing. In post-test counseling, the results and their significance are discussed, and medical management is reviewed, including screening and treatment options.<sup>20</sup> The aims of genetic counseling for conditions such as cancers include: educating patients about the genetic condition, improving the accuracy of their risk perceptions, and encouraging adoption of risk-reducing behaviors.<sup>21,22</sup> The process of genetic counseling and testing therefore raises a number of important issues, which would be salient as well to the context of hereditary kidney disease: (1) how patients understand their disease risk following genetic testing; (2) their emotional responses to the genetic information; and (3) whether they engage in recommended risk-reducing behaviors following testing. In the next section of the review, each of these issues will be addressed in turn.

## Patient responses to genetic information

### Understanding of disease risk following genetic counseling and testing

Existing research studies have examined how patients understand their disease risk following genetic counseling and testing for hereditary cancer syndromes and, to a lesser extent, whether patients recall genetic information.<sup>13</sup> In a meta-analysis, Braithwaite et al. (2004) observed that, in controlled trials, genetic counseling improved knowledge of cancer genetics but did not lead to changes in perceived risk of disease.<sup>23</sup> A more recent review found that genetic counseling may lead to more accurate disease risk perceptions among at least some patients, and that these effects were sustained at one-year follow-up.<sup>24</sup> However, a substantial proportion of patients continued to either overestimate or underestimate their cancer risk even after counseling. Other studies have found no differences in perceived risk of HBOC or HNPCC between mutation carriers and non-carriers at 12 months after genetic testing.<sup>1</sup>

When examining only studies focused on HBOC, reviews have found that genetic counseling generally increases patients' knowledge of breast cancer genetics,<sup>25</sup> and that genetic counseling and testing for familial breast cancer appears to improve somewhat accuracy of perceived risk among patients.<sup>26</sup> For example, in a study of 450 women seen at a familial breast cancer clinic, those who overestimated their disease risk before genetic counseling had significantly lower risk estimates after counseling, but still tended to overestimate their cancer risk; those who underestimated their risk initially had significantly higher risk estimates after counseling, but still underestimated their risk.<sup>27</sup> These findings suggest that biased processing of genetic information may be occurring, such that patients might not fully adjust their estimates of their disease risk to reflect the information that they receive from health care providers.<sup>28</sup>

Perceived risk may be a critical variable; one study found that patients with accurate risk perceptions for colorectal cancer were significantly more likely to have undergone appropriate screening compared to those with inaccurate perceived risk.<sup>18</sup> In the context of HNPCC, research has shown that mutation carriers had a slight increase in perceived risk of colon cancer following genetic counseling and testing, while non-carriers had a significant reduction in perceived risk.<sup>29</sup> In another study, nearly all patients remembered whether they had inherited a familial mutation at one-year follow-up, but mutation carriers understood their colorectal cancer risk significantly less than non-carriers.<sup>30</sup>

In summary, research has shown that genetic counseling and testing appears to improve knowledge of cancer genetics, but patients might not always correctly interpret their test results. There is evidence that genetic counseling and testing somewhat improves accuracy of perceived cancer risk.<sup>31</sup> However, none of the existing studies have taken into account patients' health literacy, and particularly their numeracy, or basic quantitative skills, which could certainly impact adjustment of risk perceptions.<sup>28</sup> Existing research findings suggest that genetic counseling and testing for hereditary kidney diseases might lead to improved understanding of disease, but that patients might not fully understand the meaning of their genetic test results for disease risk. These outcomes need to be examined among nephrology patients with a greater range of numeracy skills and background knowledge regarding genetics and kidney disease.

### Emotional responses to genetic information

Studies of patients' emotional responses to genetic information for hereditary cancer syndromes have generally looked for possible harmful outcomes (e.g., distress, anxiety, worry). In a 2005 review, Meiser et al. found that unaffected mutation carriers generally had no adverse psychological effects, while unaffected non-carriers had reductions in negative emotional outcomes.<sup>32</sup> Others have observed no effects of genetic counseling on general anxiety or cancer-specific worry in controlled trials, although there is some evidence of short-term reductions in these outcomes in studies with prospective designs.<sup>23</sup> Heshka et al. (2008) found no evidence of long-term effects of genetic testing for carriers or non-carriers on general distress, cancer-specific distress, anxiety or depression, although some short-term increases were observed.<sup>1</sup>

Comprehensive reviews and long-term studies focused on familial breast cancer have suggested that adverse psychological outcomes of genetic counseling and testing are uncommon, although a subgroup of individuals may be at increased risk for negative effects.<sup>33</sup> An evidence review found that, overall, more studies showed decreased breast cancer worry or anxiety after risk assessment and testing.<sup>34</sup> Butow et al. (2003) observed differences in the effects of genetic counseling and testing for familial breast cancer by carrier status; mutation carriers did not experience significant increases in depression and anxiety, while non-carriers experienced reductions.<sup>26</sup> Unaffected mutation carriers may experience a short-term increase in distress after learning their test results, with a return to pre-test levels over time, while non-carriers may have more immediate and sustained psychological benefits.<sup>31,35</sup> Some mutation carriers may feel strong relief after testing,<sup>36</sup> although only low levels of benefit finding have been reported.<sup>37</sup> In the context of HNPCC, studies have consistently shown that unaffected non-carriers experience psychological benefits, with short- and long-term decreases in anxiety and depression, while no adverse long-term effects have been observed among unaffected carriers.<sup>32,38</sup>

Therefore, the existing literature suggests that genetic testing, when conducted in concert with comprehensive genetic counseling, may lead to reductions in emotions like anxiety, distress and depression among unaffected non-carriers, with no long-term adverse effects among unaffected carriers. These findings therefore suggest that long-term negative psychological outcomes of genetic testing for hereditary kidney diseases are unlikely, although no research has directly addressed this question to date. Future studies are also needed to examine the emotional responses of nephrology patients with limited health literacy, whose lower levels of baseline skills and knowledge might affect both cognitive and emotional responses to the large amount of information generally given during the genetic counseling and testing process.

## Uptake of screening and surgical options following genetic testing

Much recent research has focused on examining patient decision making for and use of risk-reduction strategies following genetic counseling and testing for hereditary cancer syndromes. In their review, Beery et al. (2007) found that genetic testing was associated with increased adherence to surveillance and screening guidelines in some populations, with greater use of risk-reducing surgeries among mutation carriers than non-carriers.<sup>13</sup> Substantial differences have been observed across risk-reduction options and disease outcomes, however.

Heshka et al. (2008) observed that mammography rates generally increased in carriers after disclosure of *BRCA1/2* test results compared to rates pre-disclosure, with use of both breast and ovarian cancer screening generally higher in carriers than non-carriers.<sup>1</sup> Carriers' use of different screening strategies has varied substantially, however, with higher rates of mammography use than ovarian cancer screening.<sup>1</sup> Although U.S. studies published since 2003 indicate a shift toward greater uptake of prophylactic surgery in *BRCA1/2* mutation carriers, the majority of women are still opting for breast cancer surveillance as their primary risk management strategy.<sup>33</sup> Hopwood (2005) observed that, although use of screening seemed to increase appropriately in mutation carriers, adherence still might not be optimal for carriers or for non-carriers who are at population risk.<sup>31</sup> Observed effects of genetic information regarding carrier status for *BRCA1/2* on screening, chemoprevention, and surgical outcomes are summarized in Table 1.

Studies examining responses to genetic information for HNPCC have focused on colonoscopy use. Use of colonoscopy has generally been found to increase in the 12 months following genetic testing in carriers compared to non-carriers.<sup>1</sup> One recent prospective cohort study observed that carriers increased their use of cancer screening and non-carriers decreased their use as recommended.<sup>29</sup> The few studies investigating gynecological screening in this population have noted increased adherence after genetic testing.<sup>13</sup> Studies examining use of screening following genetic counseling and testing for HNPCC are summarized in Table 1.

In sum, studies to date have suggested that the majority of both carriers and non-carriers adopt appropriate screening and preventive behaviors following genetic counseling and testing for hereditary cancer syndromes.<sup>32</sup> However, rates of use vary greatly across different risk-reduction options, and a notable proportion of mutation carriers are not adherent to recommendations. Therefore, these findings suggest that genetic testing for hereditary kidney diseases might somewhat improve adherence to recommended screening such as ocular evaluation or abdominal imaging. The challenges of hereditary kidney disease, such as younger age of onset,<sup>8</sup> may require additional interventions to achieve recommended use. It is also important to note that almost all of these studies have included populations of highly educated, Caucasian women, generally seen in academic specialty centers, and it is unclear to what extent these results will generalize to broader patient populations. Decision making regarding risk reduction options may be substantially different in populations with limited health literacy, who have lower levels of background knowledge, of oral literacy skills required to interact effectively with physicians, and of numeracy skills required to interpret risk information.

## Effects of educational strategies on patient responses to genetic information

The final section of this review will address how the educational content and delivery of genetic information may play a critical role in patients' understanding of, and subsequent responses to, their test results.<sup>18</sup> Research examining the content of traditional genetic counseling sessions is limited, but considerable variation has been observed across sessions.<sup>39</sup> Edwards et al. (2008) found that adding content related to genetic risk was effective in improving knowledge and perceived risk, but that the effects of risk communication interventions on emotional, behavioral and health outcomes was unclear.<sup>40</sup> Audrain et al. (1998) found that patients value

informational elements in genetic counseling.<sup>41</sup> Another study found that discussing prophylactic surgery during genetic counseling sessions resulted in women from high-risk breast cancer families having significantly more expectations met.<sup>42</sup>

Another line of existing research has investigated different delivery approaches for genetic information, mostly focused on identifying alternatives for educating patients on the basics of inheritance.<sup>22</sup> A recent review of 13 intervention studies, which generally compared individualized genetic counseling sessions to another type of educational intervention, found that computer-based and video educational interventions led to the greatest improvement in genetic knowledge and satisfaction compared to the other approaches, and the computer-based interventions had better outcomes on psychological measures.<sup>43</sup> However, this research base is limited, as almost all studies have focused on familial breast cancer, and have generally not examined behavioral outcomes. The main findings of studies comparing educational approaches for genetic information related to breast and ovarian cancer risk are presented in Table 2.

In summary, limited research has been conducted to date to examine the effects of educational content and delivery of genetic information on patient outcomes. Initial research findings that have potential use in the nephrology context are that computer-based interventions might have the potential to educate patients about genetics and disease basics. However, as with other studies in this literature, participants in these studies have generally been highly educated and Caucasian, and there is no evidence regarding how patients with limited health literacy might understand and subsequently use presented genetic information. Additional research is needed on hereditary kidney diseases, as patients might have different reactions to genetic information related to conditions other than HBOC. Future studies could also examine how interventions can reach younger patients before the onset of disease.

## Conclusions

Despite the increasing importance of advances in the genetic basis of kidney disease, little research has examined how nephrology patients might respond to such genetic information. However, existing research on patient responses to information on hereditary breast and ovarian cancer and hereditary nonpolyposis colorectal cancer can shed light on three major issues: how patients understand genetic information; their emotional responses to it; and their uptake of recommended risk-reduction strategies. With regard to the first issue, comprehensive genetic counseling and testing seems to improve patients' understanding of genetics, but they still might not fully understand the meaning of genetic test results for their disease risk. Second, genetic counseling and testing does not appear to result in long-term negative emotional effects among either mutation carriers or non-carriers. Third, while genetic counseling and testing may improve adherence to recommended screening, rates of use have varied greatly across different risk-reduction options. Computer-based interventions might be a useful educational adjunct for nephrology patients. However, examining whether and how these prior findings relate to the context of hereditary kidney disease is an important and rich area for future research.

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Table 1

Effects of genetic information following genetic counseling and testing on use of risk-reducing surgical procedures, screening and surveillance, and chemoprevention.

| Study                                | Sample  | Follow-up timepoints after results disclosure | Major results   |
|--------------------------------------|---|---|---|
| <b>HBOC</b>                          |   |   |   |
| Chen et al. (2002) <sup>44</sup>     | 198 unaffected women without prior prophylactic or preventive procedures                | After receipt of test results                 | <i>BRCA1/2</i> carriers had significantly higher intentions for chemoprevention, PM, and PO than non-carriers or those with unclassified variants ( $p < .0001$ )   |
| Graves et al. (2007) <sup>45</sup>   | 435 women affected with unilateral breast cancer  | 1, 6 and 12 months                            | Women with uninformative <i>BRCA1/2</i> test results were less likely to have received contralateral PM at 12-months than those with positive test results (OR=0.23; 95% CI:0.08–0.66)  |
| Kinney et al. (2006) <sup>46</sup>   | 40 women from an African American <i>BRCA1</i> kindred                                  | 12 month                                      | No significant increase in mammography screening observed, regardless of carrier status   |
| Lerman et al. (2000) <sup>47</sup>   | 216 female members of HBOC families   | 12 month                                      | Carriers had significantly higher rates of mammography (68%) than non-carriers (44%) (OR= 7.1; 95% CI:1.36–37.1)  |
| Lynch et al. (2006) <sup>36</sup>    | 459 women from HBOC families  | Varying times post disclosure                 | 53% of carriers and 0% of non-carriers underwent prophylactic surgeries after disclosure, significantly different from pre-disclosure ( $p < .0001$ )<br><br>Rates of TVU significantly increased in carriers ( $p < .015$ ) and marginally decreased in non-carriers   |
| Metcalfe et al. (2005) <sup>48</sup> | 125 unaffected <i>BRCA1/2</i> carriers  | Varying times post disclosure                 | 12% had used tamoxifen, 10% raloxifene<br><br>27% underwent PM, 67% PO  |
| Peshkin et al. (2002) <sup>49</sup>  | 107 unaffected women  | 12 month                                      | No significant difference in use of mammography at 12 months between carriers (59%) and non-carriers (47%)  |
| Phillips et al. (2006) <sup>50</sup> | 142 unaffected female <i>BRCA1/2</i> carriers   | 3 years                                       | Carriers who knew their mutation positive status were significantly more likely than those who did not know mutation positive status to undergo all screening practices except BSE (OR=12.1 for PO; 7.9 for CBE; 7.2 for mammogram; 30.5 for TVU)   |
| Scheuer et al. (2002) <sup>51</sup>  | 251 unaffected <i>BRCA1/2</i> carriers  | Varying times post disclosure                 | Increase in use of mammogram ( $p = .001$ ), CBE ( $p = .001$ ), TVU ( $p < .001$ ) and CA-125 ( $p < .001$ ) after disclosure among those without risk-reducing surgeries  |
| Schwartz et al. (2003) <sup>52</sup> | 289 high-risk women   | 1 year  | Mutation negative individuals (OR=0.03; 95% CI:0.01–0.30) and <i>BRCA2</i> carriers (OR=0.23; 95% CI:0.04–1.2) significantly less likely than <i>BRCA1</i> carriers to receive bilateral PO<br><br>Positive test result also associated with increased use of CA-125 ( $p < .001$ ) and TVU ( $p = .003$ ) ovarian cancer screening |
| Schwartz et al. (2004) <sup>53</sup> | 194 newly diagnosed breast cancer patients at risk for carrying <i>BRCA1/2</i> mutation | Surgical decision after testing               | Carriers significantly more likely to choose BLM (48%) than those with uninformative results (24%) or test decliners (4%) ( $p < .001$ )  |
| Weitzel et al. (2003) <sup>54</sup>  | 37 newly diagnosed breast cancer patients   | Surgical decision after testing               | Carriers significantly more likely to undergo BLM than unilateral mastectomy ( $p < .001$ )   |
| <b>HNPCC</b>                         |   |   |   |
| Collins et al. (2005) <sup>55</sup>  | 98 unaffected individuals   | 12 months                                     | Carrier status only significant predictor of colonoscopy at follow up (OR=20; 95% CI: 5.8–68)<br><br>Among women, mutation carriers reported greater use of TVU ( $p = .004$ ) and endometrial sampling ( $p < .001$ ) than non-carriers  |
| Hadley et al. (2004) <sup>56</sup>   | 56 unaffected individuals from families with known HNPCC mutation                       | 6 and 12 months                               | Colonoscopy use significantly higher among carriers than non-carriers (OR=61.6; 95% CI:5.8–652.6)   |

| Study                               | Sample  | Follow-up timepoints after results disclosure | Major results  |
|-------------------------------------|---|---|--|
| Hadley et al. (2008) <sup>29</sup>  | 65 unaffected women in families with HNPCC mutation | 6 and 12 months                               | Carriers more likely to undergo colonoscopy than non-carriers at 12 months (p<.0001)   |
| Halbert et al. (2004) <sup>57</sup> | 98 unaffected adults from HNPCC families            | 1, 6 and 12 months                            | Carriers were significantly more likely than test decliners to have a colonoscopy at 12 months (OR=12.12; 95% CI: 3.42–42.96); no significant change in colonoscopy use among non-carriers |
| Wagner et al. (2005) <sup>58</sup>  | 94 mutation carriers                                | Varying times post disclosure                 | Colonoscopy screening increased from 31% to 88% among carriers (p<.001); gynecological screening increased among females from 17% to 69% (p<.001)  |

BLM=bilateral mastectomy

ES=endometrial sampling

PM=prophylactic mastectomy

PO=prophylactic oophorectomy

TVU=transvaginal ultrasound

**Table 2**

Effects of different educational approaches for genetic information related to breast and ovarian cancer risk on cognitive, affective and behavioral intention outcomes.

| Study                                     | Experimental design  | Sample  | Major results   |
|---|--|---|---|
| Bowen et al. (2004) <sup>59</sup>         | Randomized. GC vs. psychosocial counseling vs. control group                                 | 354 women with breast cancer family history, not consistent with <i>BRCA</i> mutation                           | Participants in both counseling groups had lower perceived risk at follow up compared to controls (p<.01)<br><br>Cancer worry decreased in both counseling groups (p<.05)   |
| Calzone et al. (2005) <sup>60</sup>       | Randomized. Group vs. individual pre-test education session                                  | 142 patients at high risk for carrying <i>BRCA</i> mutation   | No significant differences in knowledge or distress between groups  |
| Green et al. (2004) <sup>61</sup>         | Randomized. Standard one-on-one GC vs. computer-based educational program and GC             | 211 women with personal or family histories of breast cancer  | Mean increase in knowledge significantly higher in computer group among low-risk women (p=.03) but not high-risk women<br><br>Reduction in perceived risk greatest among low-risk women in counselor group (p<.001)   |
| Lerman et al. (1997) <sup>62</sup>        | Randomized. Education vs. education and non-directive psychosocial counseling vs. control    | 400 women at low to moderate risk with family history of breast and/or ovarian cancer                           | Significant increases in knowledge in both intervention groups (p<.001)<br><br>Significant increases in perceived limitations and risks of testing among counseling group (p<.01)<br><br>No difference in testing intentions between groups                     |
| Lobb et al. (2002) <sup>63</sup>          | Randomized. Received or not an audiotape of pre testing GC session                           | 193 unaffected and affected women from high risk breast cancer families   | Risk perception less accurate in audiotape group among unaffected women (p=0.05)<br><br>Women in intervention group who listened to the audiotape had greater reductions in anxiety (p=.02) and depression (p=.01)  |
| Mancini et al. (2006) <sup>64</sup>       | Non-randomized. Standard patient information booklet vs. no booklet                          | 560 affected women considering <i>BRCA1/2</i> testing   | Booklet group had greater knowledge (p=.001) and satisfaction with information provided (p=.001)<br><br>Women in booklet group had stronger testing intentions (p=.009)   |
| Miller et al. (2005) <sup>65</sup>        | Randomized. Enhanced counseling vs. general health information session following standard GC | 77 high-risk women undergoing <i>BRCA1/2</i> testing  | At 6 months, information seeking about prophylactic oophorectomy higher in enhanced counseling group (p=.04)<br><br>At 6 months, 28% of those in enhanced counseling group had undergone prophylactic oophorectomy, compared to 6% of controls (p=.03)          |
| Skinner et al. (2002) <sup>66</sup>       | Randomized. Tailored vs. non-tailored print materials  | 325 women considering <i>BRCA1/2</i> testing with personal and family histories of breast and/or ovarian cancer | Those in tailored materials group had significantly greater improvement in knowledge (p<.0001) and less over-estimation of risk of being a mutation carrier (p<.0001)<br><br>No significant differences between groups in anxiety or expectations about testing |
| van Roosmalen et al. (2004) <sup>67</sup> | Randomized. Received decision aid or control group   | 368 affected and unaffected women being tested for a <i>BRCA</i> mutation                                       | Decision aid group more frequently considered prophylactic surgery at 4 weeks (p=.02)<br><br>Decision aid group had more accurate risk perceptions and felt better informed (p<.01)<br><br>No differences between groups for anxiety,                           |

| Study                                 | Experimental design   | Sample   | Major results  |
|---------------------------------------|---|--|--|
| Wakefield et al. (2008) <sup>68</sup> | Randomized. Decision aid or control pamphlet in pre testing GC session                      | 148 affected and unaffected women considering BRCA testing | depression, cancer distress<br>Decision aid group felt more informed (p=.003) and had higher knowledge levels (p<.05)  |
| Wang et al. (2005) <sup>69</sup>      | 2×2 factorial design. CD-ROM program for patients; feedback checklist for genetic counselor | 197 women attending BRCA counseling                        | Patients who viewed CD-ROM significantly less likely to undergo genetic testing (33% vs. 47%; OR=0.63; 95% CI: 0.45–0.89)<br><br>Feedback group had greater increases in knowledge (p<.05)<br><br>Among those less worried at baseline, the CD-ROM group had no increase in worry over time, others had increase in worry (p<.005) |
| Watson et al. (1998) <sup>70</sup>    | Randomized. Audiotape vs. standard consultation   | 115 women with family history of breast cancer             | Worry decreased in audiotape condition (p=.002)<br><br>Audiotape had no effect on recall of genetic risk estimate  |

GC=genetic counseling