Conceptualizing genetic counseling as psychotherapy in the era of genomic medicine

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Abstract

Discussions about genetic contributions to medical illness have become increasingly commonplace. Physicians and other health-care providers in all quarters of medicine, from oncology to psychiatry, routinely field questions about the genetic basis of the medical conditions they treat. Communication about genetic testing and risk also enter into these conversations, as knowledge about genetics is increasingly expected of all medical specialists. Attendant to this evolving medical landscape is some uncertainty regarding the future of the genetic counseling profession, with the potential for both increases and decreases in demand for genetic counselors being possible outcomes. This emerging uncertainty provides the opportunity to explicitly conceptualize the potentially distinct value and contributions of the genetic counselor over and above education about genetics and risk that may be provided by other health professionals.

In this paper we suggest conceptualizing genetic counseling as a highly circumscribed form of psychotherapy in which effective communication of genetic information is a central therapeutic goal. While such an approach is by no means new—in 1979 Seymour Kessler explicitly described genetic counseling as a “kind of psychotherapeutic encounter,” an “interaction with a psychotherapeutic potential”—we expand on his view, and provide research evidence in support of our position. We review available evidence from process and outcome studies showing that genetic counseling is a therapeutic encounter that cannot be reduced to one where the counselor performs a simple “conduit for information” function, without losing effectiveness. We then discuss potential barriers that may have impeded greater uptake of a psychotherapeutic model of practice, and close by discussing implications for practice.

Keywords

Psychosocial; psychotherapy; therapeutic; genetic counseling; evidence based medicine

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Conflict of Interest Statement

Authors Austin, Semaka and Hadjipavlou declare that they have no conflict of interest.

Introduction

Physicians, nurses, and other health-care providers in all quarters of medicine, from oncology to psychiatry, are increasingly being expected to routinely field questions about the genetic basis of the medical conditions they treat (Guttmacher *et al* 2007). Communication about genetic testing and risk also enter into these conversations, as knowledge about genetics is increasingly expected of all medical specialists. To cater to this need, undergraduate curricula in medicine are increasing their emphasis on genetics (Salari *et al* 2013) and organizations offering continuing education for healthcare providers are providing ever more sophisticated and detailed resources (NCHPEG.org, Geneticseducation.nhs.uk). We suggest that attendant to this evolving medical landscape is some uncertainty regarding the future of the genetic counseling profession. It is possible, for example, that demand for genetic counselors could increase as a result of more frequent identification of rare genetic syndromes through more widespread implementation of genetic testing, as well as the discovery of the genetic contributions to common multifactorial diseases. On the other hand, it is possible that demand for genetic counselors could decrease, as other healthcare professionals may feel increasingly required and equipped to address the topics that would have previously been the domain of genetic counselors. This uncertainty presents the opportunity to explicitly conceptualize the distinct value and contributions of the genetic counselor over and above the provision of education about genetics and risk that may increasingly fall within the purview of other health professionals.

Genetic counseling: an activity and a profession

In considering the place of genetic counseling in the context of this evolving medical landscape, it is instructive to first explicitly acknowledge that genetic counseling can be conceptualized as both an activity and as a profession. The term “genetic counseling” arose in the 1940s, and is attributed to Sheldon Reed (Resta 1997). Reed worked with parents of children with intellectual disability, and although he was much appreciated by families for his interpersonal skills, his “compassion never left the confines of medical paternalism” (Stern 2012, p88). While he conceptualized the “genetic counseling” he engaged in as “a kind of genetic social work without the eugenic connotations,” (Reed 1975, p335) there is no direct connection between Sheldon Reed and the establishment or development of the professional discipline of genetic counseling (Resta 1997).

The first training program that established genetic counseling as a stand-alone professional discipline in its own right, rather than simply an activity that geneticists or other healthcare providers might engage in, was not established until 1969. The development of the curriculum for this program was informed not just by the rapidly accumulating medical knowledge about genetics, but also by clinical psychology—and in particular, Carl Roger’s humanist client-centered approach to psychotherapy (Veatch *et al*, 2007). In drawing from both medicine and psychotherapy, the new *profession* of genetic counseling adopted a distinct stance that diverged from the exclusively medical model origins of the *activity* of genetic counseling as conceived by Reed. But, despite this, the professional discipline of genetic counseling has had an uneasy relationship with psychotherapy. While the profession has readily identified important aspects of the genetic counseling encounter as

“psychosocial” in nature, there has been considerable reluctance to apply the term “psychotherapeutic” to genetic counseling practice, or to align the profession explicitly with psychotherapy. However, the question of “How psychotherapeutic should genetic counseling be?” (Fraser 1982, p367) has recurred sporadically in the literature for several decades (Targum 1981, Kessler *et al* 1984, Kessler 1987, Eunpu 1997, Biesecker 2001).

Defining psychotherapy and its relationship to the discipline of genetic counseling

According to the American Psychological Association (APA), psychotherapy “is the informed and intentional application of clinical methods and interpersonal stances derived from established psychological principles for the purpose of assisting people to modify their behaviors, cognitions, emotions, and/or other personal characteristics in directions that the participants deem desirable” (Norcross 1990, p218). Psychotherapy is provided by a trained healthcare provider and encompasses a diverse array of practices “designed varyingly to provide symptom relief and personality change, reduce future symptomatic episodes, enhance quality of life, promote adaptive functioning in work/school and relationships” and “increase the likelihood of making healthy life choices” (APA 2013, p102). Vastly dissimilar therapeutic approaches such as behavioral therapy for spider phobia delivered over a single session, several years of psychoanalysis to enhance self-understanding, or supportive therapy to cope adaptively with grief, all fall under the rubric of psychotherapy. At its core, psychotherapy is about a helping relationship in which one person has the knowledge and skills relevant to helping another person address a problem through conversation. We suggest that the APA’s working definition of psychotherapy is readily compatible with genetic counseling, which has been defined as the process of helping people to “understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease” (Resta *et al* 2006, p77). Thus, given the typically short-term nature of the counselor client relationship and the specific focus of the interactions, one could consider genetic counseling to be a time limited, highly circumscribed form of psychotherapy.

Psychotherapeutic elements of genetic counseling have been recognized since the inception of the profession, as has its potential to evoke feelings of shame and guilt, especially when psychological concerns are not addressed (Kessler *et al* 1984). The foundational nature of Rogerian, client-centered psychotherapy skills to the practice of genetic counseling is reflected in their identification as core competencies by the Accreditation Council for Genetic Counseling (ACGC, 2013a). The following psychotherapeutic skills are explicitly included in these competencies: the provision of short-term, client centered counseling and psychological support, empathic listening and crisis intervention skills, as well as demonstrated knowledge of psychological defenses, family dynamics, coping models, the process of grief, and reactions to illness. Accordingly, in order to obtain and maintain their accreditation through the ACGC, genetic counseling training programs are required to reflect these competencies in their curricula (ACGC 2013b).

Teaching and counseling models in genetic counseling: a review of the outcomes literature

Seymour Kessler, who has written extensively on psychological aspects of genetic counseling, delineated two contrasting models of practice—the “teaching model,” characterized by a predominantly didactic stance, and the “counseling model,” characterized by a therapeutic stance in which attention to contextual and psychological dimensions is prioritized alongside the provision of genetic information (Kessler 1997). Although in theory these two models are best conceptualized as representing opposite poles on a continuum of practice rather than discrete or dichotomous categories—with genetic counselors situated along a spectrum of teaching and counseling with those in the middle implementing more hybrid approaches—empirical research suggests otherwise.

In a systematic review of empirical studies of the content and process of genetic counseling, Meiser et al. (2008) found that the majority of genetic counselors practice from a teaching model, which emphasizes the didactic provision of biomedical information with relatively little attention to clients’ psychosocial concerns. Their conclusion is consistent with results from the largest study to date specifically designed to examine how genetic counselors conduct pre-test counseling sessions (Roter *et al* 2006), and of a large, recent study of how genetic counselors communicate about risk in the context of Alzheimer’s Disease (Lerner *et al* 2013). And yet this pervasive practice of genetic counseling is very much at odds with the Reciprocal Engagement Model (REM), a model of genetic counseling practice that was developed through consensus of 23 genetic counseling program directors from across North America (Veach *et al* 2007). Although genetic information is recognized as a key element in REM, so too are patient emotions, and at the heart of REM is the tenet that the counseling relationship “is integral to genetic counseling” (Veach *et al* 2007).

The pervasive teaching model of genetic counseling is also at odds with a growing body of evidence from outcome studies of genetic counseling, which indicate that attending to psychological dimensions of practice, such as the facilitation of understanding, empathic responses and lower levels of verbal dominance, are associated with more positive client outcomes (Meiser *et al* 2008). Although most studies investigating the outcomes of genetic counseling have focused on clients’ recall of information, accuracy of risk perception, and knowledge and understanding of medical concepts conveyed (Kessler & Antley 1981; Meiser *et al* 2008), the most important and impactful outcomes of genetic counseling from clients’ perspectives are often of a psychosocial nature—for example, healthy adjustment, empowerment, behavioral change, and satisfaction with decision-related outcomes (Edwards *et al* 2008). These outcomes are associated with a counseling model approach to genetic counseling. Specifically, research demonstrates that clients tend to express less concern and experience reduced anxiety when their expectations for reassurance and advice are met in the genetic counseling session (Meiser *et al* 2008; Michie *et al* 1997). In contrast, meeting clients’ expectations for information has not been associated with improved emotional outcomes. Higher counselor verbal dominance is also associated with higher anxiety levels and lower perceived needs fulfillment, highlighting the importance of client engagement and collaboration in the genetic counseling session (Dijkstra *et al* 2013; Meiser *et al* 2008; Roter

et al 2006). Clients also tend to have greater improvements in depression levels when counselors use more empathic responses (Duric 2003; Meiser *et al* 2008).

Clients consistently rate higher satisfaction with practice models that emphasize counseling over teaching (Roter *et al* 2006). Further, it is also worth highlighting that—perhaps counter-intuitively—counseling oriented practice also positively influences knowledge-based outcomes. Encouraging clients to articulate their feelings seems to facilitate their cognitive processing of the complex medical information provided (Ellington *et al* 2011; Meiser *et al* 2008; Veach *et al* 2007). In a systematic review of interventions to improve risk communication in clinical genetics, Edwards et al. (2008) found that supportive or emotional elements of counseling, such as addressing issues of loss, grief and relationship problems, provided greater benefits (including in areas such as knowledge and decision making) than purely informational or educational approaches.

Collectively, these data suggest that meeting the emotional needs of clients is strongly associated with better psychological results, which in turn can support knowledge-based outcomes. In other words, relational aspects of genetic counseling potentially impact outcome—even if that outcome is narrowly defined in terms of the successful provision of information. These data from process and outcome studies of genetic counseling dovetail with findings from the broader psychotherapy research literature.

Recent comprehensive meta-analyses of hundreds of controlled studies have demonstrated that several facets of the therapy relationship have a robust and significant impact on therapy outcomes across a wide range of approaches, clinical contexts, clients, and problems (Norcross & Wampold 2011). These evidence-based facets—or so-called “common factors”—include the appropriate expression of empathy, establishing an effective therapeutic alliance, collaboration, goal consensus, and positive regard, and they account for most of the explained variance in therapy outcomes (Norcross & Wampold 2011). These are effective elements of *any* therapeutic relationship and overlap directly with the principles of Rogerian client-centered therapy, which inform the practice-based competencies for genetic counselors and standards for accreditation of genetic counseling programs, listed above. We suggest that the extensive psychotherapy literature demonstrating that the therapeutic relationship has a significant influence on outcomes is directly relevant to genetic counseling. Clearly, a therapeutic approach to genetic counseling such as that implied in the model of practice described by the REM, which views the counselor-patient relationship as integral to genetic counseling, is aligned with the empirical research on evidence-based relationships, while a teaching model of genetic counseling, which implicitly eschews the importance of the counselor-patient relationship, is not.

Barriers to psychotherapeutically focused genetic counseling

While no studies have systematically assessed barriers to adopting a therapeutic model of practice in genetic counseling, several potential obstacles have been identified (Biesecker 2003; Eunpu 1997; Hartmann *et al* 2013). One view is that genetic counselors might feel comfortable providing an education-based intervention (Hartmann *et al* 2013; Kessler, 1997). This suggests that there might be a gap within training programs regarding the

development of adequate counseling ability (Biesecker 2003; Weil 2003). However, this potential explanation appears to be at odds with other data. Specifically, therapeutic skills are required core competencies to graduate from a genetic counseling program (ACGC 2013a; ACGC 2013b; Eunpu 1997; Resta *et al* 2006), and program accreditation relates to how well these competencies are addressed by the curriculum. Thus, at least according to the ACGC, the curricula of accredited programs are addressing the core competencies relating to therapeutic skills adequately.

Further exploration suggests that any lack of comfort with therapeutic skills among genetic counselors might arise from discrepancies between how genetic counseling is taught versus how it is practiced. Specifically, directors of genetic counseling programs have noted that while the curriculum emphasizes psychosocial counseling skills, in clinical rotations students are often instructed by their supervisors to focus on the provision of medical information (Biesecker 2003; Veach *et al* 2007; Weil 2003). This lack of emphasis on therapeutic skills in clinical practice is consistent with a survey of 1346 members of the National Society of Genetic Counselors (which included students) in which only 5.9% identified an interest in psychotherapy (Eunpu 1997).

The reason for this apparent divergence between training and practice has not been empirically investigated. However, there is some suggestion that it may be less a result of deficiencies of training programs and more about the constraining reality of how genetic counselors are typically situated within medical institutions (Biesecker 2003; Weil 2003). In this context, genetic counseling is implicitly conceptualized as a medical activity whose primary objective is to convey medical information about genetic diagnosis and its prognosis, treatments, recurrence risks, and reproductive options (Biesecker 2003; Weil 2003).

Further, genetic counselors often have a heavy caseload, and are routinely expected to limit sessions to a maximum of one hour, if not less. Given that genetic information can be quite extensive, it is not uncommon for this task to overwhelm the session, and diminish time available for more psychotherapeutic work (Hartmann *et al* 2013). Consequently, the primary focus of counseling sessions is not counseling, but information. Biesecker (2003) summarizes this well:

“It is difficult to diverge from the practice of medical genetics sufficiently to exercise our skills as attendees to the loss, hurt, frustration, anger, indecision, and disappointment that our clients experience. Yet it is both the cognitive and the affective meaning of genetic information and their lived experiences that are most relevant to genetic counseling clients. A focus on information may be less about our abilities (training), values, or attitudes and more about priorities in health care delivery and our status as counseling colleagues in the medical genetics team.”

(Biesecker 2003, p215)

In addition to sociological explanations in which the wider adoption of a therapeutic model of genetic counseling is hindered by outside forces such as institutional pressures, some writers have identified limiting forces that are internal to the profession. For example, Weil (2003) has suggested that the guiding principle of “non-directiveness,” once considered the

“central ethos” of genetic counseling, has hampered the development of a more psychotherapeutically oriented approach (Weil 2003). Historically, the concept of “non-directiveness” (in the sense of not directing clients to terminate pregnancies affected by genetic syndromes, for example) was instrumental in distancing the contemporary genetic counseling profession from the directive or eugenic medical practices of the past (Stern 2012). However, “non-directiveness” has also cast a constraining shadow, as it has persistently been interpreted as a stance that limits the genetic counselor to the role of information provider (see Stern 2012 for an excellent review).

Despite the fact that in recent years the promotion of client autonomy (a stance that is readily compatible with more therapeutic models of practice) has gradually gained primacy and psychotherapeutic goals have been included in the definition of genetic counseling, other writers have described a sense of unease or hesitation about “claiming the psychotherapeutic role as part of their professional self-definition” (Eunpu 1997). This is perhaps related to a tendency to eschew the terms “psychotherapy” or “psychotherapeutic” in favor of the vague—if not euphemistic—term “psychosocial” to refer to psychotherapeutic dimensions of genetic counseling practice. At least part of the hesitation to use the term “psychotherapy” seems to stem from misconceptions about psychotherapy and its relevance to genetic counseling; for instance, that “psychotherapy” requires the presence of psychopathology in the client, a long-term therapeutic relationship, and/or more extensive training than is typically covered in genetic counseling curricula.

In addition, earlier writings on psychological aspects of genetic counseling drew on the psychoanalytic paradigm that was dominant at the time. For example, some authors viewed genetic counseling as an “opportunity to divulge emotional stresses permeating the family milieu through a catharsis which circumvents such defense mechanisms as projection, rationalization, self-condemnation, repressions, and feelings of misdirected guilt and hostility” (Tiers 1964, p113 quoted in Resta 2006), while others argued for the “psychodynamic objective” of helping clients resolve unconscious conflicts that interfered with their capacity to absorb and engage with genetic information (Targum 1981). While this is a laudable goal, it is not likely compatible with the scope of current practice and the therapeutic training of most genetic counselors. Although genetic counselors with psychodynamic training may be inclined to understand clients in psychodynamic terms (e.g., unconscious conflicts)—and such understandings may prove fruitful in facilitating the effective communication of risk and other genetic information—genetic counseling can be psychotherapeutic without being psychodynamic.

Similarly, psychotherapy is sometimes conflated with psychoanalysis or long-term “personality reconstruction,” and invoked as a way to provide contrast for the counseling activities of genetic counselors (Resta 2006). This situation is quite misleading, and elides the clear overlap between genetic counseling and its client-centered therapy roots—which are evident in its core competencies—and the goals and skills of supportive psychotherapy.

Using existing data to inform clinical practice and research strategy for genetic counseling

The data currently available suggest that if we aim to conduct genetic counseling in a manner that promotes the best psychosocial outcomes for clients, that is, therapeutically, we must attend to relationship factors that have been shown to be effective in promoting good outcomes across psychotherapies and therapeutic relationships. These evidence-based elements include establishing an effective therapeutic alliance, appropriate expression of empathy, collaboration, positive regard, and shared goal setting (Norcross & Wampold 2011). Although more research is needed to explore the process and outcomes of genetic counseling in general, as well as the relationship between psychological and knowledge-based outcomes more specifically, available data suggest that these common therapeutic factors are as critical to the outcomes of genetic counseling as they are to other forms of psychotherapy.

Recently, McCarthy Veach and colleagues (2007) have elaborated a psychotherapeutic approach to genetic counseling practice—the “reciprocal-engagement model”—that explicitly recognizes the importance of these factors.

“Genetic counseling therefore is a relationally based helping activity whose outcomes are only as good as the connection established between the counselor and patient. In other words, the quality of the genetic counselor-patient relationship is as important to genetic counseling outcomes as the genetic information provided.”

(Veach et al 2007 p721)

We fully agree with this approach and would anticipate that the absence of such therapeutic factors would impede the effectiveness of genetic counseling, even when narrowly restricted to the accurate recall of information.

In sum, we suggest that trends in the emerging literature indicate that as a profession we are increasing our emphasis on examining the psychosocial outcomes for clients who receive our services, with the aim of optimizing these outcomes. The available evidence (that we have summarized above) suggests that as we engage in this important work, we need to find ways to attend to the application of psychotherapeutic skills in our clinical encounters with clients rather than focusing on applying a strictly educational model. Even in circumstances where we seek to achieve improvements in outcomes related to information retention, we suggest that use of psychotherapeutic techniques may allow clients to achieve an emotional state that is more conducive to digesting important medical information. Further, it is these therapeutic elements of practice that stand to distinguish genetic counseling as provided by genetic counselors from genetic counseling that can be provided by other healthcare professionals. As stated by Resta (2006), the:

“focus on psychological issues is what genetic counselors regard as the key issue that separates them, professionally and clinically, from other health professionals who provide genetic counseling.”

(Resta 2006 p271).

The future?

Embracing a psychotherapeutic orientation may foster greater professional development and skill acquisition. For instance, resolving anxiety inherent to genetic uncertainty is often a chief motivation for why clients seek the help of a genetic counselor (Van Zuuren & Van Schie, 1997). An overly simplistic view of this would be that providing genetic information reduces uncertainty, and by extension, anxiety. The empirical evidence we reviewed suggests otherwise. Not only is uncertainty often not so easily alleviated but new information, especially concerning risk and probability, may actually introduce new uncertainties. Helping clients adapt to these new uncertainties, especially when they emerge *after* receiving genetic information, is one psychotherapeutic dimension of genetic counseling—but only if it is identified as such. Genetic counselors who recognize this will be more likely to develop effective skills to manage these scenarios. Some of these skills can be borrowed from other psychotherapies, such as cognitive behavior therapy (CBT), which has developed techniques specifically for coping adaptively with uncertainty and associated anxiety. For example, Broley (2013) recently described the successful application of Acceptance and Commitment Therapy (ACT) techniques to help the mother of a child with 22q11 deletion alleviate intrusive, recurrent worry and cope adaptively with the troubling uncertainty inherent in her child's condition. As complex/multifactorial conditions (to which considerable uncertainty is inherent) increasingly enter the orbit of genetic counseling, and as genomic technologies become more widespread, more and more variants of uncertain significance will be identified. We suggest that as the degree of certainty in the information that can be provided decreases, psychotherapeutic approaches may become increasingly important to the practice of genetic counseling.

Conclusion

As genetics becomes integrated into all aspects of healthcare, physicians and healthcare providers will be increasingly expected to become competent in educating their clients about genetic testing relevant to their areas of practice and interpreting the clinical significance of genetic tests results. Yet while knowledge about genetics becomes more common, the skillset of genetic counselors, which integrates expertise in genetics with training in counseling, remains unique and important.

In this paper, we have proposed conceptualizing genetic counseling as a time-limited, highly circumscribed psychotherapeutic encounter—a psychoeducational process focused on the communication of genetic information that is embedded within a therapeutic relationship. Available research on the process and outcome of genetic counseling supports such a psychotherapeutic model of practice in which educational and psychosocial goals are indivisible. Indeed, such a psychotherapeutic approach not only stands to enhance client outcomes but also distinguishes genetic counselors from other healthcare professionals who provide education about genetics.

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