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Genomic Counseling: Next Generation Counseling

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Abstract

Personalized medicine continues to expand with the development and increasing use of genome-based testing. While these advances present new opportunities for diagnosis and risk assessment, they also present challenges to clinical delivery. Genetic counselors will play an important role in ushering in this new era of testing; however, it will warrant a shift from traditional genetic counseling to “genomic counseling.” This shift will be marked by a move from reactive genetic testing for diagnosis of primarily single-gene diseases to proactive genome-based testing for multiple complex diseases for the purpose of disease prevention. It will also require discussion of risk information for a number of diseases, some of which may have low relative risks or weak associations, and thus, may not substantially impact clinical care. Additionally, genomic counselors will expand their roles, particularly in the area of health promotion to reduce disease risk. This additional role will require a style of counseling that is more directive than traditional counseling and require greater knowledge about risk reducing behaviors and disease screening.

Introduction

With the increasing availability and uptake of genome-based testing for individual risk assessments, a shift from traditional genetic counseling to what has been referred to as “genomic counseling” (Sturm and Manickam 2012) may be warranted. Though genetic counselors are likely the most qualified to discuss genome-based testing with patients, the current practice paradigm was developed primarily for diagnosis of chromosomal abnormalities and single-gene diseases (Phimister et al. 2012) and therefore, would require additional training and modifications to the current service delivery model. A number of recent publications have highlighted some of the challenges associated with counseling for whole genome/exome sequencing or direct to consumer (DTC) testing (Ashley et al. 2010; Middleton 2012; Riordan et al. 2012; Roberts et al. 2011; Sturm and Manickam 2012; Waxler et al. 2012; Corpas 2012; Darst et al. 2013). These case reports provide some insight on how genetic counseling will need to evolve to manage changes with respect to both scope and type of information to be communicated to the patient. In this paper, we explore some of the general challenges and differences in genomic counseling compared to traditional genetic counseling, and offer some suggestions to advance the field of genomic counseling to keep pace with the development of genome-based testing.

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Genetic vs. Genomic Counseling

The practice of genomic counseling will differ from traditional genetic counseling in four areas: 1) the number and/or type of diseases for which testing is available and discussed, 2) purpose of testing, 3) intervention and clinical utility and 4) access to testing. The current focus on highly penetrant single-gene disorders and chromosomal aneuploidies contrasts with genomic risk assessments for complex, multi-factorial disease typically with low penetrance. For genomic testing, use of array-based platforms or whole genome or exome sequencing will generate numerous findings of varying levels of risk, clinical validity and clinical utility, even if the test is ordered for a single purpose (such as seeking a diagnosis of condition with unknown etiology). Typically, patients who undergo single-gene genetic testing are affected or at risk for a genetic disease. While this may be true for patients undergoing genome-based testing, healthy individuals without family history or known risk factors may also be included (Patel et al. 2013). Thus, the model of traditional counseling of reactive or responsive medicine, where genetic counseling and testing are intended to respond to a clinical problem or question may not be applicable. Instead, a more proactive risk reduction model will be necessary to address genome-based disease risk results, which would include careful considerations of the utility of these results, and potentially counseling for disease prevention and therapeutic decision-making. Importantly, individuals can order genome-based testing and access their test results without consultation with a health professional, which may result in a genetic counselor being called upon after testing to help interpret and/or communicate information from reports from a DTC genomic test.

Typically, traditional genetic counseling for disease diagnosis can be divided into three main steps: pretesting, delivery of results, and follow-up. While pretest counseling will likely still be provided to patients offered genome-based testing in a clinical setting, the advent of DTC testing essentially eliminates this step and consumers will make their own decisions about the benefits and risks of testing. The delivery of results and follow-up consultations will be similar in format to traditional genetic counseling though substantially expanded to accommodate the scope of genome-based testing.

With genome-based testing, particularly in the absence of pre-test counseling, the post-test visit will likely be lengthened due to the scope of information to be discussed. One potential disadvantage of a single counseling session post-testing is that patients may not be adequately prepared for possible outcomes of testing and/or their perceived understanding or expectations may not align with the actual result. Of particular concern, the lack of psychological preparation for adverse results can impede an individual's ability to adapt to those results (Middleton 2012). Even if patients do consult a genetic counselor prior to genome-based testing, it would not be feasible to anticipate and counsel for every possible finding and clinical implications, particularly for predictive whole genome/exome sequencing testing (Ormond et al. 2010; Sharp 2011). Indeed, the few published reports on genomic counseling note the increased amount of time necessary for counseling (Sturm and Manickam 2012), which may not be feasible if genome-based testing becomes more widely used.

New and Expanded Roles as Genomic Counselors

Due to the increase in information revealed about multi-factorial diseases by many genome-based tests, one important change for genetic counselors will be a greater role of health promotion and the increased time required for genomic counseling (Biesecker 2001; Gettig 2010). Many counselors already engage in health-promotion and prevention, so this role will need to be expanded to ensure coverage of all risks revealed by genomic testing. Emerging evidence in diabetes research shows that diet modifications can reduce body fat for individuals with a specific genotype (Mattei et al. 2012; McCaffery et al. 2011; Peter et al. 2012; Florez et al. 2006; Zhang et al. 2012). It is reasonable to expect that similar evidence supporting lifestyle modifications will emerge with continued genomic research. In a survey of DTC customers, no significant change in diet or exercise was observed among those that spoke with a genetic counselor to review results. However, individuals who discussed the test results with a physician showed lower fat intake and increased exercise (Bloss et al. 2011), perhaps due to the differences in approach to care. Thus, the primary role of genetic counselors may be expanded to not only help patients understand their genetic risk and non-genetic factors that may affect their overall risk, but also to provide information about interventions, screening or other recommendations to encourage uptake of these preventive behaviors and reduce disease risk. Although there is little evidence that knowledge of one's genetic risk leads to behavior change (Marteau et al. 2010), this may be attributed to the lack of guidance and encouragement currently provided by genetic counselors or other health providers when consulted.

To fulfill this new role, elements of motivational counseling should be incorporated into post-test counseling visits. Motivational counseling is a patient-centered, directive method of supporting patient change by focusing on the patient's motivation to change and has been shown to be effective in improving adherence to diet and exercise plans as well as to promoting treatment engagement, retention and adherence, and improving overall health outcomes (Hettinga et al. 2005). The incorporation of motivational counseling has already been recommended for diseases such as Type 2 diabetes and colorectal cancer, for which genome-based testing is available (Pengchit et al. 2011; Waxler et al. 2012). Though there has been a tradition of non-directiveness in genetic counseling, more directive counseling may be necessary to encourage adoption of these behaviors (Waxler et al. 2012; Baker et al. 1998; O'Daniel 2010). Directive follow-up and recommendations should occur as soon as possible after receiving results to increase likelihood of behavioral modifications, particularly in individuals found to be at increased risk (Heshka et al. 2008; Scheuner et al. 2008). Successful motivational counseling will require knowledge of patients' baseline motivation for lifestyle changes. Markowitz et al (2011) found that less motivated patients were less likely to engage in disease prevention when given a low-genetic risk. Strengthening a patient's beliefs that changing behavior will reduce risk (perceived efficacy) and that the patient has the ability to change behavior (perceived self-efficacy) is critical to making a behavior change (Marteau and Lerman 2001).

Personalized interventions may improve outcomes with respect to improved risk perception (Pierce et al. 2000) and behavioral change (Amrhein et al. 2003). By emphasizing the importance of lifestyle changes to offset the increased risk for high risk patients, and

focusing on non-genetic factors' contribution to disease risk for low risk patients – all patients would be encouraged to consider behavior changes, regardless of genetic risk (Waxler et al. 2012). This strategy would be important to balance as patients' understanding of gene-environment contributions to complex disease, reducing possibilities of underestimating risk or believing genetic to be absolute and/or unchangeable (Haga 2009; Markowitz et al. 2011; Marteau and Lerman 2001).

Preparing for Genomic Counseling Practice

The National Society of Genetic Counselors (NSGC) has recognized genomic medicine as a field of potential growth through their efforts to improve counselors' knowledge and awareness by dedicating the Annual Educational Conference to genomic health in 2011 as well as establishing a Special Interest Group for Personalized Medicine. Currently, about 1.2% of genetic counselors surveyed in the NSGC Professional Status Survey (NSGC 2012) reported specializing in genomic medicine or personal genomics/genomic profiling. Given the expansion of genome-based testing, this number is anticipated to increase and thus, new approaches to counseling and changes to training programs will be needed to emphasize counseling for common complex diseases, preventive health, and motivational tools to help patients adopt healthy behaviors. It may be anticipated that genomic counselors will emerge as a new specialty of genetic counselors, similar to counselors specializing in the prenatal, pediatric, or cancer settings. To reinforce the importance and differences associated with genome-based testing and counseling, students should have the opportunity to observe genomic counseling in a clinical setting. This will help ensure that future genetic counselors will have exposure to the different practice approach associated with genomic counseling. Currently, genomic counselors working in industry receive specialized and continuous training for genomic counseling, such as was required by the personal genomic testing company Navigenics (Levin et al. 2012), but given the anticipated widespread use, all students should have some experience in this new field.

In addition, genetic counselors can strengthen partnerships with other allied health professionals to develop a multi-disciplinary approach for genomic risk assessments. Genetic counselors have a long history of working with other health professionals such as speech pathologists, physical therapists, psychologists, and others, to coordinate comprehensive treatment for patients requiring specialized care. Such a multi-disciplinary approach could include a range of health providers including internists, geneticists, genetic counselors, and genomic nurses (Battista et al. 2012; Chan and Ginsburg 2011). Much in the way that a child with a metabolic disease may attend a specialty clinic and see a geneticist, a genetic counselor, and a nutritionist in a single visit, a personalized health clinic could be created to enable patients to review genomic testing results with a counselor, meet with a nutritionist, exercise scientist, health coach, or any other healthcare professional that could assist with lifestyle changes or screening to reduce disease risk. A qualitative survey of patients participating in a personalized medicine collaborative reported that even when patients shared their genomic test results with their primary care provider, few physicians made lifestyle or screening recommendations based on those results; the patients believed their lack of recommendations was due to the provider not knowing what to do with the results (Gordon et al. 2012). Indeed, many physicians have limited knowledge in genetics

and genomics (Haga et al. 2012; Hofman et al. 1993; Hunter et al. 1998). Genomic counselors can help fill this gap in understanding and work with physicians and other providers to deliver comprehensive-based care.

Conclusion

As genetic and genomic technologies have evolved, so must clinical practices. There is a huge potential for genetic counselors to define the appropriate delivery of information derived from genome-based tests as well as encourage the integration of genomic risk information into all areas of medicine. These changes will necessitate adapting training programs and providing professional development opportunities to increase knowledge of genome-based, disease prevention and intervention options, and using a more directive counseling approach. It is essential that the NSGC and other professional organizations as well as certification boards like the American Board of Genetic Counselors continue to recognize and support education in this field to adequately prepare the current and future counselors to practice genomic counseling. With the appropriate training and support, genomics and personalized medicine can become a thriving and valuable specialty for genetic counselors.

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