
Professional Issues

A New Definition of Genetic Counseling: National Society of Genetic Counselors' Task Force Report

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The Genetic Counseling Definition Task Force of the National Society of Genetic Counselors (NSGC) developed the following definition of genetic counseling that was approved by the NSGC Board of Directors:

Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:

- *Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.*
- *Education about inheritance, testing, management, prevention, resources and research.*
- *Counseling to promote informed choices and adaptation to the risk or condition.*

The definition was approved after a peer review process with input from the NSGC membership, genetic professional organizations, the NSGC legal counsel, and leaders of several national genetic advocacy groups.

KEY WORDS: genetic counseling; genetic counseling definition; genetic counseling history; National Society of Genetic Counselors.

INTRODUCTION

In 2003 the [National Society of Genetic Counselors \(NSGC\)](#) appointed a task force that was

charged with assessing the need for a new definition of genetic counseling. The creation of the Task Force was spurred by the advent of genomic medicine and the need to maintain a common practice base to genetic counseling as it expands beyond traditional settings into laboratory genetic medicine, public health, social and behavioral research, and common disease subspecialties (Bennett *et al.*, 2003; Ciarleglio *et al.*, 2003).

The authors constitute the NSGC's ad hoc task force on the Genetic Counseling Definition, herein referred to as the Task Force. Robin Bennett, then NSGC President, convened the task force. The composition of the Task Force reflected a wide range of

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specialization and experience. An Expert Panel comprised of Luba Djurdjinovic, M.S., Seymour Kessler, Ph.D., Wendy Uhlmann, M.S., Sharon Terry, M.A., and Jon Weil, Ph.D. advised the Task Force.

The charge assigned the Task Force was to define “genetic counseling,” rather than the profession of “genetic counselors.” This is an important distinction. Genetic counselors have many roles such as designing and conducting research, managing clinical cases, supervising trainees, teaching, making health care policy, and developing and implementing educational tools. Although many of these activities relate directly to genetic counseling, they are distinct from the counseling process. Simultaneous to our endeavor, NSGC appointed a task force to develop a scope of practice document that would capture many of these activities. Therefore, rather than defining what genetic counselors *do*, we were charged with describing what genetic counseling *is*, i.e., the essence of the relationship between the client and counselor when they interact. In defining genetic counseling practice, we do not stipulate who is qualified to provide it. This is the purview of the [American Board of Genetic Counseling](http://www.abgc.net) (www.abgc.net) and other professional organizations. However one of our aims was to create a definition that genetic counselors could use to convey their practice to other health care providers, legislators, health insurance executives and hospital administrators.

The multi-faceted purpose of this effort to define genetic counseling includes the development of common language in describing an important dimension of genomic medicine. Definitions promote understanding and awareness, and help to operationalize practice for research purposes. A definition also clarifies goals as they dictate practice (for further discussion see Biesecker 2001). The process of evaluating the need for and developing a new genetic counseling definition are described in this paper.

METHODS

Review of Existing Definitions

The Task Force first reviewed existing practice definitions (see Appendix). We searched MEDLINE for definitions from the mid-1960’s through 2004 via PubMed using the terms “genetic counseling,” “genetic counseling definition,” “genetic counseling history,” and “genetic counselor.” We also reviewed genetic counseling and medical genetics textbooks, along with bibliographies of relevant publications

since 1948. Colleagues, genetic counseling websites and professional pamphlets were also consulted. Several of the definitions were from other countries (Revel, 1995).

More than 20 published definitions were identified, the earliest being the 1948 article by Sheldon Reed where he coined the term. (For a more detailed review of the history of definitions of genetic counseling, see [Resta in press](#)). The 1975 definition adopted by the American Society of Human Genetics is the only definition created by a professional committee and is the most commonly cited in the literature. All other definitions were academic or descriptive and authored by leaders in the profession or anonymous sources, as in the case of pamphlets or brochures. The NSGC website and pamphlets describe what genetic counselors *do*, but prior to now the organization had no practice definition.

While the ASHG definition still captures important aspects of genetic counseling (Walker, 1998), it is cumbersome and lacks reference to the counseling elements (Biesecker and Peters, 2001). Other definitions were judged to be too short (Kessler, 1979; Reed, 1974; Resta, 2006), incomplete (Hsia and Hirschorn, 1979), focused primarily on disease prevention (Biesecker, 2001; Epstein, 1973; Harper, 1998) or particularly academic or overly technical for broad use (Bringle and Antley, 1980; Biesecker and Peters, 2001). Based on this review, the Task Force concluded that there was a need for a new definition.

Composing and Vetting a New Definition

The Task Force began considering a new definition by identifying target audiences and applications. These included marketing the profession, increasing public and professional awareness, crafting licensure legislation, developing practice guidelines, providing a basis for research into the counseling process and outcomes, and developing educational materials.

Care was taken to avoid scientific or technical jargon and to create a definition that was broad enough to apply to many settings. In particular, the phrase “genetic contributions to disease” was chosen to embrace the growing practice of genetic counseling for common or complex diseases. The Task Force developed and revised many versions of the definition. The Expert Panel reviewed two drafts and appropriate revisions were made.

NSGC membership comments were elicited via the NSGC listserv. The leadership of other

professional genetics societies in the United States and Canada reviewed a revised version. We gathered input from consumers who lead advocacy organizations who in turn requested comments from their own constituency organizations. After each solicitation, revisions were made to the definition by the chairs of the Task Force. The NSGC legal counsel and marketing consultant firm also both approved the final definition. In July 2005, the NSGC Board of Directors adopted the definition.

DEFINITION

The NSGC definition of genetic counseling states that genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of the genetic contributions to disease. This process integrates:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about inheritance, testing, management, prevention, resources and research.
- Counseling to promote informed choices and adaptation to the risk or condition.

The full definition may be most useful for teaching and research purposes. Alternatively, it can be shortened to only the first sentence. We anticipate that the shorter version will be most useful for marketing, policy, legislation and educational materials.

DISCUSSION

NSGC's leadership recognizes the importance of genetic counseling in meeting healthcare demands imposed by the emerging era of genomic medicine. In fostering the development of a contemporary practice definition, NSGC is taking one of many steps to facilitate genetic counselors' positioning at the forefront of this rapidly expanding field. The definition will aid unification of the field as it expands into primary care by clarifying what genetic counseling involves. The specialization of genetic counseling into cardiology, psychiatry, neurology, oncology, ophthalmology and other arenas, could lead to fractionation. Identifying the core aspects of genetic counseling practice common to all settings can offset the centrifugal effect of specialization.

A key role for genetic counselors is the education of other health care professionals about genetics

(Collins and Guttmacher, 2001). A clear practice definition is an important component of this endeavor. The delineation of genetic counseling complements efforts to determine roles of a variety of providers in the care of clients/patients (Guttmacher *et al.*, 2001).

The NSGC definition is not based on research data, but rather on clinical descriptions and experience (Biesecker and Peters, 2001). How accurately it reflects clinical practice is an empirical question. Research will help to discern whether all three elements—risk assessment, education and counseling—routinely occur and in what proportion (e.g., Roter *et al.*, *in press*). Without the benefit of process research, this definition might be considered a standard for genetic counseling practice, what it should or can entail. Some studies have found great variability in genetic counseling sessions (Matloff, 1994), suggesting that the definition broadly describes what may happen in practice and not necessarily what routinely happens.

LIMITATIONS

An undertaking by a committee to define a broad professional practice invariably involves compromise. The outcome becomes distilled down to a product that provokes the least amount of controversy and speaks to the greatest number of people. One area of controversy in developing this definition, for example, was the extent to which it would address ethical practice issues. Eventually the Task Force chose to eliminate reference to ethical issues, justified by the conviction that genetic counseling practice should be ethical. Ethical guidelines for the profession are addressed in the NSGC Code of Ethics (www.nsgc.org).

A second concern was critique of the word "process." One reviewer made a compelling case for use of the word "relationship" rather than process to draw attention to the importance of the connection between the counselor and client and the investment in the endeavor. However other reviewers were uncomfortable with use of relationship as a descriptor.

We anticipate that genetic counselors conducting research, managing the interpretation of laboratory tests, marketing testing services, or working on educational or policy projects may find the definition does not represent their current work. This suggests that genetic counseling is not a key component

of their current job responsibilities that draw on other professional skills of genetic counselors. Genetic counselors working in these and other capacities are encouraged to find their roles and responsibilities represented in the scope of practice document currently drafted by the NSGC (www.nsgc.org).

CONCLUSION

Practice definitions are not created in a social vacuum but rather reflect larger social trends, the socioeconomic background of genetic counselors, advances in medical technology and scientific knowledge, changing ethical values, and the motivation behind formulating a given practice definition (Resta, *in press*). In this rapidly evolving area, the NSGC definition forms the basis for assessing the field of genetic counseling for its relevance, its usefulness in counseling practice, and its value to society.

Appendix

#1: Genetic counseling is the process whereby an individual or family obtains information about a real or possible genetic problem.

—Hsia and Hirschorn (1979)

#2: Genetic counseling is the process of evaluating family history and medical records, ordering genetic tests, evaluating the results of this investigation, and helping parents understand and reach decisions about what to do next.

—Kids Health

#3: It (cancer genetic counseling) has been described as a communication process, which deals with human problems associated with the occurrence, or the risk of occurrence of a genetically inherited cancer in the family.

—Agashe and Shrikhande (2001)

#4: Genetic counselors work as members of a healthcare team, providing information and support to families who have members with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions. They identify families at risk, investigate the problem present in the family, interpret information about the disorder, ana-

lyze inheritance patterns and risks of recurrence, and review available options with the family.

—Human Genome Project Information

#5: An informative and supportive dialog regarding a known, potential or unsuspected genetic condition:

Understand the concerns and questions raised by the family and/or referring physician.

Acquire information: pedigree, medical history, experiential background (the personal burden), obtain relevant medical records.

Work as part of a multidisciplinary team to clarify the accuracy of the diagnosis (physical exam, relevant tests and evaluations).

If the diagnosis is known, facilitate the family's understanding and adjustment, emotionally and practically. Information is provided to help families make informed decisions and to access appropriate issues and decision-making in light of a degree of uncertainty.

If the diagnosis remains uncertain, facilitate understanding of what is known and what has been ruled out—facilitate management issues and decision-making in light of a known degree of uncertainty.

—Access Excellence

#6: A kind of genetic social work.

—Reed (1947)

#7: Genetic counseling is about helping people try to understand and cope with the effects of genetic disease on their lives and the lives of their families.

—Resta (2000)

#8: Genetic counseling is a communication process which deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family. This process usually involves an attempt by one or more appropriately trained persons to help the individual or family to:

- comprehend the medical facts including the diagnosis, probable course of the disorder, and the available management
- appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives
- understand the alternatives for dealing with the risk of recurrence
- choose a course of action which seems to them appropriate in view of their risk, their

family goals, and their ethical and religious standards and act in accordance with that decision, and to

- make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

—American Society of Human Genetics (1975)

#9: Genetic counseling is the process of providing information about the risk of occurrence and recurrence of genetic disease and, when appropriate, of taking steps to modify these risks.

—Epstein (1973)

#10: Genetic counseling is the process by which patients or relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it and of the ways in which this may be prevented, avoided or ameliorated.

—Harper (1998)

#11: An educational process that seeks to assist affected and/or at risk individuals to understand the nature of the genetic disorder, its transmission and the options open to them in management and family planning.

—Kelly (1986)

#12: Genetic counseling is a dynamic psycho-educational process centered on genetic information. Within a therapeutic relationship established between providers and clients, clients are helped to personalize technical and probabilistic genetic information and to enhance their ability to adapt over time. The goal is to facilitate clients' ability to use genetic information in a personally meaningful way that minimizes psychological distress and increases personal control.

—Biesecker and Peters (2001)

#13: Genetic counseling I take to include the following kinds of activity:

- advising adults, preconception, of the probability of their conceiving a child suffering from a genetic disorder
- advising adults, post-conception, and as a result of some method of fetal screening, as to whether or not a fetus is suffering from a genetic disorder

- alerting them to the options open to them.

—Chadwick (1993)

#14: Genetic counseling can entail any of the following activities:

- The achievement of an early, precise diagnosis of the condition causing concern, where possible, and hence an increase in knowledge and understanding. Even if unsuccessful, the attempt to achieve a precise diagnosis of the cause of a child's handicap can itself be therapeutic for the family.
- The screening for complications of genetic disease, thereby assisting in the management of affected individuals.
- The provision of social and practical support for those individuals and families with genetic disease: the affected, those who might develop the disorder in the future, and those whose children have been, already are or might in the future be, affected.
- The development and application of specific therapies for genetic disorders.
- The reduction of "handicap," itself a social construct, by working to minimize the stigma associated with disability and handicap, hoping to develop the self-esteem of affected individuals.
- The provision of information about future reproductive risks and options in particular family situations.

—Clarke (1993)

#15: A communication process which involves diagnosis, explanations and options (as in all medical consultation). In genetic counseling there is a stronger need for detail, especially in the explanations and options, for which empathetic and emotional support are an essential part. Counsellors are involved in the ethics of the "people's right to know."

—United States definition, cited in International Bioethics Committee of UNESCO (1995)

#16: Counselling entails precision of diagnosis, the estimation of risks, and a supportive role to ensure that those who are given information are

enabled to benefit from it and from the interventions that are available.

—United Kingdom definition, cited in International Bioethics Committee of UNESCO (1995)

#17: The objective, methods and indications of genetic consultation are:

Objective:

to provide information to patients (and/or blood relations of a patient) at risk of contracting a disease that may be hereditary on:
consequences of pathology in question
probability of contacting and transmitting it
possibility of keeping it in check and treating it

Methods:

construction and analysis of a pedigree
calculation of the risk of recurrence
estimation of the consanguinity coefficient
more specific analysis

When is counseling indicated:

known or presumed illness in the patient or family
congenital malformation
mental retardation
consanguinity
recurrent miscarriage, infertility

—Italian definition, cited in International Bioethics Committee of UNESCO (1995)

#18: A medical process of communication between a physician and a consultand (counselee) where scientific knowledge, data and facts are exchanged in order to provide a framework to understand the genetic problem of the patient and the family.

—Chilean definition, cited in International Bioethics Committee of UNESCO (1995)

#19: Better called “genetic advising”—a useful tool in preventive medicine.

—Argentine definition, cited in International Bioethics Committee of UNESCO (1995)

#20: Information on eventual pathology, not therapeutic but predictive.

—Zaire definition, cited in International Bioethics Committee of UNESCO (1995)

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