J Genet Counsel (2017) 26:361–378
DOI 10.1007/s10897-017-0067-x

REVIEW PAPER

# A Rapid Systematic Review of Outcomes Studies in Genetic Counseling

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Received: 5 February 2016 / Accepted: 6 January 2017 / Published online: 6 February 2017


**Abstract** As healthcare reimbursement is increasingly tied to value-of-service, it is critical for the genetic counselor (GC) profession to demonstrate the value added by GCs through outcomes research. We conducted a rapid systematic literature review to identify outcomes of genetic counseling. Web of Science (including PubMed) and CINAHL databases were systematically searched to identify articles meeting the following criteria: 1) measures were assessed before and after genetic counseling (pre-post design) or comparisons were made between a GC group vs. a non-GC group (comparative cohort design); 2) genetic counseling outcomes could be assessed independently of genetic testing outcomes, and 3) genetic counseling was conducted by masters-level genetic counselors, or non-physician providers. Twenty-three papers met the inclusion criteria. The majority of studies were in the cancer genetic setting and the most commonly measured outcomes included knowledge, anxiety or distress, satisfaction, perceived risk, genetic testing (intentions or receipt), health behaviors, and decisional conflict. Results suggest that genetic counseling can lead to increased knowledge, perceived personal control, positive health behaviors, and improved risk perception accuracy as well as decreases in anxiety, cancer-related worry, and decisional conflict. However, further studies are needed to evaluate a wider array of outcomes in more diverse genetic counseling settings.

**Keywords** Genetic counseling · Outcomes · Review

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## Introduction

Genetic counselors are health care providers who have completed specialized graduate training to function as professionals in this discipline. While the term "genetic counseling" can be used somewhat generically to describe the *activity* of genetic counseling, regardless of the type of health care provider involved, we use the term genetic counseling in this paper to mean *an intervention delivered by individuals trained as genetic counselors* specifically.

The practice of genetic counseling has been defined as, "… the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease". The definition states that the process includes three components- risk assessment, education, and counseling (NSGCDTF 2006). Outcomes that are stated or can be implied from this definition include: improved patient knowledge through effective education and attention to the impact of genetic information; accurate identification and communication of risk on the part of the genetic counselor; and informed patient decision-making. The adaptation component of the definition alludes to multiple potential outcomes

including improving patient adherence to available medical management recommendations, enhancing quality of life, reducing morbidity and mortality, and promoting patient sharing of information with at-risk relatives. In addition, genetic testing is increasingly a component of risk assessment. Appropriate genetic testing, which encompasses identifying the right genetic test(s) and offering it to appropriate patients, might also be a potential genetic counseling outcome.

Rising health care costs in the absence of concomitant improvements in the public's health are driving health care professionals, including genetic counselors, to identify and implement evidence-based strategies that improve patient outcomes. Prior work to generate a comprehensive list of genetic counseling outcomes includes work by The Western States Genetics Services Collaborative. This group developed an outcomes menu for public health and clinical genetics services based on a review of the literature, two existing documents describing genetics outcomes, and an iterative review process (Silvey et al. 2009). The group identified 12 major outcomes with 42 sub-outcomes falling into five impact areas: knowledge and information; financing; screening and identification; diagnosis, treatment and management; and population health. Outcomes specific to genetic counseling included making informed health and life decisions based on a genetic diagnosis, participating in treatment "at optimal levels" after genetic counseling, and feeling supported in managing their emotional reactions to the genetic information (Silvey et al. 2009).

Establishing a set of meaningful genetic counseling outcomes, although not without its challenges, is critical to supporting evidence-based practice in genetic counseling. The goals of this rapid systematic literature review are 1) to catalogue and summarize the outcomes that have been previously measured in the setting of genetic counseling provided by genetic counselors; 2) to identify potential gaps in the literature; 3) to discuss some of the challenges in outcomes research; and 4) make recommendations about future outcomes research in genetic counseling.

## Methods

We conducted a literature review using a "systematic rapid-review" approach (Ganann et al. 2010). The Web of Science (including Pub Med) and CINAHL databases were searched using the terms "genetic* counsel*" and "genetic consul*" to capture all papers with a title, abstract, or topic that included the phrase "genetic counselor", "genetic counseling", "genetic consult" and variant spellings. Searches were performed on July 18, 2013, no date limits were applied. Searches were refined to exclude case reports and non-peer reviewed journal articles (such as commentaries), non-English papers, and animal studies. Our primary goal was to simply catalogue the outcomes that had been used in previous research, not to apply any quality metrics or meta- analyses as might be done in a more traditional systematic review of an intervention.

The inclusion criteria were:

1) Genetic counseling was provided by non-physician genetics specialists. (Masters-level genetic counselor, advanced practice genetics nurse, or other graduate-level genetics specialist trained in accordance with the standards and accreditations appropriate to that country). In the case of multiple types of providers performing genetic counseling within a study, the study was included if the majority of providers were non-physician genetic counselors. We excluded studies that measured the outcomes of genetic *testing*. Receiving a genetic test result as part of a genetic counseling session is an event that may profoundly alter medical management or reproductive decision-making in and of itself. As such, genetic testing is likely to have outcomes that are distinct from genetic counseling outcomes.
2) Measures occurred before and after genetic counseling (pre-post design) OR measures were compared between a genetic counseling arm vs some other intervention or a control arm that did not include genetic counseling (comparative cohort design).

One author reviewed the initial list of references and papers that clearly did not meet eligibility criteria were excluded. The remaining abstracts were each reviewed by two authors independently to determine inclusion. When the two authors did not agree on eligibility, discussion between the two authors resulted in resolution in all cases. In cases where eligibility could not be determined from the publication (e.g., in cases where the paper did not indicate who provided the genetic counseling), the corresponding author of the respective paper was contacted for clarification. Once the list of eligible studies was finalized, key data elements were extracted from the published papers and summarized. Variables extracted included sample size, clinical setting, geographic location, study design, patient/family characteristics, outcome measures reported, and results.

## Results

The Web of Science/PubMed search identified a total of 13,329 papers; CINAHL identified 1038. An initial review of all abstracts resulted in retention of 1063 references for further review. A total of 23 papers met our inclusion criteria. The clinical setting, geographic location, study design, and outcomes measured are reported in Table 1.

**Table 1** Characteristics of studies that reported outcomes of genetic counseling ($n = 23$)

| | N | % |
|---|---|---|
| Setting | | |
| Cancer | 13 | 57 |
| General | 6 | 26 |
| Prenatal | 2 | 9 |
| Pediatric | 1 | 4 |
| Psychiatric | 1 | 4 |
| Country | | |
| United States | 16 | 70 |
| Canada | 2 | 9 |
| Australia | 2 | 9 |
| Israel | 1 | 4 |
| Netherlands | 1 | 4 |
| Spain | 1 | 4 |
| Study design and comparison | | |
| Observational, comparative, GC vs. no GC (Table 2) | 5 | |
| Randomized, comparative, GC vs. no GC (Table 2) | 6 | |
| Observational, single-arm with pre-post measures (Table 3) | 12 | |
| Outcomes assessed[a] | | |
| Knowledge | 13 | 57 |
| Anxiety, Depression, Distress, Concern | 10 | 43 |
| Perceived Risk | 7 | 30 |
| Satisfaction or Perceived Usefulness | 5 | 22 |
| Genetic Testing Intention or Receipt | 5 | 22 |
| Health Behavior or Health Outcome | 5 | 22 |
| Decisional Conflict | 3 | 13 |
| Family Outcomes | 2 | 9 |
| Perceived Personal Control (PPC) | 2 | 9 |
| Quality of Life (QOL) | 1 | 4 |
| Self-esteem | 1 | 4 |

[a] Total >100% since some studies measured >1 outcome

Eleven studies compared patients receiving genetic counseling to a control or comparison group (Table 2), and twelve studies used a pre- post- design (Table 3). There were six randomized trials that directly examined genetic counseling versus some other intervention. Most studies were conducted in the United States with Masters-level genetic counselors providing the genetic counseling.

Table 4 summarizes the measures used and the results of the included studies, organized by outcome domains. The most frequently measured outcomes were knowledge, satisfaction, anxiety and distress (including disease-specific distress and general distress), perceived risk, genetic testing (intent or receipt), decisional conflict, and health behaviors (including adherence). Measures used were a mix of previously validated measures and study-specific measures.

## Knowledge

The outcome studied most extensively in this review is knowledge. This is not unexpected given that education is one of the primary components of the genetic counseling process, and knowledge can be measured in a variety of settings. The majority of studies that used a pre-post design showed an increase in knowledge across several different practice settings. However, the majority of studies were in the hereditary breast cancer setting. All studies measured knowledge within 6 months of genetic counseling. The few studies that compared genetic counseling to a different educational intervention (e.g., pamphlet, computer-based modules) found that genetic counseling and the other interventions all increased knowledge.

## Anxiety, Depression, Distress

Five studies measured anxiety. Four of these studies used the State Trait Anxiety Inventory (STAI) and all studies showed either decreases or no change in anxiety levels. No studies found an increase in anxiety. Four of the five studies were conducted in the hereditary breast cancer setting.

Several studies evaluated depression, concern, negative affect, and/or distress. Notably, several studies in the cancer setting specifically used the Impact of Events Scale, where genetic counseling was the "event". The majority of studies demonstrated no change in distress, with a few showing reduced distress.

## Perceived Risk

All but one study of perceived risk were in the breast cancer setting where a variety of study-specific measures were used. Most studies showed a decrease in perceived risk. This result was interpreted as a beneficial outcome as many study participants were overestimating their cancer risk at baseline.

## Satisfaction/Perceived Usefulness

Five studies examined satisfaction with the genetic counseling process, or satisfaction with decision-making. A study by Hunter et al. (2005) is notable as it was a randomized trial of various genetic counseling approaches in the prenatal setting. The study found that women having individual genetic counseling were more satisfied than women receiving group genetic counseling or who were given a decision aid.

## Genetic Testing Intent or Receipt

All except one of the five studies looking at intention to undergo genetic testing or receipt of genetic testing were in the

**Table 2** Comparative studies- randomized or observational

| Study | Setting (Location) | Sample size | Genetic Counselor | Study population | Comparison groups | Outcomes | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | Knowledge | Anxiety, Depression, or Distress | Perceived Risk | Satisfaction or Perceived Usefulness | Genetic testing[a] | Health Behavior or Health Outcome | Decisional Conflict |
| Bowen et al. 2002 (Randomized) | Cancer (USA) | 357 | MS- GC[b] | Female relatives of breast cancer patients | GC vs group session with health counselor vs control (no GC or group) | | | | | X | | |
| Burke et al. 2000 (Randomized) | Cancer (USA) | 243 | MS- GC | Female relatives of breast cancer patients | GC vs control group (no GC) | | | X | X | | | |
| Ciske et al. 2001 | General genetics (USA) | 138 | MS- GC | Parents of children with CF[c] | GC vs no GC (per self-report) | X | | | | X | | |
| Cavanagh et al. 2010[d] | General genetics (USA) | 37 | MS- GC | Parents of children with CF[c] | GC *vs* no GC (per self-report) | X | | | | | | |
| Cheuvront et al. 1998 (Randomized) | General genetics (USA) | 288 | MS- GC | Relatives of CF patients | GC *vs* pamphlet | X | X | | | | | |
| Green et al. 2001 (Randomized) | Cancer (USA) | 72 | MS- GC | Female relatives of breast cancer patients | GC *vs* computer program vs control (knowledge assessment before GC) | X | | | | X | | |
| Green et al. 2004 (Randomized) | Cancer (USA) | 211 | MS- GC | Female relatives of breast cancer patients | GC *vs* computer program | X | X | X | X | X | | X |
| Halbert et al. 2012 | Cancer (USA) | 135 | MS- GC | African-American women | GC or Culturally tailored GC *vs* no GC (decliners) | | | | X | | | |
| Hunter et al. 2005 (Randomized) | Prenatal (Canada) | 352 | MS- GC | Pregnant wome n > age 35 & partners | GC *vs* group counseling GC *vs* decision aid +/−GC | X | X | | X | | | X |
| Randall et al. 2001 | Cancer (Australia) | 60 | MS- GC Australian graduate diploma in genetic counseling | Breast cancer patients | GC vs no GC | X | X | | | X | | |
| Rutherford et al. 2014 | Pediatrics (USA) | 198 | MS- GC | Pediatric genetics clinic patients | GC + MD vs MD only | | | | | | X | |

[a] Intention, attitude, receipt, desire, interest, or completion of Genetic Testing

[b] MS- GC (Masters-level genetic counselor)

[c] CF (cystic fibrosis)

[d] Follow-up to Ciske et al. 2001

**Table 3** Observational, single-arm studies with pre-post design

| Study | Setting (Location) | Sample size | Genetic Counselor | Study population | Timing of Measures | Outcomes | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | Know-ledge | Anxiety, Distress, Concern, or Depression | Perceiv-ed risk | Satisfaction or Perceived Usefulness[a] | Health Behavior or Health Outcome[b] | Decision-al conflict | Family outcomes[c] | PPC[d] | QOL[e] | Self-esteem |
| Austin and Honer 2008 | Psychiatric (Canada) | 13 | MS-GC[f] | Parents of children with psychiatric disorder | Baseline, Immediat-ely post-GC, 1-month post-GC | | X | | X | | | | | | |
| Baldwin et al. 2012 | General genetics (USA) | 244 | MS-GC | Deaf/hard of hearing adults | Baseline, immedi-ately post-GC | X | | | | | | | | | |
| Berkenstadt et al. 1999 | Prenatal (Israel) | 256 | MS-GC, few with MD geneticist | Patients referred for age-related prenatal diagnosis with a specific genetic problem. | Baseline, immedi-ately post-GC | | | | | | | | X | | |
| Cabrera et al. 2010 | Cancer (Spain) | 152 | Advanced practice genetics nurse | Relatives of breast cancer patients | Baseline, 1-month post-GC, 6-months post-GC | X | X | X | | | | | | X | |
| Christie et al. 2012 | Cancer (USA) | 93 | MS-GC | Breast cancer patients | Baseline, 2–-3 weeks post GC | X | X | | | | X | | | | |
| Grant et al. 2013 | General genetics (USA) | 108 | MS-GC | Overweight adults in primary care clinic | Baseline, approx. 12 weeks post-GC | | | X | | X | | | | | |
| MacDonald et al. 2007 | Cancer (USA) | 122 | Advanced practice genetics nurse; MS-GC | Women with personal/family history of breast/ovarian cancer | Baseline, 6-months post-GC | | | | | | | X | | | |
| McInerney-Leo et al. 2004, 2005, 2006 (Parts I, II and III of the same study) | Cancer (USA) | 212 | MS-GC | Men and women from 13 extended families with a BRCA mutation (This review includes only participants declining genetic testing) | Baseline, 6–-9 months post- GC | | X | X | | X | | X | | | X |

**Table 3** (continued)

| Study | Setting (Location) | Sample size | Genetic Counselor | Study population | Timing of Measures | Outcomes | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | Know-ledge | Anxiety, Distress, Concern, or Depression | Perceiv-ed risk | Satisfaction or Perceived Usefulness[a] | Health Behavior or Health Outcome[b] | Decision-al conflict | Family outcomes[c] | PPC[d] | QOL[e] | Self-esteem |
| Meiser et al. 2001 | Cancer (Australia) | 218 | MS-GC; Australian graduate diploma in genetic counseling, few MD geneticists | Women with family history of breast/ovarian cancer | Baseline, 12--months post-GC | X | X | X | | X | | | | | |
| Pal et al. 2010 | Cancer (USA) | 37 | MS-GC | African-American women with breast cancer before age 50 | Baseline, 6–-185 days post-GC | X | | | | | | | | | |
| Pieterse et al. 2011 | Cancer (Netherla-nds) | 77 | GCs as defined in the Netherlands; advanced practice genetic nurses, PhDs, and MS-level | Women presenting for BRCA genetic counseling | Baseline, immedi-ately post-GC, 6 months post-GC | X | X | X | | | | | X | | |
| Taylor and Wu 2009 | General genetics (USA) | 98 | Advanced practice genetics nurse | African-American women recruited for genetic study of hypertension | Baseline, 6 months post-GC | | | | | X | | | | | |

[a] Satisfaction with genetic counseling and/or genetic testing or perceived usefulness of genetic counseling

[b] Health behavior constructs (e.g. cancer screening in Meiser et al. 2001; BMI in Taylor and Wu 2009)

[c] Communication with family members and family relationships

[d] Perceived personal control

[e] Quality of Life

[f] Masters-level trained genetic counselors (MS-GC)

breast cancer genetics setting; in some cases the goal was to decrease inappropriate testing intentions in low-risk women.

### Health Behaviors Including Adherence

Five studies measured aspects of health behavior. One study, conducted in the diabetes setting, measured weight loss and attendance at a 12-week lifestyle balance course. Another study, conducted in the cancer setting, measured breast cancer screening practices. A third study investigated lifestyle and health changes (weight, blood pressure, sodium intake and physical activity) in a hypertensive population. The fourth investigated adherence to medical management recommendations (various) in a pediatric genetics patient population. The fifth investigated adherence to medical recommendations made in a pediatric genetics clinic.

### Decisional Conflict

Three studies measured decisional conflict; all used the Decisional Conflict scale and all showed decreases in decisional conflict. Two studies were in the cancer setting and one was in the prenatal setting.

### Other Outcomes

The review identified several outcomes that were measured in only one or two studies, including: self-esteem, quality of life, family outcomes (communication and relationships), and perceived personal control.

## Discussion

Although our findings are based on the relatively small number of studies meeting our inclusion criteria, the studies to date show that a wide variety of outcomes can be considered in genetic counseling research. Results of these studies demonstrate that genetic counseling can lead to increases in knowledge, perceived personal control, positive health behaviors, and increased accuracy of perceived risk. Anxiety, cancer-related worry, and decisional conflict often decrease following genetic counseling, and patient satisfaction is typically high. The studies we identified used a variety of different measures which limits the ability to make specific cross study comparisons. Furthermore, the majority of studies we reviewed occurred within the oncology/cancer risk setting which limits applicability to the other practice settings such as reproductive or pediatric genetics. The focus on outcomes in the cancer setting may be due to the availability of evidence-based guidelines for cancer genetic risk assessment and widespread interest and availability of genetic counseling and testing for familial breast and ovarian cancer risk.

An important strength of the current review stems from the broad search terms and methods we used to increase our ability to capture relevant papers meeting our inclusion criteria. Despite our efforts, additional studies meeting our criteria may not have been captured. Furthermore, this review must be interpreted in light of our strict criteria which deliberately focused on genetic counseling as provided by predominantly masters-level genetic counselors. We identified some additional studies that included advanced practice genetics nurses, and other types of genetic counselors as defined by their jurisdiction at the time of the study (for example, the Australian graduate diploma in genetic counseling). A recent review by McAllister and Dearing (2015) used a broader definition of "genetic services" which included genetic counseling by physicians, non-physicians, and genetic testing. In contrast, our review excluded studies that combined the outcomes of genetic counseling and testing, as our focus was to look at what outcomes had been measured in the context of the genetic counseling process, rather than as the result of a genetic test. Finally, several cross-sectional studies that showed patient satisfaction with genetic counseling or other post-genetic counseling only measures were also excluded from this review as there was no baseline comparison measure. It should also be noted that we identified two additional studies that were published following our cut-off date for the review that meet our inclusion criteria; Hippman et al. (2016) measured knowledge, risk perception, internalized stigma, and perceived control over illness in a pilot randomized trial of genetic counseling vs. an educational booklet; results indicated that genetic counseling improved risk perception and both interventions improved knowledge. Palmer et al. (2014) found that understanding of genetic test results improved after genetic counseling, and deaf identity remained stable in a sample of individuals undergoing genetic testing for mutations in genes related to deafness.

An important limitation of this study is that we did not attempt to evaluate the quality of the individual studies or identify biases that might be present in the study designs. Although randomized controlled trials are considered the gold standard, the quality can vary widely. We also chose to include pre-post study designs, even though they are typically less scientifically rigorous, because we were primarily interested in capturing a broad spectrum of the types of outcomes that have been studied. Future work is needed to incorporate scales and checklists (such as CONSORT) that have been developed to rate the quality of studies.

Despite study limitations, critical gaps that this review highlights include the relatively small number of genetic counseling outcomes studied to date and lack of studies that focus on morbidity, mortality or other long-term health outcomes. However, several outcomes may have direct or indirect influences on morbidity and mortality. For instance, genetic counseling may influence self-efficacy to follow

treatment or surveillance recommendations as well as adherence to these recommendations in cases where such recommendations have been shown or are expected to result in decreased morbidity and mortality. Self-efficacy was not an outcome evaluated in any of the studies we identified. However, after the date of our literature search, a randomized controlled trial of individuals with a first degree relative of colon cancer was published in which genetic counselors delivered a motivational interviewing risk communication intervention designed to increase perceptions of colorectal cancer risk and disease severity as well as self-efficacy and response-efficacy. That study found that rates of colonoscopy were substantially higher in the intervention group (35.4%) as compared to the comparison group who received mailed informational materials only (15.7%) (Kinney et al. 2014). Nevertheless, unlike colorectal cancer, not all genetic or familial conditions have clear treatment or prevention guidelines that reduce morbidity and mortality and therefore the effect of genetic counseling on other measures such as self-reported quality of life may be areas to explore. Likewise, in prenatal genetic counseling, where the primary goal is autonomous, informed decision making, measures such as decisional conflict and perceived personal control may be more appropriate.

Additional focus on patient-reported genetic counseling outcomes is critical as healthcare shifts to a more patient-centered focus that emphasizes value and outcomes. All potential outcomes of interest, particularly those that have not been evaluated or that were included in only a single study in our review, will require additional verification in a wider variety of settings. Identification or development of standardized measures would also be useful for assisting in the ability to make comparisons across settings and across studies. Some outcomes scales specific to the genetic counseling setting have already been developed (McAllister et al. 2011), but our search did not identify any studies meeting our inclusion criteria that used this instrument within our search dates. A Satisfaction with Genetic Counseling scale has also been published (Shiloh et al. 1990) and was used in one study in our review. Several general measures that have been validated in multiple populations were used repeatedly (e.g. STAI, Impact of Events), but these were primarily in the cancer setting and were focused on cancer-related distress or distress related to cancer genetic counseling.

At the present time, there is a limited, but growing body of literature on genetic counseling outcomes to guide evidence-based practice. However, there are a number of challenges to measuring these outcomes. One challenge is that there are a variety of health care professionals who provide genetic counseling services. Some, such as Master's trained genetic counselors, advanced practice genetics nurses, physician medical geneticists, and PhD medical geneticists have specialized training and certification in genetics whereas others may have no or minimal training in genetics. These differences may translate into variations in how genetic counseling is provided by genetics professionals versus other health professionals who offer genetic services, which limits the validity of comparing genetic counseling outcomes across professions and also highlights the need to identify which genetic counseling processes or strategies contribute most to patient outcomes.

Even amongst genetic specialists, clinical training, practice-based competencies and scopes of practice vary (ACGC 2013; ACMG 2011). For instance, in the United States and Canada, genetic counselor training is focused on four competency domains- genetics expertise and analysis, interpersonal communication, psychosocial and counseling skills, education, and professional development and practice (ACGC 2013). Competencies for the physician medical geneticists overlap with many aspects of the genetic counselor competencies in areas such as genetics knowledge, family history taking, risk assessment and genetic testing (ACMG 2011). However, the geneticist competencies include the physical examination and treatment components that are not part of the genetic counselor scope of practice. In contrast, the genetic counselor competencies have greater emphasis on the patient education and counseling skills that are key components of genetic counselor's role (ACGC 2013). As such, the outcomes of a genetic counseling session performed solely by a medical geneticist may or may not be similar to those of sessions conducted by a genetic counselor.

Another factor that makes it difficult to measure genetic counseling outcomes is the diverse practice settings in which genetic counselors work. The 2014 National Society of Genetic Counselors' Professional Status Survey revealed that 35% of clinical genetic counselors work in prenatal genetics, 12% in pediatric genetics, 29% in cancer genetics and 24% in other specialties, the most common of which are research, general genetics, cardiology, specialty disease, laboratory, infertility/IVF, metabolic disease, and neurogenetics (NSGC 2014). There are some desired outcomes that are common across practice settings such as patient satisfaction, accurate risk assessment, informed decision making and adaptation to genetic disease or risk. However even these potential "core" outcomes are conceptualized heterogeneously in different genetics settings. Some outcomes, such as reducing morbidity and mortality through screening, risk reduction and preventative measures, may be most relevant to services such as cancer or cardiovascular genetics. Measuring outcomes is further complicated by the fact that genetic counseling is strongly rooted in promoting patient/family autonomy (NSGC 2006), especially in the prenatal and infertility/assisted reproductive technology settings. As such, outcome measures like disease prevention are not applicable, while measures of decisional conflict or distress may be more suitable for such settings.

A further complicating factor in measuring genetic counseling outcomes is the tight link between genetic

**Table 4** Results of studies by outcome type

| Study | Study population | Measures | Main results |
|---|---|---|---|
| Knowledge | | | |
| Baldwin et al. 2012 | Deaf or hard of hearing individuals | 10 true/false knowledge questions on genetics given at baseline and pre- and post genetic counseling | Statistically significant increase in knowledge following pre-test genetic counseling (paired t(239) = 3.45, $p = .0007$). |
| Cabrera et al. 2010 | Patients without a cancer diagnosis presenting for BRCA genetic counseling/with a family history of breast cancer | 13 question knowledge questionnaire developed by the study investigators to evaluate prevention, diagnosis, and treatment of breast cancer, risk of breast cancer and HBOC risk. Score 0–31 with higher = more knowledge. Administered at baseline, immediately post genetic counseling (P1) and 6 months later (P2) | A significant increase in knowledge was detected from baseline (mean knowledge score of 16.37, S.D. 4.1) to Post 1 (mean score of 19.6, S.D. 4.3, $p < 0.001$) and from baseline to Post 2 (mean score of 19.6, S.D. 4.2, $p = 0.005$. Changes in knowledge were less significant for participants who were older, lower levels of education, and having 4 or more children. |
| Cavanagh et al. 2010 | Parents of children with CF | 18-item knowledge questionnaire (multiple choice and yes/no/unsure) regarding the genetics of CF and their child's sweat test results | Parents who received genetic counseling had significantly higher knowledge scores than those who did not receive genetic counseling ($r = -0.53$, 95% CI = −0.73 to −0.24. |
| Cheuvront et al. 1998 | 1st, 2nd, 3rd degree relatives of people with CF | 10-item true/false knowledge of CF disease, basic genetics, and implications of carrier status | No significant difference in knowledge with GC versus pamphlet. |
| Christie et al. 2012 | Breast cancer patients meeting NCCN cancer genetics referral criteria presenting either before definitive surgery (BDS) or after (ADS) | 15-item adapted version of National Center for Human Genome Research Knowledge Scale, to measure HBOC knowledge (mutation prevalence, inheritance, cancer risks, management) | Significant increase in knowledge between T1 (prior to pre-test genetic counseling) and T2 (after counseling) for both BDS and ADS patients; median change 4.2 ($p = 0.004$) and 2.7 ($p < 0.001$) respectively. |
| Ciske et al. 2001 | Parents of children with CF | 7 item questionnaire (true/false/unsure) | Statistically significant differences noted in five of seven knowledge questions when comparing frequency of correct responses between parents who received genetic counseling and parents who had not received genetic counseling. Correct responses ranged from a mean of 94.2 vs. 66.1, $p < 0.001$, respectively, to 93.1 vs. 71.9 $p < 0.004$, respectively. Frequency of accurate responses did not depend on which health care professional provide the genetic counseling. |
| Green et al. 2004 | Women with personal or family history of breast cancer | NHGRI 20-item multiple choice and true/false questionnaire | Knowledge scores increased in GC and computer group ($p < 0.001$) but was higher in computer group (change score = 38) compared to counselor group (change score = 29, $P = .03$). |
| Green et al. 2001 | English speaking women 18y + with a first degree relative with breast cancer | NHGRI 20-item multiple choice and true/false questionnaire | The mean percent of correct knowledge responses was significantly greater for participants seen by a GC (92%) or the interactive computer program group (96%) compared to the control group (74%), $P < .0001$. After adjusting for demographics there were no significant differences between the GC and interactive computer program groups. |
| Hunter et al. 2005 | AMA prenatal patients; gestation < =18 weeks; no previous consideration of prenatal diagnosis; English speaking | Maternal Serum Screening Knowledge Questionnaire −19 item self-report measure to examine knowledge of prenatal testing and alternatives | Women and men in the group and individual GC sessions, and those receiving a decision aid, all showed |

**Table 4** (continued)

| Study | Study population | Measures | Main results |
|---|---|---|---|
| | | | increases in knowledge from pre-to post GC ($p < 0.016$). |
| Meiser et al. 2001 | Women at risk of developing hereditary breast cancer | Breast cancer knowledge scale: 9 item true/false measure (revised from Lerman 1996) | Knowledge increased at follow up ($Z = -7.73$; $P < 0.001$). |
| Pal et al. 2010 | African American women undergoing GC for early onset breast cancer | 12 item instrument that incorporated elements of informed consent as outlined by ASCO (true/false/don't know) | Out of a maximum score of 12, the mean pre- and post-GC knowledge scores for all participants were 6.2 and 7.8, respectively, with a statistically significant increase in knowledge ($p < 0.0001$). |
| Pieterse et al. 2011 | Women presenting for BRCA genetic counseling | 7 questions (correct, incorrect, don't know), covered probabilities of carrying a BRCA ½ mutation, developing breast cancer conditional on carrier status, and necessity of surveillance | Mean knowledge scores decreased slightly after GC and at 6-month follow-up. The only statistically significant decrease was among women without a cancer history (pre-GC mean = 4.83; mean at 6-months = 4.36, $p < 0.05$, $\chi^2 = 3.84$) (6.7% decrease). |
| Randall et al. 2001 | Women with breast cancer presenting for cancer genetic counseling versus women with breast cancer but not seeking genetic counseling (control) | Nine-item true/false knowledge scale adapted from Lerman et al. | Women presenting for cancer GC had significantly greater increase in knowledge than controls ($T = 2.7$, $P < 0.05$). |
| Anxiety, depression, distress, concern | | | |
| Austin and Honer 2008 | Unaffected parents of children with a psychotic disorder | Assessed impact of genetic counseling on perceived understanding of mental illness, concern about risk to other relatives, perceived usefulness of psychiatric genetic counseling | Over 84% of participants indicated they were concerned to some degree about other relatives' risk for psychiatric disease; all indicated that genetic counseling decreased their concerns to some extent. |
| Cabrera et al. 2010 | Patients without a cancer diagnosis presenting for BRCA genetic counseling/with a family history of breast cancer | Hospital Anxiety and Depression Scale (HADS)<br>Spanish version of Cancer Worry Scale - 6-item scale with total scores ranging from 6 to 24 where a higher score indicates higher levels of cancer worries | No significant difference after counseling was noted. The mean score of 11.26, S.D. 6.91 at baseline, rose to 11.71, S.D. 7.99 at P1 ($p < 0.009$). At P2, mean score raised again to 12.33, S.D. 7.96 but this was not significantly different than baseline ($p = 0.411$).<br>Mean cancer worry significantly decreased in all risk groups (high, moderate, low risk) post counseling. The group baseline CWS was 11.42, S.D. 3.16, decreasing to 10.79, S.D. 3.32 ($p < 0.001$) at P1, and to 10.74, S.D. 3.42 ($p < 0.001$ Baseline to P2). |
| Cheuvront et al. 1998 | 1st, 2nd, 3rd degree relatives of people with CF | Positive and Negative Affect Scale (PANAS) | No significant difference in the positive versus negative affect in pamphlet versus GC. |
| Christie et al. 2012 | Breast cancer patients meeting NCCN cancer genetics referral criteria presenting either before definitive surgery (BDS) or after (ADS) | Impact of Event Scale (IES) | Significant decrease in overall cancer related distress ($p = 0.041$) and intrusive thoughts ($p = 0.014$) from pre- to post-genetic counseling sessions BDS patients but not ADS patients. |
| Green et al. 2004 | Women with personal or family history of breast cancer | State & Trait Anxiety Inventory (STAI) | Scores for the counselor group decreased significantly after counseling among high-risk ($p = .001$) and low-risk ($p = .007$) participants. For computer group participants, anxiety did not change significantly after computer use but did decline after subsequent |

**Table 4** (continued)

| Study | Study population | Measures | Main results |
|---|---|---|---|
| | | | counseling among both high-risk and low-risk women. |
| Hunter et al. 2005 | AMA prenatal pts; gestation < =18 weeks; no previous consideration of prenatal diagnosis; English speaking | State & Trait Anxiety Inventory (STAI) | No significant change before or after or by intervention (group, individual). |
| McInerney-Leo et al. 2004 | Women and men from 13 extended HBOC families with previously identified mutation and who completed baseline and follow-up questionnaire and who declined genetic testing | Center for Epidemiologic Studies Depressive Scale (CESD)<br>Impact of Event Scale (IES)<br>Breast Cancer Worries Scale (BCW) | No significant change from baseline to 6–9 months follow up in CESD, IES, or BCW scales. |
| Meiser et al. 2001 | Women at risk of developing hereditary breast cancer | Breast cancer knowledge scale : 9 item T/F measure (revised from Lerman 1996) | Knowledge increased at follow up ($Z = -7.73$; $P < 0.001$). |
| Pieterse et al. 2011 | Women presenting for BRCA genetic counseling | State & Trait Anxiety Inventory (STAI) - Form Y<br>Impact of Event Scale (IES) | Generalized anxiety decreased immediately after GC and continued to decrease over time. At 6-month follow-up anxiety was significantly lower than baseline ($p < 0.01$, $\chi^2 = 6.64$).<br>Distress related to seeking genetic counseling for hereditary cancer showed a statistically significant increase immediately following GC for women with cancer ($p < 0.01$, $\chi^2 = 6.64$). At 6-mo, unaffected women showed a decrease in distress ($p < 0.05$, $\chi^2 = 3.84$). |
| Randall et al. 2001 | Women with breast cancer presenting for cancer genetic counseling v controls (women with breast cancer but not seeking genetic counseling) | Beck Depression Inventory (BDI) – 21 item designed to measure severity of depression<br>State-Trait Anxiety Inventory (STAI)<br>Impact of Event Scale (IES) | No increase in depression following GC.<br>No significant difference in anxiety or distress after GC or over time. |
| Perceived risk | | | |
| Burke et al. 2000 | Women with at least one relative with breast cancer (fam hx suggestive of BRCA excluded) | Mean perceived personal risk on a scale from 0 to 100%, mean perceived risk of the average women's lifetime risk | GC group had a change in mean perceived risk decreased compared to controls: GC group: 49% at baseline to 24% at follow-up; control group 53% at baseline to 49% at follow-up ($F = 27.9$; df = 1235; $P < 0.001$). |
| Cabrera et al. 2010 | Patients without a cancer diagnosis presenting for BRCA genetic counseling/with a family history of breast cancer | Risk perception – study specific item: "I believe I will develop breast cancer at some time in my life" with 3 response options (I completely disagree or disagree; I am not sure or I agree or totally agree).<br>Objective Risk estimation: completed according to the Tyrer-Cuzick Model. Subjective risk estimation compared to actual risk to identify those who over- or under-estimate risk. The study compared risk perception with objective risk estimation. | No improvement in accuracy of risk perception for those who had overestimated or underestimated risk; also no significant change in risk perception over time. |
| Grant et al. 2013 | Overweight patients at increased phenotypic risk for type 2 diabetes | Recall of diabetes genetic risk status (e.g., "higher" or "lower") and numeric risk | Small favorable changes in risk perception noted, but not statistically significant. |
| Green et al. 2004 | Women with personal or family history of breast cancer | Perceived Relative Risk<br>a. In your opinion, compared to other women your age, what are your chances of developing breast cancer in the future<br>b. Responses 1 (much lower) to 5 (much higher) | All measures of perceived risk decreased by a greater amount in the genetic counseling group than in the computer intervention (the difference in relative risk decrease was not significant; the difference in absolute risk decrease was |

**Table 4** (continued)

| Study | Study population | Measures | Main results |
|---|---|---|---|
| | | Perceived Absolute Risk<br>a. What do you think your chances of getting breast cancer are on a scale of 0 to 100, where 0 is no chance of getting breast cancer and 100 means that you will definitely get it<br>Perceived risk of having a genetic susceptibility to breast cancer<br>a. In your opinion, how likely is it that you have an inherited gene mutation for breast cancer susceptibility? | significant at $p = 0.02$; the difference in perceived genetic risk was significant at $p = 0.002$). |
| McInerney-Leo et al. 2005 | Eighteen women from 13 extended HBOC families with previously identified mutation who completed baseline and follow-up questionnaire and who declined genetic testing | Likert scales measuring risk of breast cancer, risk of ovarian cancer, and risk of carrying a mutation ("In your opinion, compared to other women in your age, what are your chances of …") | No change in perceived breast cancer risk, but significant reductions in ovarian cancer risk and perceived chances of carrying a mutation ($P = 0.01$ for both). |
| Meiser et al. 2001 | Women at risk of developing hereditary breast cancer | One item asked participants to select their approximate perceived lifetime breast cancer risk from the following response options: 1,4,8, 12, 16, 25, 33, 50, 85, and 100%. | No significant changes in risk perception accuracy from baseline to follow-up. |
| Pieterse et al. 2011 | Women presenting for BRCA genetic counseling | Counselee's and counselors perceptions of lifetime risk of developing or redeveloping breast cancer with endpoints labeled 0–100%. They dichotomized into close estimation and overestimation categories. | Among those without cancer, the percentage who overestimated their risk of developing cancer pre-GC (96%) fell immediately after GC (50%) ($p < 0.001$, $\chi^2 = 10.83$) and at 6-month follow-up (57%) ($p < 0.01$, $\chi^2 = 6.64$). However, the percentage of those with cancer who overestimated risk of redeveloping cancer pre-GC (76%) decreased only slightly after GC (67%) and went back up at 6-months (77%) ($p > 0.05$). |
| Satisfaction or perceived usefulness | | | |
| Austin and Honer 2008 | Unaffected parents of children with a psychotic disorder | Questionnaire designed for study asked about perceived usefulness of psychiatric genetic counseling | All participants indicated the reason they chose genetic counseling was to increase knowledge. Immediately after the session, 12/13 (92%) indicated it was quite or very useful; 1 month after the session, all who responded to the follow up questionnaire (9/9) still found the information helpful. |
| Burke et al. 2000 | Women with at least one relative with breast cancer (family history suggestive of BRCA excluded) | Usefulness of genetic counseling on a scale of 1–5 | Of the 117 participants, 91 found it either very or moderately useful. Three found it somewhat useful and two found it not very useful. |
| Green et al. 2004 | Women with personal or family history of breast cancer | 4-point Likert scale to evaluate nine aspects of the intervention | GC had more Excellent/Good ratings than computer intervention for a) "Providing enough information to decide" among high risk women ($p = 0.01$); for b) "Providing reassurance" among low risk women ($p = 0.02$); and c) "Making good use of time" among low risk women ($p = 0.03$); no difference for other items. |
| Halbert et al. 2012 | African American women at increased risk for BRCA1/2 mutation | Satisfaction with Decision Scale (Holmes-Rovner, 1996) | Women who participated in genetic counseling were more satisfied than non-participants 18.0 vs. 16.9, respectively, $p = 0.01$. |

**Table 4** (continued)

| Study | Study population | Measures | Main results |
|---|---|---|---|
| Hunter et al. 2005 | AMA prenatal pts; gestation < =18 weeks; no previous consideration of prenatal diagnosis; English speaking | Intervention Satisfaction Questionnaire (ISQ)<br>11 item short form of satisfaction with genetic counseling scale (Shiloh et al. 1990) | Individuals who received traditional genetic counseling were significantly more satisfied than those who had group counseling ($p < 0.001$ for women and $p < 0.005$ for men) or decision aid group ($p < 0.001$ for women and $p < 0.001$ for men.). |
| Genetic Testing (Attitude, Intention, or Receipt) | | | |
| Bowen et al. 2002 | Women with one relative with breast cancer (family history suggestive of BRCA excluded) | One question "Do you think that you would be an appropriate candidate for genetic testing" | Following GC, participants were less likely to view themselves as appropriate candidates for genetic testing than controls (OR = 8.55, 95% CI =3.6–20.3), $p < 0.0001$. |
| Ciske et al. 2001 | Parents of children with CF | Reported completion of carrier testing among parents | Parents who underwent GC were more likely to undergo carrier testing ($p < 0.001$). |
| Green et al. 2001 | English speaking women 18y + with a first degree relative with breast cancer | Two questions, "if a blood test to look for an abnormality in a breast cancer susceptibility gene (BRCA1 or BRCA2) was offered to you today, what do you think you would do? (1 = I would definitely get tested; 5 = I would definitely not get tested) and "are you interested in obtaining a genetic test?" | Both GC and computer intervention groups demonstrated decreases in the proportion of low-risk women intending to pursue genetic testing, but there was no difference between the GC intervention and the computer intervention ($p = 0.70$). |
| Green et al. 2004 | women with personal or family history of breast cancer | The question, "if a blood test to look for an abnormality in a breast cancer susceptibility gene (BRCA1 or BRCA2) was offered to you today, what do you think you would do?" (1 = I would definitely get tested; −5 = I would definitely not get tested)<br>Receipt of genetic testing @ 6 months post-intervention | Both GC and computer intervention groups demonstrated decreases in the proportion of low-risk women intending to pursue genetic testing ($p < 0.001$ for both groups); more pronounced effect in the GC group ($p = 0.07$ for group comparison). Neither intervention changed the already very high levels of intent to test among the highest risk women.<br>No difference between GC and computer groups in actual receipt of testing at 1- or 6-months post intervention. |
| Randall et al. 2001 | women with breast cancer presenting for cancer genetic counseling v controls (women with breast cancer but not seeking genetic counseling) | Perceived importance of benefits and limitations of undergoing testing | The GC group had a significantly higher degree of concern about genetic testing than controls ($t56 = 2.54$, $P = 0.014$., |
| Health behavior or health outcome | | | |
| Grant et al. 2013 | Participants, 21 years or older who are overweight, met one criterion for metabolic syndrome without a diagnosis of type II diabetes and were willing to take part in a 12-week group session to achieve weight loss (diabetes prevention program) | Self-reported measures of risk perception, motivation, confidence, and stage of change upon enrollment in study, after genetic counseling intervention (before 12-week program), and then after 12-week program. Stage of change instruments are validated measures for assessing motivation | Exploratory analysis revealed that higher-risk participants were more likely to indicate that the initial genetic counseling intervention made them more "motivated to take part in the 12-week program (78.6% versus 43.8% in lower risk participants, $p < 0.003$) and to make lifestyle changes (85.7% versus 56.3% for lower risk participants, $p < 0.008$)". But this did not result in changed outcomes (see below). |
| | | Diabetes prevention classes (12-week Lifestyle Balance program) attended<br>Weight loss, % BMI reduction, % of weight | Receiving higher or lower genetic risk result + genetic counseling did not result in statistically significant changes in attendance at group classes, weight loss, BMI reduction or losing 7% of |

**Table 4** (continued)

| Study | Study population | Measures | Main results |
|---|---|---|---|
| | | | body weight when compared to the untested control group. |
| McInerney-Leo et al. 2006 | Women and Men from 13 extended HBOC families with previously identified mutation and who completed baseline and follow-up questionnaire and who declined genetic testing | Mammogram, breast self exam, CA-125 measures, pelvic ultrasound; compared self-reported screening behaviors at baseline and at 6–9 months after consultation | No change in frequency of breast self exam or CA-125; small increase in number of women having mammogram (before age 40) and increase of one woman having pelvic ultrasound (only raw data presented; no statistical tests performed) |
| Meiser et al. 2001 | Women at risk of developing hereditary breast cancer | Mammography, clinical breast exam and breast self-exam | No change in mammography or breast self-exams. Significant decrease in clinical breast exams (92% were vigilant at baseline; 86% at 12 months follow-up, $p = 0.041$). |
| Rutherford et al. 2014 | Pediatric genetics clinic patients/parents | Adherence to medical recommendations | Patients seen with a GC were more likely to follow the medical recommendations that were made at the genetics consult (79% completion rate for GC + MD vs 65% rate for MD alone; $p = 0.009$) |
| Taylor and Wu 2009 | African American women with hypertension and their 1st and 2nd degree relative | Physical activity (minutes), sodium intake (calculated using self-reported food intake), body mass index, systolic and diastolic blood pressure | Six-months after GC, systolic and diastolic blood pressures decreased slightly, women performed more physical activity, and they reduced their sodium intake. However the changes were not statistically significant. |
| Decisional conflict | | | |
| Christie et al. 2012 | breast cancer patients meeting NCCN cancer genetics referral criteria presenting either before definitive surgery (BDS) or after (ADS) | Decisional Conflict Scale | Pre-test GC led to a marginally significant decrease in overall decisional conflict (median change of −10.2, $p = 0.056$) and a significant decrease in the subscale for informed decision-making conflict (median change −25.0, $p < 0.001$) for ADS patients. |
| Green et al. 2004 | women with personal or family history of breast cancer | Decisional Conflict Scale | Overall, decisional conflict was lower in the GC group compared to computer group ($p = 0.04$). However when groups were stratified into high- and low-risk subgroups, there was no difference. |
| Hunter et al. 2005 | AMA prenatal pts; gestation < =18 weeks; no previous consideration of prenatal diagnosis; English speaking | Decisional Conflict Scale | Decrease in decisional conflict post intervention in all groups; decision aid showed greater decreases than group counseling ($p < 0.016$). |
| Family outcomes | | | |
| MacDonald et al. 2007 | Women with personal and/or family history of breast or ovarian cancer presenting for genetic counseling | Self-reported discussions with first degree relatives, and checklist of barriers to communication | Risk communication with first degree relatives increased slightly after GC, but the change was not statistically significant. Barriers to communication decreased, but the change was not statistically significant. |
| McInerney-Leo et al. 2005) | Women and Men from 13 extended HBOC families with previously identified mutation and who completed baseline and follow-up questionnaire and who declined genetic testing | Family Relationship Index (FRI) of Family Environment Scale (FES) measures cohesion (degree of commitment, help, and support), expressiveness (encouraged to act openly and express their feelings directly), and conflict (openly expressed anger, aggression, and conflict among family members) | Family cohesion improved (mean of 6.79 at baseline to 8.00 at 6-9 months, $p < 0.001$). No statistically significant change in expressiveness or conflict. |

**Table 4** (continued)

| Study | Study population | Measures | Main results |
|---|---|---|---|
| Perceived personal control | | | |
| Berkenstadt et al. 1999 | Patients with a genetic problem at the time of counseling for age-related prenatal diagnosis | Developed the Perceived Personal Control Questionnaire to investigate patients' subjective perceptions of their level of control with regard to their genetic problem. Comprised of 9 items representing three aspects of control: cognitive, behavioral and decisional. Rated on a 3-point scale of agreement: do not agree (0), somewhat agree (1), completely agree (2). | Perceived personal control significantly higher post-counseling in all three aspects of control ($p < 0.001$). Higher post-counseling PPC was associated with getting a definite diagnosis ($F = 8.32$, $p < 0.001$) and an exact recurrence risk ($F = 19.9$, $p < 0.001$) and being offered prenatal diagnosis ($F = 8.80$, $p < 0.001$). Post-counseling PPC was significantly correlated with knowledge, satisfaction, counseling evaluations, and expectation fulfillment ($p < 0.01$). |
| Pieterse et al. 2011 | Women presenting for BRCA genetic counseling | Perceived Personal Control Questionnaire – 9 questions | Perceived control improved after GC among women without a cancer diagnosis immediately after GC ($p < 0.01$, $\chi^2 = 6.64$) and at 6-month follow-up ($p < 0.001$, $\chi^2 = 10.83$). There was no change among women with a cancer diagnosis. |
| QOL | | | |
| Cabrera et al. 2010 | Patients without a cancer diagnosis presenting for BRCA genetic counseling/with a family history of breast cancer | EuroQuol 5 dimension describes 5 dimensions of the health state. There is also a visual analog scale ranging from the worst health (0) to the best health state (100) | No change in QOL from baseline to 1 month or 6 month time points. |
| Self-Esteem | | | |
| McInerney-Leo et al. 2004 | Women and men from 13 extended HBOC families with previously identified mutation and who completed baseline and follow-up questionnaire and who declined genetic testing | Rosenberg Self-Esteem Scale (Global self esteem) | No change in self-esteem from baseline to 6-9 month time point |

*BRCA* breast cancer, *HBOC* hereditary breast and ovarian cancer, *CF* cystic fibrosis, *NHGRI* National Human Genome Research Institute, *AMA* advanced maternal age

counseling and genetic testing. With genetic tests available for over several thousand genetic conditions, genetic testing is an increasingly common part of the genetic counseling process in many practice settings. If outcomes are measured after both genetic counseling and genetic testing have occurred, it is difficult to assess whether the outcomes are the result of the counseling, testing, or a combination.

To overcome the many complicating factors mentioned above, we have developed six recommendations for moving forward with genetic counseling outcomes research described below.

1. Improve published descriptions of genetic counseling interventions

   It is important to design studies that consider and better document what was done in the genetic counseling session and by whom. This may help overcome several of the challenges we describe related to a variety of provider types who offer genetic services and variability in practices that may occur even within the field of genetic counseling.
2. Design studies that distinguish outcomes of genetic counseling vs genetic testing

   Our review criteria led to the elimination of several studies because we were unable to discern the effects of these two separate, yet highly intertwined interventions. It is critical to capture how genetic counselors may be helping individuals adapt after they receive their test results. Indeed our inclusion criteria may partially explain the relatively small number of outcome types identified in our review.
3. Conduct literature reviews or longitudinal studies aimed at identifying appropriate intermediate endpoints, or at developing an indirect chain of evidence linking proximal to distal outcomes

   Research is needed to determine which, if any, of the previously studied outcomes are most strongly correlated with or influence more distal or long-term outcomes such

as morbidity, mortality, mental health, and social health. One of the primary goals of outcomes research will be to identify measures that can be used to evaluate the quality of genetic counseling delivered by various providers. Importantly, the U.S. National Quality Measures Clearinghouse does not consider most of the "outcomes" we identified in this review to be outcomes until or unless there is sufficient evidence showing they influence the aforementioned distal health outcomes.

4. Consider strategic inclusion of outcomes that are widely accepted by healthcare organizations to facilitate outcomes-based reimbursement

   Given that genetic counselors function in healthcare settings highly concerned about reimbursement, it may be strategic to focus on "outcomes" that are strongly correlated with or have been shown to influence distal health outcomes. Nevertheless, we should recognize that certain "outcomes" may be important to patients even if they are not linked to health outcomes. Outcomes that are highly valued by patients could be considered for a different type of quality measure by the National Quality Measures Clearinghouse because they reflect patient-centered care.

5. Increase the use of theoretical models or frameworks when designing outcomes studies

   Theories, frameworks, and models may help researchers consider a broader array of outcomes and they also can provide a rationale for why we expect what we do to have an effect on certain outcomes (or not). In planning our review we did not consider extracting data about whether studies were informed by theoretical models or frameworks. However, we noted that the vast majority of studies did not mention these in their design considerations. Furthermore, several studies failed to provide a compelling rationale for why they selected the measures they chose.

6. Develop a standard set of well-validated measures that can be harmonized across multiple types of genetic counseling studies

   Finally, having a standard set of defined outcomes and measures should help us more easily and more robustly build an evidence base for the genetic counseling profession; this would also allow for study comparisons and meta-analyses in the future. However, we do not believe that this review provides enough information to make recommendations about standard measures. Before making such recommendations we believe it would be prudent to further evaluate and consider of the following:

   - Data on what outcomes are most important to patients/clients as well as other stakeholders (i.e., third-party payers)
   - Theories, models, frameworks, and data to provide a rationale for or evidence linking genetic counseling processes to outcomes we identified and to distal health outcomes
   - Review of outcome studies that did not meet our inclusion criteria or that were conducted in other healthcare contexts outside genetic counseling
   - The extent to which the measures have been demonstrated to be reliable, valid, and sensitive to change in various genetic counseling settings

## Conclusions

To date, there are no consistent measures of genetic counseling outcomes across studies. However, there is evidence that genetic counseling can increase knowledge, decrease distress, and lead to benefits for patients across several outcome measures. There is a need for further outcomes research measuring longer term and health outcomes and for research in a wider variety of genetic counseling settings.

**Acknowledgements** Support for Deborah Cragun's time was provided by a NCI R25T training grant awarded to Moffitt Cancer Center (5R25CA147832-04). The authors thank Eliza Jeong for assistance with formatting.

This manuscript was unfunded work completed as part of the National Society of Genetic Counselors Outcomes work group. We would like to thank NSGC leadership for helping to formulate the working group's study inclusion criteria for this review and for providing comments on an earlier draft of this manuscript.

**Compliance with Ethical Standards**

**Conflicts of Interest** Lisa Madlensky, Angela M. Trepanier, Deborah Cragun, Barbara Lerner, Kristen M. Shannon and Heather Zierhut declare that they have no conflict of interest.

**Human Studies and Informed Consent** No human studies were carried out by the authors for this article.

**Animal Studies** No animal studies were carried out by the authors for this article.

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