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The future of genetic counselling: an international perspective

Barbara Bowles Biesecker[1] & Theresa M. Marteau[2]

The focus of clinical genetics, and thus genetic counselling, is forecast to expand from the diagnosis and prediction of rare, often untreatable conditions, to the prediction of common, often treatable or preventable conditions[1]. Whether this evolution is likely to proceed rapidly or at a pace that permits sensible integration of molecular genetic tools is unknown and a source of debate[2,3]. It is clear, however, that genetic discoveries will modify the way in which disease and risk are conceptualized. Here, we predict how genetic counselling, specifically for more common diseases, might be provided in the decades to come. We envisage an expansion of professional roles and expertise for many health care providers and highlight the need for counselling practices to become more evidence based. Although we support an evidentiary-based approach to the integration of genetic testing into practice, genetic advance is unlikely to occur in an orderly and standardized manner within countries, much less among different countries and health care systems[4]. Geneticists will become increasingly involved in professional education and policy-making regarding genetic testing and screening programs.

Present practice

It is important to distinguish genetic counselling from clinical genetics services and genetic testing or screening. The former is a communication and, in some cases, a psychotherapeutic process, while the latter are diagnostic or prognostic services. Current clinical genetics services and accompanying genetic counselling commonly involve the diagnosis (and prediction) of what are for the most part rare and untreatable conditions in fetuses, children and adults. Genetic diagnosis has traditionally been based on physical examination or family history but increasingly relies on molecular testing.

Worldwide variance in the professional identity and training of providers of genetic counselling implies that the practice differs between countries. Most clinical genetics services (with or without genetic counselling) are provided by physicians who are trained as medical geneticists[5]. In the United States and Canada, a significant degree of genetic counselling is also provided by genetic counsellors trained to the master's degree level[6,7]. Genetic counselling in Australasia and the United Kingdom is usually provided by nurses working alongside medical geneticists—and more recently by counsellors trained to the master's level[8–10]. In other parts of Europe, social workers or geneticists provide counselling[5]. No systematic comparison of the process of genetic counselling among differing countries has been conducted, although some surveys have been carried out[11]. These reveal differences in the attitudes and values of providers[12,13] and suggest significant differences in the practice of providing genetic information to patients worldwide.

Goals of genetic counselling

The goals of genetic counselling inform its practice. The goals of clinical genetics services and those of genetic counselling are often confused[15]. The counselling process may seek to facilitate informed and autonomous decision-making, appreciation of the inheritance of a genetic condition, integration of genetic information into a useful framework, or improvement in the emotional well-being of those affected or their family members. The objective of non-directiveness is also frequently discussed, although it is often unclear whether it is part of the process or a desired outcome[16,17]. Non-directiveness is based on the desire to uphold the personal nature of reproductive decision-making and a reluctance to pass judgement on the worthiness of the life of a person affected with a genetic condition[18]. Yet, in practice, patients may be influenced by the kind of information provided and the manner in which it is given[19]. Preliminary research suggests that patients may feel persuaded by providers[20]. Additional studies are required to determine the role of non-directiveness and its relationship to desired outcomes of genetic counselling.

The goal of public health programs is, generally speaking, to improve the health of a populace. It is possible to perceive this broad goal as being in conflict with autonomous reproductive decision-making. If a goal of some clinical genetics services (and supporting genetic counselling services) is to reduce the incidence of children with birth defects or genetic conditions, then it is implied that couples at increased risk ought not to reproduce and/or that a woman carrying a fetus affected with a genetic condition ought to terminate her pregnancy. Such a directive stance is inconsistent with the goals of genetic services in many Westernized countries, but providers, for example, in China and certain South American countries, openly strive to reduce the incidence of affected births[14,21]. And yet, the slant of 'directiveness' varies even between European countries—for example, Portuguese geneticists are more likely than their German counterparts to encourage termination of affected pregnancies[22].

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Notably, an explicit goal to reduce the incidence of individuals affected by genetic conditions is not in conflict with genetic counselling goals when the condition is treatable and hence the method of reducing the incidence does not depend on termination of affected pregnancies. Geneticists have been resigned to this conflict in goals for years, declaring that reproductive decision-making should be autonomous and informed, but that when a genetic condition can be treated, genetic testing should not only be offered but even mandated, as in the case of newborn screening.

The process of genetic counselling

Genetic counselling has been described by the American Society of Human Genetics as a communication process[23] and elsewhere as the provision of genetics education coupled with psychosocial counselling[24]. As more counsellors trained to the master's level have entered the field in the US, Canada and the UK, there has been increasing attention in the literature to the short-term psychotherapeutic aspects of genetic counselling[25,26]. Yet practice standards for genetic counselling are nearly non-existent or vary even within the same region or country[27]. For the use of certain genetic tests, several published policies or guidelines exist in the US, Canada and the UK, but the types of information given to clients and the manner in which the information is communicated are far from standardized, even for a specified test[28–30]. For example, the degree of psychological counselling and support varies significantly.

Our claims of practice variation are difficult to document; they are based on the sparse literature[19,31] and our experience training genetic counsellors in a variety of settings (B.B.B.) and observing genetic counselling sessions (T.M.M.). Efforts have begun to standardize the information provided in specific genetic counselling encounters, but they remain preliminary, are not evidence based and do not address the counselling process[32]. Accreditation by the American Board of Genetic Counseling has provided certain professional competency standards for genetic counsellors in the US (ref. 33); however, the delivery of clinical genetics services often involves time and reimbursement constraints that limit the degree (and probably the effectiveness) of genetic counselling. Genetic counselling practice may be shaped as much by the structure and limitations of various health care systems and by clinical genetics services as by the expertise of the counsellors. Counsellors may thus be constrained from using their counselling expertise to facilitate client decision-making and provide support.

The process and outcomes of genetic counselling have been documented by empirical studies infrequently[34,35]. The outcomes that have been studied include patient knowledge, reproductive decisions and patient satisfaction, but none of these successfully documents the overall effectiveness of genetic counselling. The process studies that have been conducted demonstrate that genetic counselling primarily involves the provision of genetic information and its implications but pays relatively little attention to the social, emotional and familial aspects of the information[36–38]. Whether current genetic counselling practices of providing information meet the needs of patients is largely unexplored, making it difficult to predict how successfully patient needs will be met in the future.

Genetic counselling in obstetrics

Historically, the majority of genetic counselling worldwide has occurred in the context of obstetric settings. Prenatal screening and diagnosis of fetal abnormalities, many of which are genetic, are now routine in Western countries. In Europe, the genetic counselling that accompanies such testing is primarily provided in antenatal clinics by obstetricians, in the UK and the Netherlands by midwives[39]. In these countries, clinical geneticists are most often involved after the diagnosis of a rare genetic condition[40]. In the US, much of the prenatal diagnostic counselling over the past 20 years has been provided by genetic counsellors, particularly for women eligible for amniocentesis or chorionic villus sampling because of their age-related risks of having a child with Down syndrome. Yet this is now changing, as it routinely falls to obstetricians[41] to offer prenatal screening for Down syndrome or spina bifida (by biochemical marker and ultrasound), who may do so without the accompanying pre-test counselling or the participation of genetic counsellors[42].

We believe that aspects of prenatal counselling should remain under the purview of genetic counsellors. Such professionals strive to encourage the informed and autonomous decision-making that is critical[43] in reproductive counselling. While insufficient evidence exists that genetic counsellors successfully achieve this, research from Europe and the US has shown that providing information that allows informed decisions about whether to undergo prenatal screening tests has not been as highly valued by non-genetics providers. This is shown by the brief, sometimes inaccurate, information given to patients by obstetricians[41,44,45]. This may be because some obstetricians do not appreciate the importance of information in making reproductive decisions or because they receive insufficient reimbursement for time spent on the education and counselling necessary to properly offer testing. Furthermore, research has shown that obstetricians in the US are more directive in giving advice than genetics providers[42,45]. Absent changes in priorities held by obstetric providers and sufficient resources allocated to support the provision of information in antenatal care, it is difficult to see how this situation will improve. The allocation of additional financial resources to improve prenatal diagnostic screening services and the determination of the most effective and efficient providers to uphold the goal of personal informed decision-making will require 'outcomes' research.

Future practice

A direct consequence of international human genome sequencing efforts will be the development of new molecular tests that some claim will herald a revolution in the practice of medicine[46,47]. Implementation of these tests will also drive diversification of genetic counselling practice. Some tests will identify mutations in genes that lead to mendelian disorders, while an increasing number will identify low-penetrance mutations in genes that contribute to common diseases and act in concert with environmental risk factors. These technical developments are likely to continue to outpace the collection of data needed to accurately interpret certain genetic test results and to recommend sound treatment or prevention strategies. It may take decades to collect sufficient epidemiological data to interpret tests for common diseases, and some tests are likely to be offered prematurely. For example, clinical testing for *BRCA1* and *BRCA2* mutations was made available in the US before molecular and epidemiological data necessary for the accurate interpretation of the test results were available[48,49]. Currently, when new tests become available, it is clinical geneticists, genetic counsellors and genetics nurses who are enlisted to assist in their implementation. We forecast that eventually genetic tests will be used directly by primary care providers and medical specialists to predict the risk for common diseases.

Specialization among genetic providers

The US and UK are seeing a trend toward specialization of genetic counselling. This movement has been driven largely by

the increasing availability of new genetic tests, which have introduced 'genetics care' into new, more common categories of disease. Cancer genetic counselling is an example of a recently evolved sub-specialization. Within the past five years, approximately 300 US genetic counsellors have become expert in this area (National Society of Genetic Counselors, Inc., Cancer Special Interest Group, NSGC Membership Directory; 1998). Similarly, there are national efforts afoot for haematology/oncology nurses to become specialized in genetics[50]. US genetic counsellors have moved into cancer genetics in response to employment opportunities. Similar movements are likely to occur in the future in psychiatry and cardiology, among others. There will not be sufficient numbers of genetic counsellors and genetics nurses to fill these new areas of specialization over the coming decades[51,52].

Genetic counsellors, genetics nurses and clinical geneticists will likely serve as pioneer providers as testing is introduced into new specialty areas; most primary care providers or specialists in other fields have insufficient expertise in molecular genetics to offer and interpret predictive genetic tests[53–55]. In the foreseeable future, however, genetic tests will become well integrated into medical practice, and pre-test education is likely to fall to nurses working in specialty clinics, for example, cardiology clinics. With parallel research into useful medical or lifestyle interventions for those identified at increased risk, it is likely that genetic testing will come to resemble other medical tests for risk prediction, such as cholesterol levels or prostate specific antigen screening. We speculate that genetic counsellors and genetics nurses will continue to facilitate decision-making for patients whose uncertainty about genetic testing is agonizing, for those who view the potential outcomes as life altering, or for those who are facing increased risk for conditions where prediction is available but there is no intervention.

Whether current genetic counselling practices of providing information meet the needs of patients is largely unexplored, making it difficult to predict how successfully patient needs will be met in the future.

Conflict between health and wealth creation

Genetic counselling practice in the US, UK, Canada and several European countries has advocated a non-directive approach to offering genetic testing. This practice may be challenged by aggressive marketing of genetic tests. Much of genetic testing for rare disorders historically has been offered through genetic services in academic centres around the globe. For-profit laboratories have commercialized testing in the US (ref. 56). The first commercial laboratories in the US that accepted DNA samples for testing strongly encouraged their clients to send samples only from patients who had undergone genetic counselling[57]. Some companies hired genetic counsellors and even today offer the service themselves. These laboratories assumed a responsibility to ensure that patients had adequate information to consent to testing. For-profit laboratories today more commonly place responsibility for pre-test counselling with the physician and will accept a sample sent by virtually any provider[58]. When the provider owns the laboratory, there are also financial incentives to submit samples. Traditionally, reimbursement for laboratory testing has exceeded that for clinical examination or counselling services[59,60]. In a milieu in which marketing materials promote testing and providers have incentives to encourage patients to undergo testing, non-coercive, personal decision-making about genetic testing may well be compromised.

The need for a financial return on investments could drive persuasive marketing of new molecular tests. A recent study comparing the written information presented about cystic fibrosis in 28 population-screening programs in the US and UK showed that commercial organizations may present genetic testing more persuasively than non-commercial organizations[61]. Pamphlets from commercial testing services used fewer positive statements when describing cystic fibrosis than did non-commercial services. This more pessimistic disease depiction may lead to more interest in testing and in avoiding bearing a child with the disease. It is important to note that providers in non-commercial settings may also encourage individuals to undergo testing. Research predicts that when providers are dependent on revenues, they may be resistant to presenting information about these screening tests in ways that might lead to lower uptake.

In addition to the persuasive ways[61] in which genetic testing is presented to patients, there are financial issues that directly affect their interest in pursuing testing. In the US, patients with health insurance and those with greater economic resources are more likely to choose to undergo testing[62]. Concern about health insurance discrimination also deters patients from testing[63,64]. Patients from countries with nationalized health care are more likely to avail themselves of genetic testing when it is offered as part of general health care[65]. While non-directive counselling is upheld in the UK and Canada, testing that is routinely provided under the health care system may be accepted as "physician recommended or endorsed." For example, because of population screening programs in the UK, citizens there are more likely to know whether they harbour a mutation for cystic fibrosis than are US citizens. Thus, issues of reimbursement and patient expectations within a specific health care system affect use of genetic testing. As more tests become available, it will be interesting to observe the extent to which nationalized health care systems encourage genetic testing and what criteria they use to make their determinations.

While tensions between providing patient choice regarding genetic testing and maximizing returns on investments in testing services are already evident, it seems likely that they will become more frequent as the number of commercial laboratories continues to rise[57]. Clinical geneticists, genetic counsellors and genetics nurses have important roles in helping to offset these potential conflicts for patients and to uphold the principle of autonomous decision-making. The expertise of geneticists and counsellors is critical when policies are being established for appropriate use of predictive tests. In response to recommendations from two National Institutes of Health–Department of Energy committees, the Task Force on Genetic Testing and the Working Group on Ethical, Legal and Societal Implications of Human Genome Research, the US government recently chartered the Secretary's Advisory Committee on Genetic Testing, which is slated to have a central role in developing a meaningful system to evaluate utility and validity before a test is marketed for profit (US Federal Government Charter; 1998).

Communicating risk and facilitating behaviour change

In predicting multifactorial or common disease, genetic status will be one of many factors contributing to an individual's overall risk. Those conducting genetic screening programs will need skills not only in communicating risk but also in facilitating behavioural changes to reduce the risk. Predictive testing for treatable conditions will require a different approach than testing for untreatable

conditions, placing less emphasis on the individual decision to partake in risk assessment and more emphasis on strategies to avoid preventable conditions. Debate has already begun on the nature and extent of the pre-test education and counselling needed to ensure informed consent when testing is offered for common diseases[66]. When there are specific medical or lifestyle recommendations that can be made to those identified at increased risk, the 'counselling' will become health education. The evolution from pre-test genetic education and counselling practices to making recommendations to undergo testing will probably vary according to disease, test and health care system. There will, however, remain genetic tests for untreatable conditions and other conditions for which genetic testing is complicated or personally threatening, necessitating more extensive genetic counselling by specialists.

Individuals' perceptions of risk affect their decisions about whether to undergo risk assessment and the likelihood that they will engage in risk-reduction behaviours. One of the aims of presenting risks in a clinical context is to convey a likelihood of an adverse event, avoiding false reassurance or fatalism, and without causing undue anxiety. The relative merits of the methods to communicate risk and avoid false assurance or fatalism need researching. Use of various media, including interactive modes, to present risk information effectively will be an important aspect of this research.

The potential benefits of risk assessment programs that include genetic tests will depend largely on how people use this information. Reducing risks will often require that they alter their lifestyle by taking medication, changing their diets, or increasing their levels of physical activity. A wealth of data from many cultures now exist that evaluate interventions aimed at changing such behaviours, using a range of methods in a variety of populations[67]. These data should form the basis for the study design of interventions to follow risk assessments that include genetic tests.

Professional education needs

General health care providers currently do not have expertise in clinical genetics or in molecular genetic testing[54,68]. Widespread education for health professionals in genetics, risk assessment and behaviour change is needed across the globe. While changes in medical, nursing and social work curricula target future providers, provision of continuing education for practitioners will be more challenging. Funds from science education foundations and governments will need to be earmarked for far-reaching efforts. Clinical geneticists, genetic counsellors and genetics nurses provide important expertise in educating their health care colleagues. Providers will need to understand basic genetic principles and clinical application of these new technologies as the genetic revolution unfolds.

The public also needs to become more acquainted with genetic tests. Improvements in public awareness will alleviate some of the extensive pre-test education needs that currently exist. Working to shape the expectations of patients about the optional nature of current tests and the importance of informed decision-making will help to ensure quality services.

Professional change lies ahead in genetic counselling. Change commonly leads to anxiety, but it can also be invigorating. Anticipating change offers an opportunity to be prepared. We speculate that within the next several decades, genetic counselling will expand to a more evidence-based profession. In anticipation of this new form of genetic medicine, we call for widespread gains in professional expertise in clinical and molecular genetics, and attention to the need for research on genetic counselling, risk assessment and behaviour change. In this way, some of the improvements in health promised by human genome research may be realized, if not maximized.

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