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Report of the Banbury Summit Meeting on the evolving role of the medical geneticist, February 12–14, 2006

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In October 2004, a meeting was held at the Banbury Conference Center in Cold Spring Harbor, Long Island, NY to discuss the training of physicians in medical genetics. The goal of the meeting was to consider strategies to increase the number of medical genetics physician trainees. Several initiatives were proposed[1] that have subsequently been the subject of further discussion and action (Table 1). One of the critical issues discussed at the 2004 meeting concerned the scope of medical genetics practice. Defining this would seem critical to attracting additional trainees, yet the issue is of sufficient complexity that it became clear that an additional meeting would be required to address it. Therefore, a follow-up meeting was organized and sponsored by the American College of Medical Genetics and took place at the Banbury Conference Center February 14–16, 2006. The invited attendees (see Appendix) represented a breadth of perspectives and stakeholder constituencies. This document summarizes the major conclusions and recommendations from that meeting.

STATEMENT OF THE PROBLEM

Medical genetics is a relatively young discipline and faces a challenge in establishing its identity that may be unique among medical specialties. Most specialty areas, such as neurology or cardiology, focus on a particular organ system, and others, such as pediatrics or geriatrics, on a particular age group. It is much more difficult to define the domain of medical genetics, as genetic issues apply to all organ systems, periods of life, disease entities, etc. The historical focus in medical genetics has been on prevention, diagnosis and management of congenital anomalies, rare single gene disorders, chromosomal abnormalities, and inborn errors of metabolism. This is a broad territory, encompassing both multisystem disorders such as Down syndrome and single system conditions such as cardiomyopathy. Within this territory, the roles of medical genetics may differ from condition to condition or from institution to institution or both. Various kinds of genetics professionals may be involved, including the physician geneticist, laboratory geneticist, genetic counselor, and genetics nurse (In this document the term "physician geneticist" or "MD geneticist" will refer to the M.D. Clinical Geneticist certified by the American Board of Medical Genetics (ABMG); "laboratory geneticist" will refer to the Clinical Cytogeneticist, Clinical Molecular Geneticist, or Clinical Biochemical Geneticist certified by the ABMG; "genetic counselor" will refer to the genetic counselor certified by the American Board of Genetic Counseling (or, previously, by the ABMG); "genetics nurse" will refer to the Advanced Practice Nurse in Genetics certified by the Genetics Nurse Credentialing Commission; "medical geneticist" will refer collectively to this group of diverse genetics professionals). Defining the scope of practice becomes an even more complex task when new approaches for risk assessment and management of common disorders, such as diabetes or hypertension, are considered.

The task is further complicated by two factors: first, genetics units have evolved differently at different institutions; second, the manner by which genetics can contribute to maintenance of health and treatment of disease is constantly changing with the rapid pace of advances in the field. The major goal of this meeting was to identify the principles that define the scope of practice of medical genetics. A critical premise of the discussions was that a medical genetics scope of practice must be defined in a manner that transcends individual institutional cultures and traditions. The role of a medical genetics unit should be just as clear in any institution as is the role of a neurology, internal medicine, or pediatrics. Individual practitioners within the unit might have distinct roles and areas of specialization, but the overall role of the unit should cover a comprehensive and well-defined collection of services.

CONTEXT

This effort at self-examination of a medical specialty is not unique. Similar initiatives have occurred, for example, in internal medicine[2] and child neurology.[3] Each of these disciplines, and many others, are facing challenges similar to those impinging upon medical genetics. These include rapid advances in technology that are changing patterns of practice; rising levels of medical student indebtedness that are driving students to more lucrative areas of medical practice; career

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Table 1
Follow-up on 2004 Banbury Summit Meeting by participating group

American College of Medical Genetics (ACMG)
Produce brochure about genetics training, distribute to medical students and residents in other disciplines (this has been done)
Provide session for genetics residency program directors at annual meeting (this has occurred, though now these meetings will take place at the APHMG meeting)
Develop white paper on the evolving role of the medical geneticist (a meeting on the medical genetics curriculum is being planned)
Association of Professor of Human and Medical Genetics (APHMG)
Create network of training program directors and course directors (this has been done)
American Board of Medical Genetics (ABMG)
Develop joint training programs (internal medicine, pediatrics, maternal-fetal medicine) (these have been developed)
Create committee to write general examination for certifying examination (the exam has been extensively reviewed and updated)
Broaden coverage of medical genetics examination (the exam has been extensively reviewed and updated)
Residency Review Committee (RRC)
Review and revise program curricular requirements (completed)
Consider alternative pathways of genetics training (in progress)
American Society of Hum Genet (ASHG)
Develop educational programs in genetics for K-college students (ongoing)
Invite MD/PhD students to participate in annual meeting (this has been done)

choice driven by lifestyle issues; reimbursement schemes that pay physicians less and less to do more and more, etc.

In the United Kingdom, the Clinical Genetics Society issued a statement on the Role of the Clinical Geneticist in 2000 (www.bshg.org.uk/documents/official_docs/clingenrole.htm). They noted:

"There remains a great deal of ignorance and mythology among many of our medical colleagues, let alone administrators and commissioners, regarding the exact role and function of a Clinical Geneticist within the health care system. Geneticists therefore have to continue to explain to the uninitiated what our responsibilities are and what training is required to perform the varying and challenging roles in this particular specialty in the year 2000."

In 2004, the Royal College of Physicians estimated a need for one whole time equivalent (WTE) geneticist per 250,000 population,[4] which equated to 200 WTE given the UK population at that time. Murray and Davies (www.clingensoc.org/Docs/CGS_Workforce_Planning.PDF) surveyed clinical geneticists in the United Kingdom that year and found 82.5 WTE clinical geneticists. A competency-based curriculum in clinical genetics was formulated by the Joint Committee on Higher Medical Training in the United Kingdom in 2005 (www.jchmt.org.uk/clingen/curr_clingenetics.pdf).

The European Society of Human Genetics held a workshop on provision of genetic services in September 2000. Their meeting report[5] laid down a set of principles and recommendations. They stated:

"Clear guidelines for best practice will ensure that the provision of genetic services develops in a way that is beneficial to its customers, be they health professionals or the public, especially since the coordination of clinical, laboratory, and research perspectives within a single organizational structure permits a degree of coherence not often found in other specialties. It is time that medical genetics is recognized by the EU as a specialty in all countries. In each country, adherence to the organizational principles of prioritization, regionalization, and integration into related health services will maximize the cost effectiveness of genetic actions."

Using the Royal College of Physicians estimate of the one WTE geneticists per 250,000 population, the United States would require approximately 1200 WTE. There are currently 1178 M.D. clinical geneticists certified by the American Board of Medical Genetics (ABMG) (http://www.abmg.org/genetics/abmg/stats-allyears.htm), but it is not known how many are in practice, or how many WTEs this represents. The Kaiser-Permanente system, which was highlighted in a talk at the Banbury Conference, employs 13 physician geneticists for a practice that covers 3.1 million members, for a ratio of one WTE per 218,000 members.

In the United States, findings from a workforce survey were published in the same issue of *Genetics in Medicine* as the report of the first Banbury Center conference.[6] Most physician geneticists who responded to the survey were board certified in another medical specialty in addition to genetics and 21% of MD geneticists also had a PhD degree. About 70% reported that their practices were full or nearly full, with limited ability to expand capacity to meet new demands. Not surprisingly, the majority was dissatisfied with current levels of reimbursement for genetic services. The uneven distribution of genetics professionals across the United States and across different areas of genetics practice was also noted as problematic. The report concluded that "there is a serious mismatch between the expansion of knowledge and clinical applications in the field of medical genetics and the size of the medical genetics workforce." A subsequent analysis of these survey data according to clinical subgroup corroborated the original findings across all subgroups and revealed that the biochemical geneticist workforce is in the greatest danger of being unable to meet demand in the next 5–10 years.[7] This is particularly troubling at a time when newborn screening programs are significantly expanding.

STATEMENT OF PRINCIPLES

The following principles regarding the practice of medical genetics were agreed upon by the conference attendees:

1. Medical genetics is a primary medical specialty dedicated to the use and interpretation of genetic information to

maintain and improve the health of individuals, their families, and their communities.
2. The primary medical specialty, as recognized by the American Board of Medical Specialties (ABMS), is comprised of board certified physician geneticists and clinical laboratory geneticists.
3. The purview of medical genetics services includes all of the following:
   a. Clinical and laboratory diagnosis, risk assessment, pedigree analysis, counseling, provision of therapy, and longitudinal medical care.
   b. Care to patients across the age spectrum and for conditions involving any organ system.
   c. Patients and their families.
   d. Practices and policies pertaining to public health and disease prevention.
4. The rapid pace of discovery in medical genetics necessitates a dynamic approach to training and implementation of new paradigms of care.

The ABMG was recognized as the 24th primary medical specialty board by the ABMS in 1991. Formal recognition of the ABMG by the ABMS required that separate groups certify nurses and genetic counselors. Regardless of how medical genetics professionals are represented in the world of organized medicine, however, within the health care system their roles should be understood to be complementary and coordinated. Criteria for approval can be found at http://www.abms.org/newbrds.asp. A candidate medical specialty board must establish that the new specialty constitutes ". . . a distinct and well-defined field of medical practice." The field must be represented by a single examining board and the American Council of Graduate Medical Education must approve training. Medical genetics is not a subspecialty of any other discipline. Two years of residency training in another discipline may be required as a component of the training of a physician geneticist; however specific training in medical genetics is viewed as distinctive and absolutely necessary to provide patient care deemed to be within the scope of practice of medical genetics. A physician can be recognized as a medical geneticist by state licensing boards, hospital staff credentialing committees, and third-party payers. The ability of the American College of Medical Genetics to be represented in the AMA House of Delegates, which is a major decision-making body of the AMA, requires that, due to its small membership base, 50% of ACMG members be physicians, of which 35% must be AMA members. This allows ACMG to actively participate in the development of the Current Procedural Terminology (CPT) billing codes through AMA and their related reimbursement through the AMA Resource-Based Relative Value Scale (RBRVS) Update Committee.

Genetics as a discipline deals with the coding, storage, transmission, and expression of information in the cell, how this information guides development and the interaction of the organism with the environment, and how variation in DNA sequence and copy number affect phenotype. Medical genetics consists of the application of this discipline to the improvement of health at the level of individuals, families, and communities. This application, which constitutes the scope of practice of medical genetics, includes clinical and laboratory diagnosis, risk assessment, pedigree analysis, counseling, and provision of care, including both management and treatment of disease involving any organ system in any patient across the lifespan of the individual.

This statement is not meant to imply that only medical geneticists can deal with these issues. Rather, medical geneticists have unique expertise in these areas. Any physician can treat a patient with headaches or seizures, and many primary care physicians do so; yet neurologists who focus on these areas are understood to be experts in handling these problems, and are called upon to provide care or consultation in the management of patients, especially those with complex problems. Similarly, genetics is expected to be incorporated into routine care across all of medicine and whereas a board-certified physician geneticist will not be involved in every medical decision based on family history information or interpretation of a genetic test, a physician geneticist will be understood to be the expert in these areas. Individual physician geneticists may have specific areas of focus, but collectively they will play a central role in the care of some patients with complex multisystem disorders with a primary genetic etiology, and a consultative role in the care of a much larger number of patients where genetic factors contribute to patient management. Physician geneticists will also play a critical role in providing expert clinical interpretation of the results of genetic laboratory tests, working in partnership with laboratory geneticist colleagues who direct these laboratories.

Any statement of scope of practice in any medical specialty must recognize the rapid pace of change in medical science. Genetics is the paradigmatic example of a rapidly evolving area of medicine. This represents a significant opportunity for medical geneticists, but also entails a responsibility. The opportunity is to stay at the leading edge of the integration of genetics into medicine, which is appropriate for a group of professionals who are intimately familiar with the principles of constitutional and somatic genetic variation and the relationship of genetic variation to health and disease. The responsibility is to retain the flexibility to move quickly into new areas that come to be illuminated by the genetics "spotlight." Geneticists will be called upon to assist in the care of patients with disorders that may be far different from those they encountered during training or in previous practice. This is illustrated by the rapid ascent of cancer genetics as a clinical discipline that was essentially nonexistent 15 years ago. It is also illustrated by major public health initiatives attributable to advances in genetics such as expanded newborn screening. We are entering an era in which genetic testing of every patient as a routine medical practice may be possible; hence genetics training and practice models will need to incorporate flexibility and versatility as core competencies.

POINTS FOR CONSIDERATION

Conference attendees recommended the following Points for Consideration to facilitate the provision of high quality medical genetic services, optimize the skills of medical geneticists, and position medical genetics for the future:

1. Medical genetics services are best provided by a physician geneticist working together with a team of professionals, including clinical laboratory geneticists, genetic counselors or genetics nurses or both.
2. The realization of an integrated model is best achieved through establishment of a consolidated administrative unit that has equal status in the academic institution or health system or both with other primary medical specialties.
3. Medical geneticists should provide leadership in the responsible introduction of new technologies, their integration into medical care, and monitoring of outcomes. Current examples include:
   a. Prenatal and newborn screening for an expanding range of genetic conditions.
   b. Application of genomic technologies to high throughput diagnostic testing.
   c. Development and application of informatic approaches to incorporate genetic and genomic data into patient care.
   d. Predictive testing to assess genetic risk of common disorders and to guide prevention and management.
   e. Clinical applications of pharmacogenetics.
   f. New approaches to therapy of inherited disorders.
4. Genetics training and certification should recognize the competencies expected of medical geneticists and the rapid pace of change of the discipline.
5. The medical genetics community should actively promote its services and educate colleagues in other specialties and should facilitate the integration of genetics into medical and public health policy and practice.
6. Medical genetic services must be made accessible to the entire population.
7. The medical genetics workforce must be increased to meet current and anticipated needs.
8. To prepare for the future practice of medical genetics, training and continuing education programs should include substantial exposure to molecular and population genetics, epidemiology, bioinformatics, and public health.
9. The pool of trainees who enter the field of medical genetics must be increased and broadened, and training pathways and the certification process must be aligned with this goal.

Medical genetics encompasses a broad range of services, offered to patients at all ages, with a variety of disease states or risks. Several distinct career paths have emerged under the umbrella of medical genetics. These careers involve different forms of training and certification, and play complementary roles in patient care. Some types of clinical services may be predominantly provided by one type of genetics professional; others are best provided by teams consisting of multiple types of genetics professionals. The roles of specific genetics professionals will differ depending on clinical needs and on availability of staff at a particular institution. Physician geneticists commonly provide diagnostic evaluations, including obtaining medical and family history, performing a physical examination, ordering, and interpreting laboratory tests. They formulate a management plan and may play a role in counseling, management, and treatment. Genetic counselors typically provide risk assessment and counseling to patients and family members, and to individuals planning to have a child. In many instances they may work as part of a team with a physician, obtaining medical and family history information, providing counseling, facilitating decision-making, participating in implementation of a management plan, and serving as case managers. Some or all of these functions may be provided by genetics nurses in some institutions; genetics nurses also may participate in treatment of patients with genetic disorders. Laboratory geneticists perform and interpret genetic laboratory tests, including cytogenetic, biochemical genetic, and molecular genetic tests. They often direct the operation of a clinical genetics laboratory, including the interpretation of laboratory test results, test development, and supervision of laboratory staff, and specimen preparation and analysis. Laboratory geneticists also provide consultation to other medical professionals on the interpretation of laboratory studies and their use in medical decision-making.

Health care systems, including hospitals and schools of medicine, are organized into administrative units. Some institutions have departments, centers, institutes, etc., in which genetics is recognized as a discrete unit on equal standing with other primary specialties such as pediatrics and internal medicine. In others, medical genetics may exist as a division within a primary specialty. Genetics laboratory services may be provided within the genetics unit, by another unit such as pathology or laboratory medicine, or by outsourcing to commercial laboratories.

Positioning genetics alongside other primary medical specialties is consistent with national recognition of the ABMG by ABMS. A very strong argument can be made for the consolidation of medical genetics professionals into a distinct unit, ideally a department on equal standing with other departments, and including a formal medical genetics service within the hospital. Genetic conditions do not respect traditional specialty or age boundaries. Care of families with genetic disorders often requires attention to the medical needs of both children and adults. A critical mass of genetics professionals can be assembled within a genetics unit to deal with genetic service needs across the institution. A reservoir of expertise required for training can be established that will serve diverse audiences, including students and practicing medical professionals. Genetic testing laboratories can coordinate with clinical geneticists to provide consultation on the interpretation of labora-

tory test results and their use in medical decision-making. Ideally the genetics unit would be a department of human or medical genetics, but other arrangements, such as cross-departmental centers, that may achieve the same goal of creating a critical mass of medical geneticists, will be successful in some institutions. The specific strategy used to create a genetics unit will need to be customized to the individual institution, but the principles of having a recognized unit and a medical genetics service at the hospital should apply across all institutions.

Genetics practice occurs in a diversity of settings in addition to academic health centers, including health maintenance organizations and private practice. Nonacademic settings for genetics practice will likely increase with the increasing relevance of genetics in common disorders. The argument that genetic services should be consolidated in a discrete unit applies to HMOs, though it is obviously less relevant to those in private practice (although private practitioners of genetics might still appreciate the possibility of being appointed to a hospital staff as a medical geneticist).

There is an increasing array of new approaches to prevention, diagnosis, and management of disease that is based on knowledge of genetics, and medical geneticists can play a critical role in introducing these within the health care system. Geneticists have long played a role in carrier screening for couples planning a pregnancy or in the screening of newborns for a variety of congenital disorders, especially inborn errors of metabolism. Molecular genetic testing has increased the possibilities for carrier screening and technologies such as tandem mass spectrometry are expanding the list of disorders that can be detected by newborn screening. Medical geneticists can work alongside obstetricians for the former and pediatricians for the latter to guide introduction of these new approaches and provide patient counseling and care. There are new opportunities to partner with physicians across all disciplines in the interpretation of presymptomatic and predispositional tests. Diagnostic tests are rapidly incorporating genomic technologies, such as array comparative genomic hybridization or expression array analysis, to rapidly query large segments of the genome. Geneticists can work alongside pathologists in the application and interpretation of tissue-based studies, and with clinicians for diagnostic tests performed on constitutional DNA. Genomic approaches will dramatically increase the complexity of data sets that can be used for patient management, requiring the use of informatics systems to guide physicians in the interpretation of these data. Medical genetics can be conceptualized as a form of information science and geneticists can help lead the way in the implementation of informatics in medical care. In some cases, genetic tests will be used to determine risk of disease before onset of symptoms, to stratify common disorders to guide choice of therapy, and to customize the type and dosage of drug to a patient's individual patterns of metabolism. Although geneticists may not have primary contact with all of the patients whose disorders are managed in this manner, they will have a major role behind the scenes in testing, data analysis, and informing medical decision-making. The array of therapeutic approaches available to treat genetic disorders is also rapidly expanding. Many of the patients geneticists have traditionally diagnosed, counseled, and followed longitudinally may eventually be managed with nonsurgical therapies. With their established relationships with these patients and their families, expertise in all aspects of genetic health care, and related case management role, medical geneticists will have increasing opportunities to provide a coordinated, patient-centered "medical home[8]" for these patients, which will include delivering these treatments and after and documenting patient outcomes.

These diverse roles for medical geneticists place significant demands on training program curricula. It is clearly insufficient to focus training only on prenatal diagnosis, dysmorphology, and biochemical genetics, the traditional mainstays of medical genetics training. Increased exposure to genetics of common disorders, risk assessment, adult-onset disorders, molecular genetics, pharmacogenetics, informatics, epidemiology, public health, and population genetics, among other areas, will be critical if geneticists are to lead the way toward what is variously called "genomic" or "personalized" medicine. Training programs will face the challenge of providing state-of-the-art exposure to a broad array of genetic and genomic approaches that may exceed the training experience of the teaching staff. The current generation of medical genetics professionals will need to remain current in the face of a rapidly evolving field and must instill in trainees the skills to maintain their currency throughout their careers.

The opportunities to integrate genetics and genomics into all areas of medicine, and to serve the entire patient population, will place major strains on the genetics workforce. This challenge will be compounded by the physician workforce shortage predicted in America by 2015 and beyond, and will call for creative approaches to trainee recruitment.[9–11] Increasing the size of this workforce will be critical. To some extent, the increasing visibility of geneticists as members of the patient care team will provide role models to attract new trainees. The diversity of career paths in medical genetics, including roles as physician, laboratory geneticist, genetic counselor, and genetics nurse, and opportunities in patient care, clinical investigation, and translational research should appeal to a broad array of interests. Careful attention will need to be given, however, to aligning these opportunities with training pathways. The core competencies required of all medical geneticists will need to be identified and training program curricula coordinated with the certification process.

CONCLUSION AND NEXT STEPS

The two Banbury Conferences have focused on the needs to attract an increasing number of physician genetics trainees and to define the scope of practice of the medical geneticist. Several of the recommendations of the two meetings are currently being addressed by groups including the ACMG, ABMG, and the genetics Residency Review Committee (RRC) (Table). One of the critical points common to all these efforts is the need to define a set of core competencies in medical genetics. One can

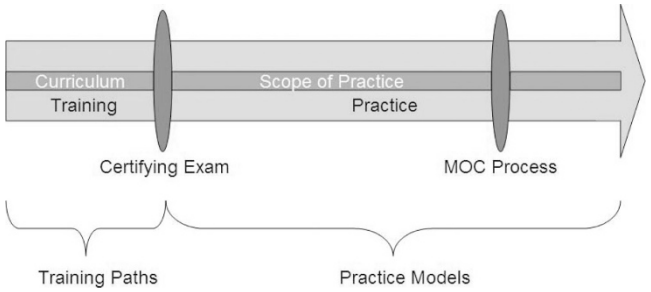

Fig. 1. Vector of genetics career path. The curriculum of the training program is consistent with the statement of the scope of practice in medical genetics, the certification examination and the maintenance of certification process assuring the quality of the training and the continuing education processes.

conceptualize a "vector" that spans the entire career of a medical geneticist, from initial training, through the board certification process, to continuing education and maintenance of certification (Fig. 1). Defining these core competencies is tantamount to defining the essence of what it means to be a medical geneticist, and will inform efforts to align the training programs, certification examination, continuing education, and maintenance of certification process. It will also help to forge the identity of the medical geneticist as a distinct player within the health care system. Efforts are now underway to define these core competencies through joint efforts of the ACMG, ABMG, and RRC.

APPENDIX: MEETING PARTICIPANTS

Meeting Organizers: Bruce R. Korf, MD, PhD (Chair), University of Alabama at Birmingham; David H. Ledbetter, PhD (Cochair), Emory University School of Medicine; Michael F. Murray, MD (Cochair), Brigham and Women's Hospital. Participants: Ronald P. Bachman, MD, Kaiser Permanente Medical Center (Oakland); John Belmont, MD, PhD, Baylor College of Medicine; Judith L. Benkendorf, MS, CGC; American College of Medical Genetics; Miriam G. Blitzer, PhD, University of Maryland School of Medicine; Leah W. Burke, MD, Vermont Regional Genetics Center; Garry R. Cutting, MD, Johns Hopkins School of Medicine; Charles J. Epstein, MD, University of California San Francisco; Gerald Feldman, MD, PhD, Wayne State University School of Medicine; Lynn D. Fleisher, PhD, JD, Sidley Austin LLP; David Ginsburg, MD, University of Michigan, HHMI; Marilyn C. Jones, MD, Children's Hospital, UCSD; Dale Lea, RN, MPH, Foundation for Blood Research; Michael Mennuti, MD, University of Pennsylvania Medical Center; Reed E. Pyeritz, MD, PhD, University of Pennsylvania School of Medicine; Piero Rinaldo, MD, PhD, Mayo Clinic College of Medicine; David S. Rosenblatt, MD, MUHC Montreal General Hospital; Maren T. Scheuner, MD, MPH, RAND Corporation; Joe Leigh Simpson, MD, Florida International University College of Medicine; Angela Trepanier, MS, CGC, National Society of Genetic Counselors; Jerry Vockley, MD, PhD, Children's Hospital of Pittsburgh; Michael S. Watson, PhD, American College of Medical Genetics; Georgia Weisner, MD, MS, Case Western Reserve University School of Medicine; Richard J. Wenstrup, MD, Children's Hospital Research Foundation; Jonathan Zonana, MD, Oregon Health Sciences University.

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