

GENETIC COUNSELING

How Inclusion of Genetic Counselors on the Research Team Can Benefit Translational Science

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Translational research in medicine is necessarily a team-based endeavor, and, indeed, translational research teams often include researchers from many different disciplines. We outline some of the practical challenges that are particularly salient to translational research, both for scientists and study participants, and propose that genetic counselors—a group of specialty-trained health care professionals who are as yet only infrequently recruited to collaborate in translational research teams—could contribute a unique perspective and skill set that would be invaluable in the effective navigation of these challenges. We propose that collaboration with genetic counselors could not only benefit individual translational research teams but also potentially help shift the research agenda for translational medicine.

CHALLENGES PRESENTED BY TRANSLATIONAL RESEARCH

Although there is no universally agreed-upon definition of the concept, translational medicine fundamentally aims to make the connection between medical research and improved patient outcomes more immediate (1). The merit of this type of endeavor is essentially universally recognized, but there are a number of challenges associated with the implementation of a translational approach to medical research.

Challenges for research teams

Participant recruitment, retention, and informed consent. Translational studies typically rely on the participation of human subjects. Unfortunately, the fact that few individuals who present for initial screening for participation in research actually enroll in studies has been highlighted as a major challenge for translational research (2). Perhaps related to this, consent forms related to translational medical research are often extensive and detailed and concern complex study-related procedures. In fact, some have argued that they can be so challenging for potential subjects to understand that they actually discourage participation (2). The net result is that recruitment and retention of participants, and ensuring that consent to participate is informed, can represent substantial challenges to translational research teams.

Clinical responsibility or research activity? One of the consequences of making the connection between medical research and improved patient outcomes more immediate is that the distinction between clinical responsibilities and research activities becomes less clear, and appropriately addressing situations that arise can be challenging for translational research teams. For example, consider the following scenario: A group of clinical researchers develops a concept for a study that involves taking a “rare diseases” approach to identify the etiology of craniofacial anomalies that affect patients at their clinic. Specifically, the team proposes to sequence genomes of patients and their parents in an attempt to identify genomic copy number variants that might be responsible for the phenotype. The team members recognize that they could generate data indicating that a particular variant may be responsible for a particular individual’s phenotypic outcome—which is the primary goal of their study—so should such findings be shared with the relevant family? If so, under what circumstances, by whom, and how? They also recognize that there is potential that in their comprehensive interrogation of the genome, they could stumble across incidental findings that might be of clinical importance for a research participant and so appreciate that they need to establish a strategy about how to respond in such a situation. For example, what if they stumbled across a genetic variant known to cause a condition such as familial adenomatous polyposis (an autosomal dominant condition in which individuals often devel-

op colon cancer at a young age), for which there are screening and prophylactic treatment protocols?

Challenges for participants

Translational research creates challenges for participants as well as for the research team. For example, in some translational research studies the sharing of genetic results or other kinds of information with research participants is an integral part of the study. Indeed, genetic testing may be a prerequisite to confirm eligibility to participate. Sharing of such test results with research participants is a sensitive and controversial issue in translational research for at least two main reasons. First, many genetic test results (as well as other kinds of information that may be shared with participants in the context of translational research) are likely to be of a conditional, uncertain, or probabilistic nature (for example, finding that a particular genetic variant may increase vulnerability to depression when an individual is faced with stressful life events). Second, even those that are of a more straightforward nature (direct cause-and-effect relationships between a particular vari-



Fig. 1. A beneficial addition to the translational research team. By communicating effectively with patients about complex, technical information, genetic counselors could help recruit participants for clinical studies. Additionally, genetic counselors have a patient-focused perspective that could help shift translational research toward a more clinical direction.

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ant and a health outcome) may be difficult for participants to meaningfully integrate into their sense of self and could provoke powerful or unanticipated emotional reactions (3).

CAN GENETIC COUNSELORS HELP OVERCOME THESE CHALLENGES?

Because translational research is the domain of the multidisciplinary team, in which each participating researcher provides input toward the team's goals on the basis of his or her unique areas of expertise, we propose that particularly in the context of translational research projects in which genetic/genomic information is to be shared with participants (but also in a broad array of other situations, including those alluded to in the scenario described above), genetic counselors (Fig. 1) could be a valuable addition to translational research teams.

What is genetic counseling?

The term “genetic counseling” first emerged in the 1940s as a concept credited to Sheldon Reed, who envisaged the activity as a “kind of genetic social work without the eugenic connotations” (4). The most recent definition of genetic counseling is more specific, but Reed's concept is retained: “Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease.” (5) The first graduate-level genetic counseling training program opened at Sarah Lawrence University in 1969 (6), and today there are dozens located around the globe. The first professional organization for genetic counselors—the National Society of Genetic Counselors (NSGC)—was formed in the 1970s (7), and standards and processes for board certification of genetic counselors were established by the American Board of Medical Genetics in 1981 [now administered by the American Board of Genetic Counseling (ABGC)]. Certification of genetic counselors provided a means to define the skills of a genetic counselor by focusing training on a set of practice-based competencies.

Today, a typical genetic counseling program is a professional, two-year master's degree. Although most programs are professional (rather than research-based), in 2003 the ABGC formally mandated that all accredited training programs must include a core research component. In fact, students usually perform independent research and/or work in multidisciplinary research teams as part of their training. Admission to genetic counseling programs is highly competitive,

and most successful applicants have a strong academic background (usually in science or psychology). Graduate program course work entails both didactic and clinical practicum components that focus on topics such as human genetics, molecular biology, biochemistry, biostatistics, risk assessment, professional ethics, and research methods (8); content is constantly updated in response to technological advances. Graduates of these programs often work in clinical service capacities, but a substantial proportion are engaged primarily in research roles, and some have made substantial contributions in this domain. At some institutions (particularly large, tertiary care centers with an academic focus that house genetic counseling M.S. programs), genetic counselors are already integrated into some translational research teams. Barriers to the broader inclusion of this group in such teams at other centers probably include lack of awareness of the relevance of the genetic counselor's skill set. In fact, individuals graduating from approved training programs emerge with a high level of genomics-oriented scientific knowledge and expertise in communicating complex concepts in lay language as part of a unique skill set that we suggest would prove an invaluable asset to any translational research team.

Genetic counselors and translational research

Fundamentally, genetic counselors' training is devoted to producing professionals who excel at two interrelated tasks, both of which could beneficially be exploited by translational research teams. First, genetic counselors are experts in communicating about complex, often very technical issues with clients from a variety of educational, socioeconomic, and ethnic backgrounds. This communication process encompasses eliciting pertinent medical and psychological information and discussing risks, benefits, and limitations to various available courses of action and tests. Effective communication related to such complex and potentially sensitive issues is founded on the development of good rapport between the genetic counselor and the patient/client. This relationship relates to the second task at which genetic counselors are experts—namely, identifying and addressing client concerns and emotional reactions related to the discussion. Genetic counselors are explicitly trained (in a range of interviewing techniques and to provide short-term counseling and psychological support) to help clients understand their

experiences, behaviors, emotions, and attitudes in such a way as to clarify their own beliefs and values and to promote autonomous and informed decision-making.

As such, genetic counselors' expertise is obviously directly applicable to the challenging process of obtaining informed consent for translational research studies (2). Genetic counselors are ideally placed to help potential research subjects comprehend the lengthy, detailed consent forms that are often necessary for translational research studies. By doing so, they could both help mitigate the potential negative impact of these complex forms on participation rates and promote autonomous informed decision-making by potential participants.

Moreover, given the fact that of those individuals who are initially willing to participate in studies, the proportion who actually enroll is small (2), it seems that investing in the “front end” of studies by engaging team members who are specially trained in rapport-building and communication about complex issues (such as study procedures) may be worthwhile. Genetic counselors have a skill set that suggests that they may be able to facilitate both recruitment and retention of participants in the context of translational research studies. In each of these domains, we propose that genetic counselors' skills would serve both research teams and study subjects well.

Genetic counselors' training and skill set equips them to contribute more to a translational research team than simply through the types of direct interactions with study participants described above. For example, training programs and clinical experience all equip genetic counselors to manage large numbers of client (or research subject) case files, organize data, and communicate with team members, as well as track and summarize pertinent information—all skills that are crucial for a good research coordinator to have. Further, genetic counselors would be well placed to help a research team design the best approach to navigate the ethical and practical challenges that arise.

GENETIC COUNSELORS AS A CATALYST TO SHIFT THE T1 TO T2-PLUS IMBALANCE IN TRANSLATIONAL RESEARCH?

Recently, there have been attempts to introduce specific terminology to the concept of “translational research” by deconstructing it into its constituent components (1, 9, 10). Although there is no consensus about

the number of constituent components, the general idea is that the first layer of translational research (T1) involves using basic science to drive the development of clinical studies. Other layers of translational research address issues such as more patient-specific approaches, the development of evidence-based practice guidelines, and population-based outcomes research. Non-T1 translational research layers have been variously categorized as T2 (1), T2 and T3 (9), and T2, T3, and T4 (10).

A point on which there is little disagreement is that much of the translational research published to date focuses on the foundational T1 layer (1, 2). Furthermore, although translational research is usually conceptualized as a bidirectional continuum, that which has been published to date tends to focus on one direction: basic science to clinical efficacy. Certainly, the reasons underlying this phenomenon are complex and probably involve such factors as the pressure of funding agencies on basic science to engage in research that has more immediate health implications, and a shortage of clinician scientists who are well placed to drive research from clinical problems to basic research (11). Another potential factor is that, as yet, perhaps multidisciplinary translational research teams do not tend to include a sufficient number of individuals who would bring an alternate perspective to shift the focus of the research agenda toward T2 (and indeed T3 and T4) studies.

In addition to the practical and immediate consequences of including genetic counselors as part of the multidisciplinary research team, we suggest that with their

client-centered (or research subject-centered) perspective and the depth of engagement/interaction required of participants, genetic counselors are ideally placed to contribute to translational research teams at a higher level. Specifically, genetic counselors would arguably be uniquely able to identify and bring to the team new, participant-driven issues that may be worthy of investigation in the context of purpose-designed research studies. In contributing to the development of new study ideas, genetic counselors may bring a different perspective to the translational research team that could contribute to a shift in the focus of the research agenda toward the layers of translational research that, although important, have received less attention to date.

SUMMARY

We suggest that genetic counselors constitute a group of professionals with whom closer collaborative ties are likely to be invaluable to translational research teams, and we invite interested researchers to identify individual genetic counselors for potential research collaboration via the Web site of the NSGC (7).

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12. **Acknowledgments:** The authors thank S. Aufox for constructive comments on an earlier draft of this manuscript. **Funding:** J.A. was supported by the BC Mental Health and Addictions Research Institute, the Michael Smith Foundation for Health Research, the Canadian Institutes of Health Research, and the Canada Research Chairs program. H.Z. was supported by the University of Minnesota and University of Minnesota Medical Center–Fairview. **Competing interests:** The authors declare that they have no competing interests.

10.1126/scitranslmed.3001898

Citation: H. Zierhut, J. Austin, How inclusion of genetic counselors on the research team can benefit translational science. *Sci. Transl. Med.* **3**, 74cm7 (2011).

Science Translational Medicine

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Sci Transl Med 3, 74cm774cm7.
DOI: 10.1126/scitranslmed.3001898

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