Fostering applications of genetics in primary care: What will it take?

Radical new approaches and resources will be needed to facilitate the appropriate use of genetic information in all aspects of medical care. As new discoveries are made, family history and personal genetic information will increasingly influence clinical decisions involving prevention, diagnosis, and therapy.[1] Yet most of these applications are in their infancy, with many still uninvestigated questions about how genetic information will become part of primary health care. These questions are important because only by involving public health planners and primary care clinicians will the broadest range of patients have access to the benefits and protection from the harms arising from advances in human genetics.

In a literature review in this issue, Suther and Goodson[2] provide a useful summary and critique of evidence about barriers that must be addressed as applications of genetics are incorporated into primary care. Reviewing 18 studies published between 1993 and 2001, the authors characterize barriers to "the routine provision of genetic services" that were perceived by primary care physicians as (1) lack of genetic knowledge, (2) lack of detailed or updated family history, and (3) lack of referral guidelines.

Identification of these important barriers is a start toward surmounting them, but the analysis is constrained by limitations of the studies that have been published to date. Most of the studies reviewed by Suther and Goodson were not designed primarily to identify barriers preventing primary care physicians from providing genetic services. Some attempted to describe or to identify gaps in the current state of practice, some to collect primary care physicians' opinions about what additional resources would be needed in the future. Most of the data are based on primary care physicians' self-report, rather than observed behavior. Their perspective is that of North American and European primary care physicians or genetic specialists, but they do not analyze barriers from the perspective of staff, patients, families, ethnic groups, payors, health system planners, clinicians in other parts of the world, or public health practitioners. The authors do not comment on whether, during the 8 years spanned by these published studies, clinicians' interest and knowledge or skill in using genetics in primary care changed. However, it appears that earlier studies focused on clinicians' lack of knowledge about genetics, while more of the later studies pointed out barriers related to implementing referral guidelines and constraints on the use of family history. Only a few of these published studies evaluated methods to increase the ability of primary care physicians to systematically apply genetics in practice, which is the most important issue for capacity-building in the near future.

What will it take? It may be instructive to consider the question, What specific "genetic services" should in the near future be within the province of primary care practice? Then "barriers" and ways to facilitate providing these services can be discovered. Suther and Goodson[2] infer that primary care practice is likely to include genetic risk stratification for prevention, counseling regarding genetic test results, referring patients with genetic risks, prescribing genetic therapies as they become available, and "comprehensive" family history-taking. I would add that primary care will inevitably involve revisiting genetic information as it affects people throughout the life cycle, and thus will require enduring, portable, searchable, and readily updated records of the family genetic history.

Implementing guidelines: Build on tasks familiar to primary care clinicians

Currently most genetic services are still provided in a specialty referral model. The Genetics in Primary Care (GPC) faculty development initiative[3] brought together educators in primary care and genetics, who agreed that in preparing primary care clinicians for a larger role in genetic medicine, it would be important to build on tasks already familiar to primary care physicians (pilot curriculum, October 2000 revision available at http://genes-r-us.uthscsa.edu/resources/genetics/primary_care.htm; accessed August 20, 2002). Certainly, identifying the need for specialty consultation and facilitating referral is a core task in primary care, upon which genetic education and resources can build. Identifying, counseling, and referring couples who have an increased risk for a child with a genetic disorder is familiar to all providers of prenatal care (e.g., obstetricians, midwives, 30% of family physicians in the United States)[4] and important for other primary care clinicians when patients have an affected child or are considering conception. Most physicians providing prenatal care appear to do minimal counseling before screening for genetic risk (e.g., the "triple serum marker screen")[5] but do routinely refer patients with identified risk factors for genetic counseling and prenatal diagnosis. Implementation of new guidelines, such as the recommendation to offer screening for carriers of cystic fibrosis,[6] poses an opportunity to systematize screening, to refine counseling protocols (including input from childbearing couples), and to define tasks for specialists and primary care physicians, and it should proceed hand-in-hand with further research on implementation and outcomes.

Scarcity of genetics professionals

Widespread implementation of more genetic screening is likely to increase greatly the demand on genetics laboratories

and counselors, potentially highlighting a barrier that was not explicitly addressed in the studies reviewed: namely, the scarcity or maldistribution of genetic consultants, which poses a barrier to some primary care physicians attempting to provide their patients with state-of-the-art genetic services. Recent data from a representative national sample of US family practitioners[7] indicate that 11% practice more than 2 hours' drive from a geneticist and 23% believe that it is very difficult or impossible for their patients to consult a geneticist or genetic counselor. While training primary care physicians to do a good job with initial counseling and screening will help, new forms of consultation resources are likely to be needed for remote rural and international settings.

"Lack of knowledge" about genetic tests sometimes means lack of evidence about clinical utility

As Suther and Goodson note,[2] evidence about the applicability of predictive genetic testing to the care of common diseases is still in its early stages of development. Therefore, "lack of knowledge" of genetic tests and "skepticism" about their clinical utility are generic concerns, not specific to primary care, but increasingly recognized within genetics and public health as key matters on which more evidence is needed.[8] At present, the most common examples of clinically applicable predictive genetic tests are those for hereditary cancer susceptibility.[9] Eighteen percent of family physicians in a 2000 nationwide survey[7] and 36% in a Philadelphia-New Jersey sample (Randa Sifri, MD, written communication, January 7, 2003) had referred patients for cancer susceptibility testing within the previous year; most family physicians had discussed concerns about a family history of breast, ovarian, or colon cancer with several families in the course of a year. As genetic tests for cancer susceptibility are marketed to nongeneticist physicians and to the public, primary care physicians will need to be able to perform the initial assessment of familial cancer risk, help eligible patients decide how and when to pursue genetic counseling, and manage patients at low and moderate risk by providing reassurance and optimal preventive care. In the area of familial cancer risk, because of frequent questions arising from patients, primary care physicians may be motivated to refine their knowledge and skills and to keep abreast of current evidence regarding preventive options for people in various risk categories. Management of moderate risk has not been studied nearly as much as identification and management of high risk. Each will have a different set of challenges. Primary care physicians may become the most appropriate clinicians to manage moderate-risk cases, ordinarily without genetics referral.

Balancing potential harms and benefits before referral for genetic testing

Primary care clinicians are conscious that there may be harms of pursuing genetic information, and they may be reluctant to advise patients to proceed in the absence of clear-cut potential benefit.[7,9] This "barrier" to genetic consultation is different from lack of knowledge or lack of clear guidelines about eligibility for referral. Genetic testing can significantly affect family relationships, lifestyle choices, and reproductive decision-making. The risks can also include costly and unnecessary testing, anxiety, misinterpreted results, and inappropriate or hazardous therapies. Primary care physicians in the *initial* stages of helping families consider whether and when to pursue genetic counseling and testing will need many of the same skills that genetic counselors use for assessing and minimizing potential harms. We will also need up-to-date empirical assessments of the incidence and natural history of harmful effects of genetic information.

Working within competing demands for clinician time and attention

A central difference between generalist and specialty practice is that patients infrequently visit their primary care physician with a single concern. Primary care physicians routinely "triage" and prioritize multiple medical issues with each patient during most encounters, and therefore they may perceive different "barriers" to addressing any particular health issue from one patient encounter to the next.[10]

Scant time is available for family history-taking and counseling in primary care. Our study involving direct observation of visits with family physicians showed that the average time spent when family history was discussed was less than 3 minutes.[11] E.C. Rich, MD (unpublished data, 2002), calculated that with current coding and reimbursement criteria, an internist could be reimbursed for 10 minutes of time for collecting, documenting, and acting on the family medical history. Waters[12] estimated that obtaining a typical three-generation pedigree requires 30 minutes. Two conclusions follow from these data: (1) new techniques to support the efficient collection and interpretation of the family genetic history will be essential in primary care, and (2) selective, rather than "comprehensive," family history-taking may be appropriate for many patients. Both ideas are ripe for research and development in primary care settings. Indeed, more efficient ways of assembling family history records could benefit medical geneticists, whose activities to document family history have also been severely underreimbursed.[13]

Selective approaches to family history-taking

It is unrealistic to assume that every patient in primary care will need the same intensive evaluation as that provided during genetic consultation. *Selecting* which patients require a comprehensive pedigree, verification of diagnoses and test results in family members, and counseling about the pros and cons of genetic testing may be the skill most required by primary care physicians. Thus it is an open research question, still unanswered, what type of family history-taking is indicated in various situations in primary care, and whether a multitiered process of family history-taking might be most efficient. The Family History Working Group of the GPC faculty development initiative suggested screening questions (patient's concerns, history of unusually early onset of common diseases, multiple affected family members, congenital conditions, ethnic background, and additional questions for reproductive-age

individuals) followed by elaboration of positive family history items, and they called for research to determine which elements of family history are most critical in different commonly encountered situations.[14]

Facilitating collection and interpretation of the family genetic history

This group also highlighted the need for tools to circumvent the time constraints, such as Web-based or otherwise portable, electronic family history records, initially completed by patients, that could be reviewed and updated, rather than constructed anew during various clinical encounters (E.C. Rich, MD, unpublished data, 2002). Techniques currently in development can potentially link the graphical family history to some forms of pedigree analysis, risk stratification, and practice guidelines (e.g., CancerGene, available at http://www.swmed.edu/home_pages/cancergene, accessed May 20, 2002, and Centers for Disease Control Draft Family History Tool, available at http://www.cdc.gov/genomics/info/conference/files/FamHistSurv.pdf, accessed January 23, 2003; other examples are given on the reference list[15–17]).

Much work remains to be done in the development and testing of tools that could overcome current barriers to the thorough collection and accurate interpretation of the family history. Ultimately, information technology has great potential to facilitate the application of genetic advances in primary care.

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