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ORIGINAL RESEARCH

Defining Our Clinical Practice: The Identification of Genetic Counseling Outcomes Utilizing the Reciprocal Engagement Model

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Abstract The need for evidence-based medicine, including comparative effectiveness studies and patient-centered outcomes research, has become a major healthcare focus. To date, a comprehensive list of genetic counseling outcomes, as espoused by genetic counselors, has not been established and thus, identification of outcomes unique to genetic counseling services has become a priority for the National Society of Genetic Counselors (NSGC). The purpose of this study was to take a critical first step at identifying a more comprehensive list of genetic counseling outcomes. This paper describes the results of a focus group study using the Reciprocal-Engagement Model (REM) as a framework to characterize patient-centered outcomes of genetic counseling clinical practice. Five focus groups were conducted with 27 peer nominated participants who were clinical genetic counselors, genetic counseling program directors, and/or outcomes researchers in genetic counseling. Members of each focus group were asked to identify genetic counseling outcomes for four to five of the 17 goals of the REM. A theory-driven, thematic analysis of focus group data yielded 194 genetic counseling outcomes across the 17 goals. Participants noted some concerns about how genetic counseling outcomes will be measured and evaluated given varying stakeholders and the long-term nature of genetic concerns. The present results provide a list of outcomes for use in future genetic counseling outcomes research and for empirically-supported clinical interventions.

Keywords Reciprocal-engagement model · Genetic counseling · Outcomes · Focus group · Goals · Outcomes research

Introduction

With continual advancements in genetic technology, the genetic counseling profession sits at the forefront of the changing landscape of genetic medicine. Genetic counseling is a relatively recent (~45 years) and specialized medical profession that involves helping "people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease" (Resta et al. 2006, p. 77). Genetic counselors serve many roles (e.g., educator, supporter, medical liaison, multidisciplinary team member, resource provider) as they work to provide information and support for individuals and families dealing with complex genetic information. With new technologies (e.g., whole genome sequencing), integration of genetics into primary care settings, and a changing healthcare climate, the profession continues to evolve. As with all areas of clinical healthcare, there is a need to better establish evidence-based practice.

Evidence-based medicine, informed by comparative effectiveness studies and patient-centered outcomes research, has become a major healthcare focus, especially in response to the passing of the Patient Protection and Affordable Care Act of 2010 (Supreme Court of the United States 2013). The necessity to identify a comprehensive set of outcomes and outcome measures for use in a mandatory reporting system (Physician

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Quality Reporting System) is a fast approaching reality in the United States (US) Healthcare System (Centers for Medicare and Medicaid Services 2013). The mandatory reporting system is important in that all recognized providers in Centers for Medicare and Medicaid Services (CMS) must report outcomes for full reimbursement of services. Without an agreed upon set of outcomes and quality measures, if genetic counselors do become members of CMS, the field would lack an agreed upon set of evidence-based outcomes. Momentum now exists to demonstrate all clinical interventions, including genetic counseling, have beneficial outcomes. To that end, identification of outcomes unique to the profession has become a top priority for the National Society of Genetic Counselors (NSGC; Access and Service Delivery 2014).

Outcomes in Genetic Counseling

Definitions of the term "outcome" vary, and they are context-dependent. In the health care literature, healthcare outcomes have been defined as "the end result of what happens to patients as a consequence of their encounter(s) with the healthcare system" (Krousel-Wood 2000, p. 235). Thus, research on health care outcomes specifically seeks to understand the end result of particular health care practices and interventions (Outcomes Research 2000). An "outcome measure" can be defined as an assessment tool used to provide a valid and reliable evaluation of this end result or outcome.

To date, a limited range of outcomes has been evaluated in genetic counseling research. Historically, the major goals of genetic counseling were to "educate clients about etiology and recurrence risks and to assist clients in reproductive decision making" (Bernhardt et al. 2000, p. 189). As such, the bulk of the genetic counseling outcome research has focused on measuring educational (learning) and reproductive (decision making) variables (Berkenstadt et al. 1999). Four outcomes, in particular, are the focus of recent studies: (1) *patient knowledge* (Armeli et al. 2005; Baldwin et al. 2011; Hunter et al. 2005; Meiser et al. 2005); (2) *decision-making* – including patients' self-reported satisfaction with the decision-making processes, or their evaluation of their actual decisions and/or decision-related outcomes (Matloff et al. 2006; Nagle et al. 2006; Peterson et al. 2006; Rowe et al. 2006; Wakefield et al. 2007); (3) *patient satisfaction* (Bjorvatn et al. 2007; Davey et al. 2005; Hippman et al. 2013; Skirton et al. 2005); and (4) *psychological adaptation* to genetic information and/or genetic counseling services –defined, for example, as patient distress (Codori et al. 2005; Meiser et al. 2008), anxiety (Hunter et al. 2005; Pieterse et al. 2007), adaptation to genetic information (Read et al. 2005), and family functioning and communication (Gaff et al. 2005; Hallowell et al. 2005; Hamilton et al. 2005; Van Oostrom et al. 2006).

Although these outcomes comprise valid "end points," they fail to capture the full scope of genetic counseling practice. In a review of 67 validated measures of clinical genetic services (CGS) that included both "non-genetics specific" and "genetics-specific" measures, Payne et al. (2008) found a variety of outcomes measures, from anxiety to patient satisfaction and knowledge, were used, and they varied in the quality and the extent to which their psychometric properties had been demonstrated (e.g., validity, reliability). They noted that, "No single validated outcome measure encompasses all aspects of the potential patient benefits from using clinical genetic services" (Payne et al. 2008, pp. 504–505). They also concluded that outcome measurements evaluating other patient benefits, for example, hope in the patient and future generations, were not being used currently.

In response to this gap, McAllister and colleagues (McAllister et al. 2011) developed an outcome measure to evaluate clinical genetic services - the Genetic Counseling Outcomes Scale (GCOS-24). The GCOS-24 is a patient-reported outcome measure based on the construct of empowerment; it evaluates additional dimensions of perceived personal control that include patients' decisional, behavioral and cognitive control, as well as the constructs of emotional regulation and hope (McAllister et al. 2008, 2010). The GCOS-24 "fills a gap in the literature of clinical genetic service evaluation" (Pithara 2014, p. 230) and provides an important step towards a comprehensive measure of one aspect of genetic counseling. Of note, however, the GCOS-24 is based on a patient-reported outcome that was originally designed to evaluate outcomes of the Clinical Genetic Services (CGS) as a whole and not genetic counseling exclusively (Pithara 2014). Moreover, the measure was based on a CGS in the United Kingdom and while the systems may be similar, the measure has not been validated in the US. Arguably, the context of genetic counseling in the US (versus the CGS in the United Kingdom) varies; moreover, there remains a need to specifically examine genetic counseling outcomes within a framework of an exclusive genetic counseling model of practice.

The Reciprocal-Engagement Model (REM)

A model of practice for genetic counseling provides a critical framework as the field continues to document the processes and outcomes of genetic counseling (Bernhardt et al. 2000; McCarthy Veach et al. 2007). In 2007, McCarthy Veach et al. proposed the Reciprocal-Engagement Model (REM) as a more specific practice model for genetic counseling. The REM is informed by the results of a 2-day consensus conference attended by genetic counseling program directors or their representatives ($N=23$) from 20 of the 30 genetic counseling graduate programs in North America accredited at that time, extant literature, and the REM co-authors' professional experiences as educators, researchers, and practitioners (McCarthy Veach et al. 2007). Conference attendees were asked to define four components of the model of genetic counseling practice being taught at the time in genetic counseling programs. McCarthy Veach et al. (2007) used Rieh and Ray's (1974) definitions of the four components of a model: 1) *Tenet* - a principle, doctrine, or belief held in common by members of a group; 2) *Goal* - an aim or purpose; content specified as an aim for activity; 3) *Strategy* - a careful plan or method, especially for achieving an end; and 4) *Behavior* - an action or reaction; personal conduct. Utilizing these definitions, participants worked to develop consensus about the specific tenets and goals of genetic counseling. Time constraints precluded their ability to articulate more than a handful of strategies and behaviors and to discuss outcomes.

The REM consists of five, mutually influential genetic counseling tenets and 17 related goals (goals listed in Tables 3, 4, 5 and 6). The five tenets are: genetic information is key, relationship is integral to genetic counseling, patient autonomy must be supported, patients are resilient, and patient emotions make a difference. The genetic counselor-patient relationship is at the center of the model. Indeed, the counselor-patient relationship is the "conduit for processes and outcomes of genetic counseling" (McCarthy Veach et al. 2010, p. 3). The REM authors postulate three broad counseling outcomes: "Patient understands and applies information to: make decisions, manage condition, and adapt to situation" (McCarthy Veach et al. 2007, p. 724). They based these outcomes on REM process goals and say they are a "work in progress" (McCarthy Veach et al., p. 724); that is, they do not comprise an exhaustive outcomes list.

Overall, the REM provides a comprehensive model of practice on which to base education, training, and clinical practice of genetic counselors. Additionally, given the limited amount of empirical research on genetic counseling processes and outcomes, the REM provides a basis on which to "develop and execute research that evaluates genetic counseling service provision in the clinical setting" (McCarthy Veach et al. 2007, p. 714) and directly link specific aspects of genetic counseling processes to genetic counseling outcomes.

Recently Hartman and colleagues (Hartmann et al. 2015) obtained evidence for the validity of the REM's 17 goals in a survey study of 194 genetic counselors. Factor analysis of the 17 goals yielded four factors that accounted for 51 % of the variance in respondents' ratings of the importance of each goal: *Understanding and Appreciation*, *Support and Guidance*, *Facilitative Decision-Making* and *Patient-Centered Education*. Mean importance ratings suggest practicing counselors regarded each goal as important. The researchers also content-analyzed participants' comments about REM goals. A prevalent theme concerns the importance of goals and goal attainment; specifically, participants commented that goals may vary in relevance and feasibility based on patient and/or genetic counseling specialty. In addition, some individuals noted goal importance and attainment are influenced by logistical factors such as time-constraints of sessions. Hartmann et al. (2015) concluded these themes further support the need for clearly articulated genetic counseling outcomes.

Purpose of the Study

While literature exists on genetic counseling outcomes in various sub-specialties, and outcome measures have been used in some genetic counseling research (as reviewed in Kasparian et al. 2007), the profession continues to lack a comprehensive list of distinct genetic counseling outcomes. This lack is partly due to the fact that extant studies are not grounded in an empirically-informed theoretical model of genetic counseling such as the REM which would serve to cohesively frame the practice of genetic counseling. As Zierhut and Cohen (2013, p. 2) assert,

> Researchers appear to be taking a "backward approach." Instead of working from a framework that describes the provision of genetic counseling as an intervention, researchers have looked at selected outcomes that are loosely connected to theories or models of genetic counseling.

The present study takes the first step towards identifying a comprehensive list of genetic counseling outcomes[1] by using the REM of genetic counseling practice as a guiding framework. A more thorough documentation of genetic counseling outcomes will not only help define the field's current contributions in healthcare, but also provide a foundation on which to base future research and empirically-supported clinical interventions. Accordingly, focus groups were conducted with nominated participants (practicing genetic counselors, outcomes researchers, and/or genetic counseling program directors) who were asked to identify patient-centered genetic counseling outcomes and genetic counselor behaviors associated with the 17 goals of the REM. The findings reported herein focus on patient-centered genetic counselor outcomes.

Methods

Participants

Upon receipt of approval from the University of Minnesota and St. Vincent Hospital Institutional Review Boards, focus group participants were identified through a nomination process overseen by the Access and Service Delivery (ASD) Committee of the NSGC. A total of 91 nominations were elicited from the ASD committee for genetic counselors who met at least one of the following criteria: greater than 5 years of genetic counseling experience, expertise in genetic counseling outcomes research, or genetic counseling graduate program director. The Genetic Counseling Outcomes Measures Working Group, a sub-committee of the ASD, reviewed the nominations to determine a final list of 54 invitees. Possible invitees were also asked to nominate additional participants. The final sample consisted of 27 participants.

Four focus groups were conducted at the 2013 NSGC Annual Education Conference (AEC) in Anaheim, California, and one additional focus group was conducted following the AEC in December of 2013.

Instrumentation

Interview Guide

A detailed focus group guide was developed by the first author and reviewed by the principle investigator (Dr. Heather Zierhut) and Dr. Patricia McCarthy Veach, co-author of the REM. The guide consisted of two broad questions and corresponding prompts that were asked of participants in reference to each of the 17 REM goals. The specific questions and prompts were:[1]

1) What does this [REM goal] look like in a genetic counseling session?

   Facilitator prompt: How could you "measure" this?

2) How would a genetic counselor know if this REM goal is achieved?

   Facilitator prompts:

   - What does the genetic counselor say or do to know if this is achieved? (Non-verbal behaviors? Verbal behaviors? Give an example of what you would say to a patient? What would you do?)
   - What does the patient say or do to know if goals are achieved? (What about non-verbal behaviors? Verbal behaviors?)

Demographic Form

Demographic data were collected on each focus group participant. Questions asked about participant age, gender, years of experience (as a practicing genetic counselor, training director, and/or outcomes researcher), NSGC region, specialty area of practice, and primary work setting. Participants were also asked about their experience with genetic counseling outcomes through two questions, respectively asking them to rate their prior knowledge of genetic counseling outcomes (4-point scale with behavioral anchors for two number: 1=*no knowledge* to 4=*extensive knowledge*), and whether their institution had approached them about tracking outcomes and if they were currently doing this (open-ended).

Procedures

One week prior to the focus groups, participants received logistical information about the focus groups, written materials describing the REM, and the McCarthy Veach et al. (2007) and Hartmann et al. (2015) publications. In interest of time and a desire to have a more thorough discussion of each REM goal, each focus group was assign to discuss four to five of the 17 REM goals. Due to a clerical error, four REM goals (i.e., REM goals 10–13) were not covered at the AEC focus groups. Therefore, an additional invitation-only focus group was conducted in December of 2013 to discuss these four goals. Participants in this additional focus group were part of the initial nomination list and invited to participate based on their expertise in the genetic counseling process and/or familiarity with research in genetic counseling outcomes. For consistency, this additional focus group utilized the same procedure as the groups conducted at the AEC.

Each focus group was facilitated by an experienced focus group moderator/genetic counseling researcher. A conference call prior to the AEC took place with the principle investigator (Dr. Heather Zierhut) and the four focus group moderators (Bonnie LeRoy, Dr. Patricia McCarthy Veach, Dr. Ian MacFarlane, and Ms. Krista Redlinger-Grosse). During this call, the moderator guide, REM, and the focus group goals and procedures were reviewed. In addition, the process of the focus group was discussed in detail and there were no further modifications made to the moderator guide. Minor changes to the demographic form were made to re-order questions and clarify response categories. In addition, a note-taker was assigned to each focus group and was responsible for recording the discussions and taking detailed notes.

The focus groups were 2 hours in length and audio-recorded. At the beginning of each group, participants reviewed an information sheet about the study, which served as informed consent. They also completed a short, demographic survey and reviewed their assigned REM goals and the study definition of *outcome*. For this study, an *outcome* was defined as, "the end result of what happens to patients as a consequence of their encounter(s) with the healthcare system" (Krousel-Wood 2000, p. 235). Participants were also given an example of a patient-centered genetic counselor behavior, the outcome and measure of that outcome (See Fig. 1).

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[1] Throughout this paper, *genetic counseling outcomes* refer to outcomes espoused by genetic counselors providing genetic counseling, in other words, genetic counselor-defined outcomes.

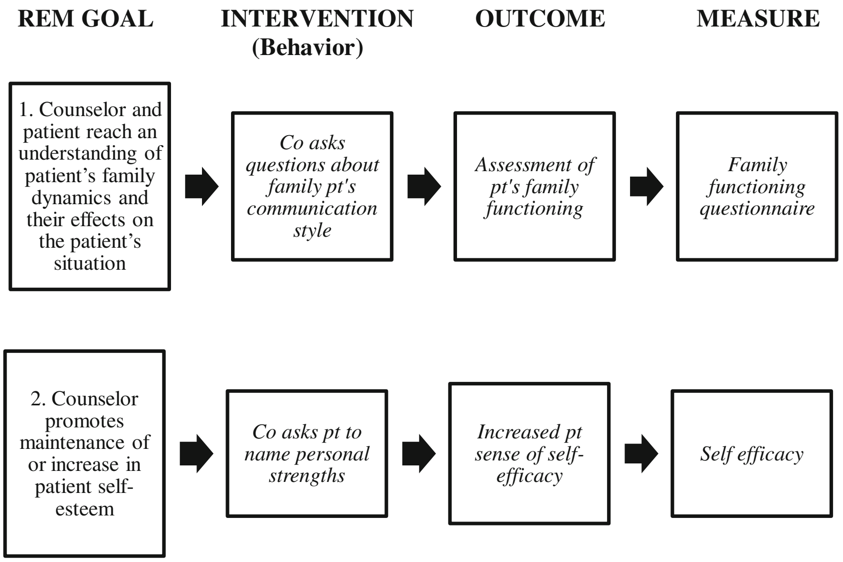

Fig. 1 Focus group visual of REM goal and associated genetic counselor behavior, outcomes, and measure

Open discussion regarding behaviors and outcomes associated with the REM goals ensued. Moderators read each assigned REM goal one at a time and posed questions to participants regarding that REM goal.

Data Analysis

Audio recordings of each focus group were transcribed verbatim and qualitatively analyzed. The first author used a theory-driven process utilizing the REM to analyze the focus group data (MacFarlane and O'Reilly-de Brun 2012). Following the process described by MacFarlane and O'Reilly-de Brun (2012), she engaged in an iterative process to extract themes, regarding genetic counseling outcomes using the 17 REM goals and the four factors described by Hartmann et al. (2015) as a framework. Specifically, she reviewed each focus group transcript to extract comments regarding outcomes that she then grouped into outcome domains (i.e., rationally derived topic areas) that reflected each of the 17 REM goals. She continually reviewed each domain, making modifications to better reflect the data. Throughout this iterative process, she made notes to provide context to the domains. The last author audited the outcome domains, and any disagreements were discussed to reach consensus. The first author again reviewed the transcripts using a final codebook until the point of saturation (i.e., no additional outcomes were identified). Next, she grouped the final list of genetic counseling outcomes for the 17 REM goals into the four factors identified by Hartmann et al. (2015) that were earlier described (e.g., *Understanding and Appreciation*, *Support and Guidance*, *Facilitative Decision-Making* and *Patient-Centered Education*). She used NVivo 10 software to aid in the coding process. For REM goals 14–17 that were covered in two separate focus groups, the data analyst used cross-case analysis to check for consistency in the identified outcomes.

Descriptive statistics were calculated for demographic data and self-rated knowledge regarding genetic counseling outcomes.

Results

Participants Characteristics

Twenty-seven individuals participated in one of five focus groups (11 genetic counseling graduate program directors, 10 practicing genetic counselors, and 6 outcomes researchers). All were female and their mean age was 47.3 years (*SD*= 10.10). Participants had a mean of 14.3 years of clinical experience (*SD*=9.08), 7.0 years of research experience (*SD*=5.97), and 9.5 years in genetic counseling related activities (teaching and program directors; *SD*=6.44). The median self-rated prior knowledge of genetic counseling outcomes was 3.0 (Range=1–4). Additional demographic and professional characteristics are presented in Tables 1 and 2.

Seven participants indicated they were involved in ongoing outcomes research in genetic counseling, and six participants had published in the area of genetic counseling outcomes (Range=1–3 publications). Five participants indicated their institutions had approached them about tracking genetic counseling outcomes, with suggested measures including test uptake, adherence, attendance, and compliance with quality indicators. Three participants stated their institutions already implemented measurement of genetic counseling outcomes ("tracking through patient registries," "examining management adherence," "patient satisfaction, knowledge").

Table 1 Demographic and professional characteristics of focus group participants (N=27)

Variable	Total n (%)
Gender	
Female	27 (100 %)
Male	0 (0 %)
Age (Years)	
20–29	1 (3.7 %)
30–39	5 (18.5 %)
40–49	12 (44.4 %)
50–59	5 (18.5.%)
60–69	3 (11.1 %)
>70	1 (3.7 %)
Geographic region[a]	
Region I	2 (7.4 %)
Region II	4 (14.8 %)
Region III	2 (7.4 %)
Region IV	13 (48.1 %)
Region V	1 (3.7 %)
Region VI	5 (18.5 %)
Primary work setting[b]	
University medical center	19 (70.4 %)
Other	8 (29.6 %)
Specialty area[b]	
Cancer	7 (25.9 %)
Prenatal	6 (22.2 %)
Public health	5 (18.5 %)
Pediatric	4 (14.8 %)
Psychiatric	2 (7.4 %)
Molecular/Cytogenetics	1 (3.7 %)
Other	14 (51.9 %)
Primary role	
Teaching/Education/Supervision	12 (44.4 %)
Clinical care	6 (22.2 %)
Research/Study coordinator	5 (18.5 %)
Clinical management	1 (3.7 %)
Healthcare administration	1 (3.7 %)
Management (for profit/non-profit)	1 (3.7 %)
Other	1 (3.7 %)

[a] US regions defined by the NSGC; [b] Participants could select more than one category

Genetic Counseling Outcomes

Participants in each focus group identified genetic counseling outcomes for a different subset of each of the 17 REM Goals. Across the groups, they identified a total of 194 genetic counseling outcomes that fit within the theoretical framework of the REM and the 17 goals. Every outcome mentioned by focus group participants was able to be classified within one of the REM goals.

Tables 3, 4, 5 and 6 contain a list of the outcomes, organized according to each REM goal, which in turn, is categorized within one of the four factors identified by Hartmann et al. (2015) in their validation study of the REM: *Understanding and Appreciation* (see Table 3); *Support and Guidance* (see Table 4); *Facilitative Decision-Making* (see Table 5); and *Patient-Centered Education* (see Table 6). The number of genetic counseling outcomes identified varied across goals and factors (see Table 7).

Forty-one genetic counseling outcomes were identified in multiple REM goals (see Table 8). The range in which outcomes were mentioned in multiple goals was two to eight REM goals. Four outcomes were mentioned across all four factors: A change in patient's ability to engage in the genetic counseling process; A change in patient's medical management; A patient is satisfied with genetic counseling service; and A change in patient's genetic knowledge.

Perceived Challenges in Evaluating Genetic Counseling Outcomes

Participants in all five focus groups voiced unprompted opinions and concerns regarding how genetic counseling outcomes would be measured and evaluated. With regard to measurement, participants in every group discussed the challenge of quantifying genetic counseling outcomes. For instance, some commented that outcomes and how they are measured would change as genetic technology changes:

> The theory right now, in my understanding, is that genomics medicine will be more focused on health behaviors, and that's going to be the outcome. That changes how you think about what the measures are here,

Table 2 Years of experience of focus group participants (N=27)

	Mean	SD	Median	Range
Clinical genetic counseling[a]	14.9	8.8	15	2–34
Genetic counseling research[b]	8.2	5.6	8	1–20
Genetic counseling related activities (Teaching, Program directors)[c]	11	5.6	10	1–20

[a] Total respondents n=24; [b] Total Respondents n=17; [c] Total Respondents n=19

Table 3 Genetic counselor outcomes by factor "*Understanding and Appreciation*" and inclusive of REM goals 1–6

REM Goal	Outcome
(1) "Counselor and patient reach an understanding of patient's family dynamics and their effects on the patient's situation"	An assessment of patient's family functioning
	An assessment of patient's support system
	A patient receives care that is congruent with their needs
	A patient considers their family in decision making
	A patient's informational needs are met
	A change in patient's level of regret regarding their decision
	A change in patient's resolution of crisis
	A change in the communication of genetic information to/within the patient's family
	A change in the patient's understanding of the long term impact of their decision
	A changes in uptake of genetic counseling by the patient's family members
(2) "Counselor promotes maintenance of or increase in patient self-esteem"	A change in patient's locus of control
	A change in patient's perceived power of control
	A change in patient's quality of life
	A change in patient's self-efficacy
	A change in patient's self-esteem
	A patient is empowered
(3) "Counselor facilitates the patient's feelings of empowerment"	A change in communication regarding genetic information within the patient's family
	A change in patient's comfort level with their decision
	A change in patient's communication with their referring provider
	A change in patient's emotional state
	A change in patient's empowerment
	A change in patient's follow through on health recommendations
	A change in patient's medical management
	A change in patient's perception that they are validated
	A change in patient's level of regret regarding their decision
	A patient overcomes barriers to their care
	A patient takes proactive behavior or steps in their care
(4) "Counselor integrates the patient's familial and cultural context into the counseling relationship and decision-making"	A change in patient's ability to engage in the genetic counseling process
	A change in patient's adaptation
	A change in patient's level of regret regarding their decision
	A patient is empowered
	A patient is satisfied with genetic counseling service
	A patient receives competent care
	A patient receives relevant information
	A patient's culture is incorporated into their decision making process
	A patient's decision is implemented
	A patient's decision making is autonomous
	A patient's decision making is facilitated
	A patient's informed decision making
	A patient's religious culture is incorporated into a discussion in the session
	An alliance between the patient and genetic counselor is formed
(5) "Counselor works with patient to recognize concerns that are triggering the patient's emotions"	An alliance between the patient and genetic counselor is formed
	A change in patient's ability to comprehend genetic information
	A change in patient's ability to engage in the genetic counseling process
	A change in patient's autonomy
	A change in patient's distress
	A change in patient's navigation of medical system
	A change in patient's perception that their needs are respected

Table 3 (continued)

REM Goal	Outcome
(6) "Counselor establishes a working contract with a patient"	A change in patient's daily functioning
	A change in patient's genetic knowledge
	A change in patient's medical follow up
	A change in patient's medical management
	A change in patient's perception that they receive empathy
	A change in patient's risk perception
	A change in patient's self-esteem
	A change in patient's level of regret regarding their decision
	A patient feels reassured
	A patient is empowered
	A patient is satisfied with genetic counseling service
	An alliance between the patient and genetic counselor is formed
	An assessment of patient's informational needs
	An assessment of patient's emotional needs
	An assessment of patient's supportive needs
	An educational agenda of the session is met

> because if the goal in genomic medicine is knowing enough to be able to change your behavior, then all the outcomes would be "Did your behavior change?" Right? (FG-1)

Some participants also expressed that measurement and evaluation of outcomes are dependent on specialty area. For example:

> Like what exactly are your outcomes? In cancer or cardiovascular, adherence and better health outcomes [that] reduce morbidity and mortality are pretty easy to measure, but I don't know how you measure in prenatal and even in pediatrics, that adaptation. How do you actually measure that? (FG-3)

Participants also mentioned the outcomes measured may differ for various stakeholders (e.g., genetic counselors, patients, third party payers). One participant stated:

> I think from a practical standpoint, we have to think about whom do we need to prove these outcomes to? Are we trying to prove it to ourselves to make ourselves better, which is a great goal, but we also need to prove this to the people who are paying for our services. What are truly the most important outcomes from that? Even if they are maybe not the best outcomes for us, or the ones that are most appealing to us, maybe they're the ones we need to focus on. I was trying to think of what those are (FG-3)

Participants also voiced concerns that varying outcomes for different stakeholders may make it difficult for genetic counselors to prove the "value-added" nature of genetic counseling that would be meaningful for all stakeholders.

The timing of when to evaluate genetic counseling outcomes was also discussed in all focus groups. Participants said genetic counseling outcomes may not be able to be evaluated within the session. As one individual stated, "I guess there's no time limit on when the outcome could happen. We're talking about events that happen during the session, but the outcome could be a year down the road" (FG-4). Many participants thought evaluating outcomes in genetic counseling may need to be done longitudinally. For example:

> But the hard thing is that even if in the ideal world you could get outcome feedback from patients, you'd have to do it at more than one time, because the patient that was so distraught that you ruined their day, a year down the road when they're pregnant again, they're going to, hopefully, realize the utility of that visit. But if you ask them that 45 min after the visit, the answer is not going to be the same. (FG-2)

Discussion

The present focus group study was designed to identify genetic counseling outcomes using the empirically-informed theoretical framework of the Reciprocal-Engagement Model (REM) of genetic counseling practice. Five focus groups with 27 peer nominated participants yielded 194 genetic counseling outcomes encompassing all 17 REM goals (see Tables 3, 4, 5,

Table 4 Genetic counselor outcomes by factor "*Support and Guidance*" and inclusive of REM goals 7–12

REM Goal	Outcome
(7) "Counselor recognizes patient strengths"	An alliance between the patient and genetic counselor is formed
	A patient provides accurate family history to the genetic counselor
	A patient receives accurate identification of tests
	A patient receives competent care
	A change in patient's distress
	A change in patient's emotional stability
	A change in patient's locus of control
	A change in patient's medical management
	A change in patient's genetic knowledge
	A patient re-contacts genetic counselor
	A patient is satisfied with genetic counseling service
	A patient's strengths are recognized
(8) "Counselor and patient establish a bond"	A trust is formed between the patient and genetic counselor
	A change in patient's ability to engage in the genetic counseling process
	A change in patient's genetic knowledge
	A changes in patient's confidence in the genetic counseling process
	A patient's emotional needs are addressed
	A patient's informational needs are addressed
	A patient's medical needs are addressed
	An alliance between the patient and genetic counselor is formed
(9) "Counselor's characteristics positively influence the process of relationship-building and communication between counselor and patient"	A patient receives care that is focused on their needs
	A change in patient's ability to engage in the genetic counseling process
	A change in patient's coordination of support resources
	A patient experiences empathy from the genetic counselor
	A patient receives compassion
(10) "Counselor helps the patient to gain new perspectives"	A change in patient's feeling of stigma
	A change in patient's ability to engage in the genetic counseling process
	A change in patient's ability to gain a new perspective
	A change in patient's anxiety
	A change in patient's confidence
	A change in patient's guilt
	A change in patient's hope
	A change in patient's mental health
	A change in patient's psychological resolution
	A change in the patient's family's health behaviors
	A change of patient's comfort level during genetic counseling
	A patient perceives their decision to be consistent with their values
	A patient receives medical options
	A patient's informed decision making
(11) "Counselor helps patient to adapt to his or her situation"	A change in patient's adaptation
	A change in patient's adherence to health behaviors
	A change in patient's distress
	A change in patient's genetic knowledge
	A change in patient's medical management
	A change in patient's perception that they receive empathy
	A patient is satisfied with genetic counseling service
	A patient takes ownership of their care
	An improvement in the patient's dissemination of genetic information within family
(12) "Counselor helps the patient to feel in control"	A change in patient's ability to ask appropriate questions regarding their medical care

Table 4 (continued)

REM Goal	Outcome
	A change in patient's ability to gain a new perspective A change in patient's ability to process information A change in patient's adaptation A change in patient's agitation A change in patient's coping A change in patient's genetic knowledge A change in patient's medical management A change in patient's perceived power of control A patient establishes a medical plan

and 6). To date, this is the first study to exclusively identify a list of genetic counseling outcomes specific to a model of genetic counseling practice.

As will be discussed, the findings not only contribute to the genetic counseling literature by documenting a more comprehensive set of practice-based outcomes, but they also provide a "database" per se that can utilized by future researchers interested in conducting focused outcomes research. In addition, the study findings further articulate the REM by providing additional and more specific outcomes stemming from each of the 17 goals. In the process of identifying outcomes, participants voiced unprompted concerns and anticipated challenges in measuring the identified outcomes that warrant acknowledgement. Overall, as the profession looks to better define its clinical practice, these findings point to several future avenues of research to more clearly and empirically establish genetic counseling specific outcomes.

Genetic Counseling Outcomes

In the original REM, genetic counseling outcomes were broadly identified as "Patient understands and applies information to: make decisions, manage condition, and adapt to situation" (McCarthy Veach et al. 2007, p 724). The present findings further operationalize and extend these broad "macro" outcomes.

Participants identified wide-ranging outcomes for each of the 17 REM goals that comprise four conceptual factors identified by Hartmann et al. (2015): Understanding and Appreciation, Support and Guidance, Facilitative Decision-making, and Patient-Centered Education. Within each of these factors, identified outcomes reference assessing patients' informational, emotional, and supportive needs. Other outcomes involve changes in patients' emotional states (e.g., changes in their adaptation, empowerment, and perceived power or control). Participants also identified outcomes that affect dynamics of the counseling session such as building of rapport, trust and engagement in the genetic counseling process, and working alliance between the patient and genetic counselor. The sample also mentioned longer-range outcomes that involve effects on patients' healthcare through test identification, decision-making, and changes in health behaviors and management. And finally, they identified outcomes that affect not only the patient but also their family member(s) (e.g., communication of genetic information within family, referral of family members for genetic counseling).

The wide range of outcomes identified by focus group participants has not been previously documented in the genetic counseling literature. Prior genetic counseling research has focused on outcomes related to educational and reproductive decision- making (Berkenstadt et al. 1999). More recent outcomes research involves variables related to patient knowledge (Armeli et al. 2005; Baldwin et al. 2011; Hunter et al. 2005; Meiser et al. 2005); decision-making (Matloff et al. 2006; Nagle et al. 2006; Peterson et al. 2006; Rowe et al. 2006; Wakefield et al. 2007); patient satisfaction (Bjorvatn et al. 2007; Davey et al. 2005; Hippman et al. 2013; Skirton et al. 2005); psychological adaptation (Codori et al. 2005; Hunter et al. 2005; Meiser et al. 2008; Pieterse et al. 2007; Read et al. 2005), and family functioning and communication (Gaff et al. 2005; Hallowell et al. 2005; Hamilton et al. 2005; Van Oostrom et al. 2006). In contrast, the outcomes identified in the present study represent a fuller picture of genetic counseling outcomes that warrant acknowledgement and further study.

In addition, the wide range of outcomes identified in this study alludes to the far-reaching impact of the work done by genetic counselors. This is not surprising given the current definition of genetic counseling states the process of genetic counseling integrates numerous aspects of work: "Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; education about inheritance, testing, management, prevention, resources, and research; and counseling to promote informed choices and adaptation to the risk or condition" (Resta et al. 2006, p. 77). Similarly, the REM of genetic counseling practice supports the complexity of the genetic counseling process in the areas of patient-focused support and guidance, understanding and appreciation, education, and

Table 5 Genetic counselor outcomes by factor "*Facilitative Decision-Making*" and inclusive of REM goals 13–15

REM Goal	Outcome
(13) "Counselor helps the patient to feel informed"	A change in communication regarding genetic information within the patient's family
	A change in patient's ability to ask appropriate questions regarding their medical care
	A change in patient's ability to engage in the genetic counseling process
	A change in patient's genetic knowledge
	A patient is empowered
	A patient receives a short-term counseling
(14) "The counselor knows what information to impart to each patient"	A change in patient's adaptation
	A change in patient's adherence to health recommendations
	A change in patient's comfort level with their decision
	A change in patient's genetic knowledge
	A change in patient's grieving
	A change in patient's guilt
	A change in patient's medical management
	A change in patient's morbidity
	A change in patient's mortality
	A change in patient's perceived power of control
	A change in patient's psychological state
	A mutual agenda for genetic counseling is set between genetic counselor and patient
	A patient is satisfied with genetic counseling service
	A patient perceives their decision to be consistent with their values
	A patient receives care that is congruent with their needs
	A patient receives relevent information
	A patient's decision making is autonomous
	A patient's expectations of genetic counseling are met
	A patient's needs are prioritized in the session
(15) "Counselor facilitates collaborative decisions with the patient"	A change in patient's ability to engage in the genetic counseling process
	A change in patient's ability to make an actionable plan based on information
	A change in patient's autonomy
	A change in patient's comfort level with their decision
	A change in patient's perceived power of control
	A change in patient's perception that they are respected
	A change in patient's perception that they are supported
	A change in patient's understanding of implications of genetic information
	A change in patient's understanding of the implications of their decision
	A change in patient's level of regret regarding their decision
	A collaborative conversation between the patient and genetic counselor
	A patient's decision making is autonomous

Table 6 Genetic counselor outcomes by factor "*Patient-Centered Education*" and inclusive of REM goals 16–17

REM Goal	Outcome
(16) "Counselor presents genetic information in way that the patient can understand"	A change in patient's adjustment
	A change in patient's anxiety
	A change in patient's coping
	A change in patient's feeling of stigma
	A change in patient's genetic knowledge
	A change in patient's level of engagement with information
	A change in patient's medical management
	A change in patient's understanding of implications of genetic information
	A change in the information exchanged between the genetic counselor and patient
	A collaborative conversation between the patient and genetic counselor
	A patient receives information presented to them at an appropriate level
	An alliance between the patient and genetic counselor is formed
(17) "Good counselor-patient communication occurs"	A change in patient's medical follow up
	A patient's emotional needs are addressed
	A change in patient's ability to engage in the genetic counseling process
	A change in patient's ability to express their needs
	A change in patient's adherence to health behaviors
	A change in patient's adjustment
	A change in patient's comfort level with their decision
	A change in patient's empowerment
	A change in patient's feeling of stigma
	A change in patient's medical management
	A change in patient's perception that they are supported
	A change in patient's perception that they receive empathy
	A change in the information exchanged between the genetic counselor and patient
	A change in the level of communication between the patient and genetic counselor
	A decision is made collaboratively between the patient and genetic counselor
	A patient is satisfied with genetic counseling service
	A patient utilizes genetic counselor as a resource
	A patient's informational needs are addressed
	A patient's informational needs are met
	A patient's medical needs are addressed,
	A trust is formed between the patient and genetic counselor
	An alliance between the patient and genetic counselor is formed
	Rapport is built between genetic counselor and patient(s)

decision-making (Hartmann et al. 2015; McCarthy Veach et al. 2007). Thus, it is not unexpected that the identified outcomes in the present study support the genetic counseling process described in both the definition of genetic counseling and the REM. In addition, the findings illustrate ways in which processes and outcomes are inevitably intertwined and provide evidence for the extensive work done by genetic counselors (Bernhardt et al. 2000; Biesecker and Peters 2001).

Table 7 Description of outcomes by REM factors and goals

REM Factor and Goals	Number of Outcomes
Understanding and appreciation	64
(1) Counselor and patient reach an understanding of patient's family dynamics and their effects on the patient's situation	10
(2) Counselor promotes maintenance of or increase in patient self-esteem	6
(3) Counselor facilitates the patient's feelings of empowerment	11
(4) Counselor integrates the patient's familial and cultural context into the counseling relationship and decision-making	14
(5) Counselor works with patient to recognize concerns that are triggering the patient's emotions	7
(6) Counselor establishes a working contract with a patient	16
Support and guidance	58
(7) Counselor recognizes patient strengths	12
(8) Counselor and patient establish a bond	8
(9) Counselor's characteristics positively influence the process of relationship-building and communication between counselor and patient	5
(10) Counselor helps the patient to gain new perspectives	14
(11) Counselor helps patient to adapt to his or her situation	9
(12) Counselor helps the patient to feel in control	10
Facilitative decision-making	37
(13) Counselor helps the patient to feel informed	6
(14) The counselor knows what information to impart to each patient	19
(15) Counselor facilitates collaborative decisions with the patient	12
Patient-centered education	35
(16) Counselor presents genetic information in way that the patient can understand	12
(17) Good counselor-patient communication occurs	23

A Comprehensive "Database" of Outcomes

The REM was initially developed as a process model which could potentially serve as a "foundation for evaluating the effectiveness of the service" (McCarthy Veach et al. 2007, p. 726) by providing a framework for research in genetic counseling that is based on a model of practice (Fox et al. 2007). As such, the comprehensive list of outcomes identified in this study comprises the next step in constructing such a framework by providing a "database" per se of genetic counseling outcomes that can be used in future investigations.

The 194 genetic counseling outcomes identified by focus group participants are presented in their "raw" or unaltered forms in Tables 3, 4, 5 and 6. They are purposefully presented in a format that allows future researchers to use the list to help guide and develop outcomes studies. We anticipate that researchers who draw from an empirically derived list of outcomes grounded in a genetic counseling model of practice will employ measures that more fully capture the scope of genetic counseling practice. Such research will yield evidence-based outcomes that better represent the unique contribution of genetic counselors. Research grounded in practice is critical as the profession anticipates the needs for evidence-based practice and established outcomes to respond to the changing milieu of healthcare and reimbursement for services (Supreme Court of the United States 2013).

Researchers interested in a particular set of outcomes could review the presented list to find empirically based outcomes that are of interest and/or relevance to their study aims. They could use the listed outcomes as a basis for further operationalizing their chosen outcome(s) and, when possible, select established measures that reliably and validly assess their desired outcome.

The following hypothetical example illustrates the potential utility of the study outcomes in a research process: Envision a research team is interested in examining the patient-counselor relationship in genetic counseling. Specifically, these researchers have observed in clinic that genetic counselors and patients establish a connection that appears to be unique from the relationships that patients develop with other health care providers. The researchers want to more fully understand how this distinct relationship influences the outcomes of the genetic counseling session. Given the desired focus on the relationship in the genetic counseling process, the researchers could refer to the eighth goal of the REM "Counselor and patient establish a bond." Utilizing the associated outcomes list (see Table 4 "Support and Guidance"), the researcher may focus in on the outcome "The patient's emotional needs are met." Thus, their study may aim to examine how the genetic counselor and patient relationship influences the patient's perceptions that their emotional needs were met. The researchers may then further operationalize the counselor-patient relationship through a working alliance measure such as the Working Alliance Inventory (Horvath and Greenberg 1989). They may hypothesize that a stronger working alliance will predict patients' self-reports that their emotional needs were met by the genetic counselor. With an empirically based outcome and established outcome measure in hand, the researchers could then continue with additional study design and implementation. Thus, in this example, the researchers were able to utilize their clinical observation, along with the documented outcomes list to develop an initial study question and hypothesis.

This hypothetical example illustrates one of several ways the list of outcomes is potentially useful for researchers. First, the outcome list could be utilized to refine a research question. In the above example, the researcher's clinical observation was the impetus for their research question. The outcome list could also be used to generate research questions. For

Table 8 Genetic counseling outcomes identified in multiple REM factors and associated REM goals

Genetic counseling outcome	Factor 1: Understanding and appreciation						Factor 2: Support and guidance						Factor 3: Facilitative decision-making			Factor 4: Patient-centered education	
	1[a]	2[b]	3[c]	4[d]	5[e]	6[f]	7[g]	8[h]	9[i]	10[j]	11[k]	12[l]	13[m]	14[n]	15[o]	16[p]	17[q]
A change in communication regarding genetic information within the patient's family			X										X				
A change in patient's ability to ask appropriate questions regarding their medical care												X	X				
A change in patient's ability to engage in the genetic counseling process				X	X			X	X	X			X		X		X
A change in patient's ability to gain a new perspective										X		X					
A change in patient's adaptation				X							X	X		X			
A change in patient's adherence to health behaviors											X						X
A change in patient's adjustment																X	X
A change in patient's anxiety										X						X	
A change in patient's autonomy					X										X		
A change in patient's comfort level with their decision			X											X	X		X
A change in patient's coping												X				X	
A change in patient's distress					X		X				X						
A change in patient's empowerment			X														X
A change in patient's feeling of stigma										X						X	X
A change in patient's genetic knowledge						X	X	X			X	X	X	X		X	
A change in patient's guilt										X				X			
A change in patient's locus of control		X					X										
A change in patient's medical follow up							X										X
A change in patient's medical management			X			X	X				X	X		X		X	X
A change in patient's perceived power of control		X										X		X	X		
A change in patient's perception that they are supported															X		X
A change in patient's perception that they receive empathy						X					X						X
A change in patient's self-esteem		X				X											
A change in patient's understanding of implications of genetic information															X	X	

Table 8 (continued)

Genetic counseling outcome	Factor 1: Understanding and appreciation						Factor 2: Support and guidance						Factor 3: Facilitative decision-making			Factor 4: Patient-centered education	
	1[a]	2[b]	3[c]	4[d]	5[e]	6[f]	7[g]	8[h]	9[i]	10[j]	11[k]	12[l]	13[m]	14[n]	15[o]	16[p]	17[q]
A change in patient's level of regret regarding their decision			X	X		X									X		
A change in the information exchanged between the genetic counselor and patient																X	X
A collaborative conversation between the patient and genetic counselor															X	X	
A patient is empowered		X		X		X							X				
A patient is satisfied with genetic counseling service				X		X	X				X			X			X
A patient perceives their decision to be consistent with their values										X				X			
A patient receives care that is congruent with their needs	X													X			
A patient receives competent care				X			X										
A patient receives relevent information				X										X			
A patient's decision making is autonomous				X										X	X		
A patient's emotional needs are addressed								X									X
A patient's informational needs are addressed								X									X
A patient's informational needs are met	X																X
A patient's informed decision making				X						X							
A patient's medical needs are addressed								X									X
A trust is formed between the patient and genetic counselor								X									X
An alliance between the patient and genetic counselor is formed				X	X	X	X	X								X	X

REM Goals: [a] 1) Counselor and patient reach an understanding of patient's family dynamics and their effects on the patient's situation; [b] 2) Counselor promotes maintenance of or increase in patient self-esteem; [c] 3) Counselor facilitates the patient's feelings of empowerment; [d] 4) Counselor integrates the patient's familial and cultural context into the counseling relationship and decision-making; [e] 5) Counselor works with patient to recognize concerns that are triggering the patient's emotions; [f] 6) Counselor establishes a working contract with a patient; [g] 7) Counselor recognizes patient strengths;[i] 8) Counselor and patient establish a bond; [i] 9) Counselor's characteristics positively influence the process of relationship-building and communication between counselor and patient; [j] 10) Counselor helps the patient to gain new perspectives; [k] 11) Counselor helps patient to adapt to his or her situation; [l] 12) Counselor helps the patient to feel in control; [m] 13) Counselor helps the patient to feel informed; [n] 14) The counselor knows what information to impart to each patient; [o] 15) Counselor facilitates collaborative decisions with the patient; [p] 16) Counselor presents genetic information in way that the patient can understand; [q] 17) Good counselor-patient communication occurs

example, genetic counseling students could review the outcome list to find areas of interest and subsequently, generate research questions. Investigators with an already established research question could draw upon the outcomes list to identify ways to operationally define and measure their proposed question.

Second, while further work is needed to connect the outcomes identified in this study with established outcome measures, the list also provide researchers with a wider-range of outcomes in which to examine concepts such as decision-making or patient-centered education. And finally, researchers can use the outcomes list to connect specific *outcomes* with an aspect(s) of genetic counseling *process* they wish to study. For example, researchers interested in patient satisfaction may note that this outcome was named as an outcome for four REM goals (e.g., Goal 4: Counselor integrates the patient's familial and cultural context into the counseling relationship and decision-making; Goal 11: Counselor helps patient to adapt to his or her situation; Goal 14: The counselor knows what information to impart to each patient; and Goal 17: Good counselor-patient communication occurs). In designing their study, they would further refine what aspect of genetic counseling processes they wish to examine in regards to patient satisfaction. Thus, they may choose to focus their study on decision-making or communication, for example, in order to more precisely assess the impact of these processes on the outcome of patient satisfaction.

The Reciprocal-Engagement Model

While not a specific aim of this study, the genetic counseling outcomes derived from the focus groups further articulate the REM of genetic counseling practice. As its name implies, the REM emphasizes the complementary nature of each element in the model. In the present study, 41 identified outcomes were identified in multiple goals and four outcomes were seen in all four of the factors identified by Hartmann et al. (2015). Given that the majority of the REM goals were only discussed in one focus group, these findings are preliminary, but they lend support to McCarthy Veach et al.'s (2007) assertion that REM elements are reciprocal in nature (p. 724).

Participants identified a greater number of outcomes for certain REM goals. For example, REM Goal 6 (Counselor establishes a working contract with a patient) yielded 30 outcomes; whereas REM Goal 9 (Counselor's characteristics positively influence the process of relationship-building and communication between counselor and patient) yielded five outcomes. While these findings do not speak to the importance of specific goals per se, they may illustrate ways in which some goals and outcomes vary in priority, or the way in which they would be achieved given the setting or patient-specific characteristics. As Hartmann et al.'s (2015) validation study demonstrated, mean frequency ratings for the REM goals were lower than mean importance for every goal. Hartmann et al. (2015) speculated that some goals may be "difficult to achieve, and/or are not necessarily applicable to each patient" (p. 234). In addition, some outcomes, such as changes in a patient's psychological state or long-term coping, are more difficult to conceptualize and measure. This may have influenced the frequency in which they were noted by the focus group participants. Further research is needed to better understand reasons varying numbers of outcomes across the 17 goals.

Anticipated Challenges of Genetic Counseling Outcomes

In every focus groups, participants voiced opinions and concerns regarding perceived challenges in evaluating genetic counseling outcomes such that the results are reliable, valid, and comprehensive. Concerns ranged from a need to modify genetic counseling outcomes measures based on advances in genetic technology, to determining how best to measure outcomes based on specialty areas and stakeholder groups. Participants also raised concerns regarding the timing of evaluations, especially given the difficulty of capturing the effects of genetic counseling on patients by measuring outcomes at a single, discrete period of time. Participants stressed the longitudinal nature of genetic counseling outcomes such that "impact" may continue to occur days, weeks, or years after a genetic counseling session. These findings are consistent with prior authors who note similar challenges (Bernhardt et al. 2000; Clarke et al. 1996; Payne et al. 2008). Bernhardt et al. (2000) discuss challenges in measuring outcomes in genetic counseling given that different outcomes may vary based on time. They differentiate between short-term goals (e.g., client's sense of being supported) and long-term goals (e.g., improved communication about genetic issues within the family), and they discuss the need for further research measuring genetic counseling outcomes longitudinally.

In addition to timing challenges, the sheer number of genetic counseling outcomes identified in the present study indicates a single, exhaustive outcomes scale is neither feasible, nor desirable. Payne et al. (2008), in their review of validated outcomes measures for clinical genetic services, demonstrated the lack of a single validated measure that evaluates every aspect of clinical genetic services. McAllister et al.'s (2011) GCOS - 24 incorporates several important outcomes of a CGS including perceived personal control, emotional regulation, and hope. While these outcomes were similarly named in the current study, many others are not captured. Further discussion within the profession is needed to examine whether a goal of a single, comprehensive outcome measure of genetic counseling is even advantageous.

The current findings, along with prior work, speak to the challenges of evaluating genetic counseling outcomes with reliable and valid measures that capture the many nuances involved in genetic counseling (e.g., priority and timing of measurements, practice differences by specialty, patients' individual and cultural characteristics, priorities of stakeholders). These challenges need to be acknowledged and the complexities should perhaps be embraced as they represent what makes genetic counselors and genetic counseling unique. More extensive research on genetic counseling outcomes and in turn, development of valid appropriate outcomes measures are warranted in order to capture the breadth, depth, and nuance of the services that are provided.

Study Limitations

While focus group participants identified many genetic counseling outcomes, the sample represents a small number of practicing genetic counselors, outcomes researchers, and training directors. Thus, the outcomes they identified may not represent the opinions of the wider genetic counseling community. Moreover, the extent to which the outcomes represent the perspectives of stakeholders outside of the profession of genetic counseling is unknown. Certain limitations inherent to focus groups warrant acknowledgement (Krueger and Casey 2008). Other focus group members, as well as the moderators may have influenced participants' opinions and thoughts. An additional focus group was conducted following the NSGC meeting. While this focus group followed the same protocol, logistical differences (e.g., location, timing) may have influenced results. Given only four REM goals were discussed by more than one focus group (REM Goals 14–17), the identified outcomes for the other REM goals represent the perspective of a small number of participants. In addition, the REM was used to guide the focus group participants in their identification of genetic counseling outcomes. Participants were not asked to identify outcomes they believed are outside the 17 REM goals. Thus, the theory-driven process used in this study likely influenced the themes identified in the data analysis. Finally, respondent review of the data analysis would have been helpful in providing testimonial validity to the identified outcomes. Given these limitations, conclusions about the outcomes (i.e., their external validity) should be made with caution.

Future Research Recommendations

This focus group study and its' identified outcomes comprise one of many steps needed to empirically characterize and prioritize genetic counseling outcomes. In this study, the REM and Hartman et al.'s (2015) four factors provided a theoretical framework to categorize the identified genetic counseling outcomes. While this provided a helpful initial framework, the intricate way in which the genetic counseling outcomes overlap and intertwine suggest the need for a more sophisticated characterization process. Many of the identified outcomes represent stepping-stones to other outcomes, despite participants being instructed to define outcomes as "end-points" of a clinical intervention. Proctor et al. (2011) discusses these types of outcomes as "implementation outcomes" or outcomes defined as "the effects of deliberate and purposive actions to implement new treatments, practices, and services" with an important function of being "key intermediate outcomes" (p. 65). Consistent with this definition, many of the outcomes identified in the present study are implementation or process outcomes and are thus, distinct from service outcomes (e.g., safety, effectiveness) and client outcomes (e.g., satisfaction). Proctor et al.'s (2011) classification system is just one example of how to characterize outcomes; further research is needed to more systematically characterize the 194 outcomes identified in this study.

The present findings ultimately may be better categorized using an overarching structure based on an outcomes model. Pithara (2014), for example, discusses the use of composite outcomes measurement to gauge outcomes of clinical genetic services. Composite outcomes measures utilize a combined metric to capture many aspects of care including quality of care, overall and individual metrics, as well as a continual measurement across components of care or providers. Research should be done to investigate the utility of composite outcomes measures for capturing the varied genetic counseling outcomes among different stakeholders involved in genetic counseling services.

Further research is also needed to determine the full scope of genetic counseling outcomes. The present study focused on patient-centered outcomes, although some outcomes that occur outside of the session were identified as well. Moreover, anecdotal evidence from discussion at an Educational Breakout Session at the 2013 AEC suggests additional genetic counselor roles and the outcomes of their work extend beyond the session with patients (Zierhut, H, personal communication, October 12, 2013). Case preparation and follow-up management are just two examples of genetic counseling activities that inevitably impact patient outcomes. Further studies should be done to more comprehensively explore the many outcomes of pre- and post-session work and how these "out of session" outcomes may vary based on specialty setting (e.g., clinical, laboratory, public health).

Conclusion

The purpose of this study was to identify genetic counseling outcomes using the framework of the REM of genetic counseling practice. The wide-range of genetic counseling outcomes identified not only extend the current literature on genetic counseling outcomes, they also provide a starting point (a "database" per se) for future researchers who desire to conduct more focused genetic counseling outcome studies. The findings of this study further articulate the REM by providing additional and more specific outcomes stemming from each of the 17 REM goals. This study is the first step in a line of research intended to comprehensively identify genetic counseling outcomes. Such research ultimately will yield a corpus of empirical evidence to define what genetic counselors do, guide measurement of the effectiveness and utility of genetic counseling services, and promote evidence-based-practice.

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Human Studies and Informed Consent All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

Informed consent was obtained from all individual participants included in the study.

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

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