



## BACKGROUND

In 2005, we, as members of an NSGC task force, proposed a definition of genetic counseling practice, which was approved by the NSGC Board and published in the *Journal of Genetic Counseling* in 2006 (Resta R, Biesecker BB, Bennett RL et al. *J Genet Couns.* 5:77; PMID 16761103). The definition is as follows:

**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

Because the practice of genetic counseling has evolved and expanded since 2005, we questioned whether this definition was still relevant. As a proxy for relevance, we investigated whether the published definition is being used in genetic counseling research and training.

## METHODS

Because there are no standardized measures for determining the utility of a professional practice definition, we measured utilization two ways:

- 1) Standard citation sources (PubMed Central®, Google Scholar, Research Gate, Crossref) were reviewed to determine how often the definition was included in scientific publications since 2005.
- 2) A convenience sample of Program Directors of US and international genetic counseling training programs were surveyed about whether and how they incorporate the definition into curricula.

## RESULTS Citation History

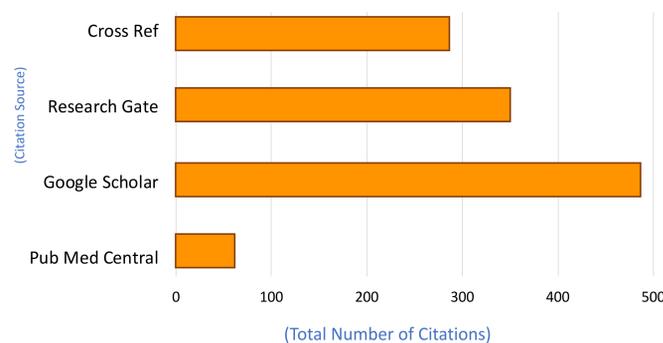
As of November 10, 2018, the definition was cited in 65 references in PubMed Central, 492 references in Google scholar, 349 references in ResearchGate, and 290 references in Crossref. The definition article was in the 99th percentile for citations in the *Journal of Genetic Counseling*.

Differences in citation numbers reflect differences in core publications in each database, methodology in surveying publications, and goals of each citation service.

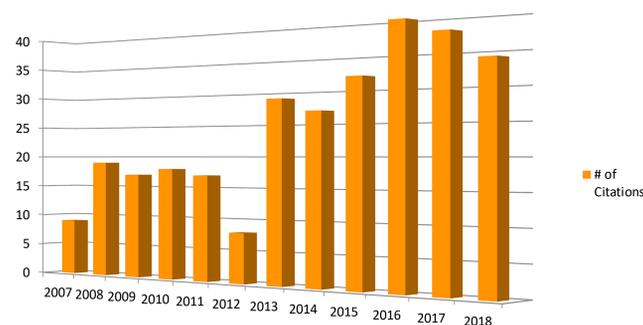
The number of citations has increased over the past 5 years compared to the first five years after publication and the definition has been cited in a wide range of journals and texts.

## RESULTS Citation History

### GC Definition Article Citations 2006 - 2018



### Number of GC Definition Article Citations Per Year



The paper itself is in the 99<sup>th</sup> percentile of papers cited in *Journal of Genetic Counseling*.

## RESULTS Program Usage

Nine United States-based and ten international graduate program directors in genetic counseling responded to our request for information. All of the US-based program directors indicated that they had incorporated the definition into their program curriculum as an essential element. There were also thoughts about ways to broaden it.

*"I use this definition all the time, with GC students, healthcare providers, interested students, and lay people. I think the three components of what [GCs] do remain the same..."*

*"I do teach it and I think it still works well, but might be a bit limited..."* This respondent also noted that the NSGC vision statement *"is also a good definition of genetic*

*counseling: 'Integrating genetics and genomics to improve health for all.'"*

Other respondents noted that while this definition was used in their program curriculum, it was also compared to other definitions of genetic counseling primarily for the purpose of providing historical context.

For the international respondents, all ten indicated that they use this specific definition for genetic counseling within their programs.

*"The elements listed in the definition guides the teaching [of the students]."*

*"I ask students to read the 2006 paper for discussion in the subsequent class about how the definition has changed and why... [it helps] set the tone for the types of philosophical discussions we will have throughout the program."*

## CONCLUSIONS

The NSGC genetic counseling definition has been widely used in research and training of genetic counselors in North America, Western Europe, Australia, and other global locations.

While we were unable to locate published criticism of the definition, we did ask the program directors whether they had any issues with how it was currently written. One respondent suggested that with the increasing study of how genetic factors not only contribute to disease but also to health, she stresses this broader application. Another respondent noted:

*"I think genetic counseling has become more than what is in the definition, but what is in the definition is necessary."*

Others stressed the difficulty of separating the process of genetic counseling from the skills, activities and roles performed by genetic counselors. Specifically, consideration was given to whether this definition still applies to a profession with an increasingly diverse employer base; particularly within the commercial and laboratory sectors.

While the definition appears to remain sufficient at present, given the dynamic role of genetic counselors and the changing practice of genetic counseling, this definition should be regularly re-evaluated to assess its continued utility.

## FUTURE AREAS OF STUDY

It would be interesting to formally evaluate the use of this definition in all genetic counseling training programs worldwide; to determine if this definition has been used in licensure bills; and to determine if the definition resonates with genetic counseling clients.

# What is Genetic Counseling ?