

## CONFERENCE SUMMARY

### **Genetic Testing and Public Policy: Preparing Health Professionals**

**May 13, 2002**

**Hyatt Regency**

**Baltimore, Maryland**

On May 13, 2002, the Secretary's Advisory Committee on Genetic Testing (SACGT) convened a public meeting on genetics education of health professionals, *Genetic Testing and Public Policy: Preparing Health Professionals*, in Baltimore, Maryland. This meeting was the culmination of more than a year of information gathering, analysis, and consultation with experts in the field by the SACGT Education Work Group, chaired by Dr. Joann Boughman. The conference considered the challenges of integrating genetic testing into clinical and public health practice, identified obstacles and gaps in genetics education of health professionals, and proposed recommendations to be considered by SACGT. More than 200 individuals from academia, public health, industry, government, patient advocacy groups, and medical or specialty organizations were in attendance.

The meeting was organized into a morning plenary session with three speakers, a panel discussion of a hypothetical case study and the roles of different healthcare providers, four concurrent afternoon focus groups, and a report on the outcomes of the focus group discussions.

Dr. Georges Benjamin, Secretary of the Maryland Department of Health and Mental Hygiene, opened the meeting by welcoming the conference attendees to Maryland. Dr. Benjamin recognized the extraordinary pace at which genetic information is being generated and the challenges to educating health professional students and practitioners. The state of Maryland was one of the first states to offer newborn screening and continues to expand and will be updating its program with the use of tandem mass spectrometry in the next few years. Dr. Benjamin emphasized the important need to understand the impact of genetic information as new technologies and tests are developed.

Dr. Eve Slater, Assistant Secretary for Health, U.S. Department of Health and Human Services, delivered a welcoming address by video. Dr. Slater recognized the numerous positive developments in the field of genetics, but emphasized the need to have an adequately educated health workforce to appropriately apply and interpret genetic tests. More than 30 years ago, the need for enhanced genetics education for health professionals was recognized and since then, several schools, foundations, professional organizations, and government have responded to this need. In addition, innovative approaches in continuing education are critically needed to ensure the integration of genetics into the clinical and public health settings. However, understanding the science and the medical benefits of testing are only part of ensuring the appropriate use of

genetic tests. Health professionals should also be aware of the ethical, social, and legal implications associated with this field including concerns about the misuse of genetic information. In conclusion, Dr. Slater extended best wishes for a successful meeting and looks forward to hearing the outcomes.

Prior to introducing the three plenary speakers, Dr. Boughman briefly reviewed the conference agenda and reminded the audience of the interactive format of the conference and that the success of the conference depended on the mutual exchange of information. The first plenary speaker was Dr. David Mallott, Associate Dean of Medical Education at the University of Maryland School of Medicine. Dr. Mallott began his presentation by reflecting on what were considered new tests more than 25 years ago – prenatal ultrasonography and CT scan – and the issues that were raised about their usefulness and application. Looking back, however, even though there were questions about the utility of these new tests early on, the field of imaging and radiology dramatically changed how disease was diagnosed and stimulated new advances in healthcare. Similarly, the field of genetics presents a new way to diagnose and understand disease and is on the cusp of a new revolution in medical testing. Dr. Mallott reviewed the uses of genetic testing and highlighted the differences between genetic tests and other types of medical tests.

Genetics holds great potential for improving healthcare and it is this potential that will generate the pressure to educate providers about this new and exciting field. A major obstacle to enhancing genetics education is the fact that healthcare is still organized in traditional disciplines and departments. Genetics is pervasive through traditional disciplines and the question of who will speak for genetics and who defines the level of expertise on genetics in primary care is of great concern. It will be important for health professionals to be able to demonstrate competency and not just familiarity about genetics. Dr. Mallott asked what should students be taught and at what point in the continuum of education – high school, undergraduate, first year of health professional education, training/residency, continuing education – should they be taught. In addition, the invisible world of genetics (the antithesis of the field of imaging) presents difficult challenges in public comprehension and understanding and calls for a tremendous translational effort to explain the invisible world to the public.

The next speaker, Dr. Eugene Rich, Chair of the Department of Medicine at Creighton University School of Medicine, discussed the use of the family history as the primary predictive test. Dr. Rich began by describing the impending applications of genetics in primary care and the major role that family history has played in the field of medical genetics. In genetics, family history is typically collected through a family history questionnaire, validation of family medical information, and a telephone and/or face-to-face interview. The family history information is recorded as a pedigree in the medical record and can take three to five hours, more than half of the service time occurring before and after the clinic visit. The family history is particularly important when used in determining the predictive value of genetic tests, and is included in the calculation of the patient's personal risk or prior probability.

Dr. Rich then moved on to examine the current state of family history in adult primary care. The traditional teaching about family history in primary care has focused on the age and health or age and causes of death of the parents and siblings of the patient as well as a yes/no checklist of exposure to several diseases and conditions. Over the years, medical education about family history has improved by better teaching the type of information that should be collected in a family history and the usefulness of the information. In family medicine, family history has been considered a “ritualistic inquiry,” but that the primary objective is to detect possible threats to the health of family members and their future offspring. Often, a family genogram is used to record a family’s background and potential health problems. The genogram differs slightly from a pedigree in that it displays not only biological, but also social and interpersonal relationships as well as the major elements of a pedigree.

In adult primary care, the implementation of family history has not been successful as compared to medical genetics. In a study by Crouch and Thiedke (J Fam Pract, 1986), family history of alcoholism, emphysema, and mental illness were seldom recorded. Similarly, in another study, 63 percent of family practitioners reported obtaining a family history in comparison to 85 percent of obstetricians/gynecologists (Acton et al., Acad Med, 2000). It has been noted that younger, residency-trained physicians have a higher rate for obtaining a family history, however, the average time devoted to a family history discussion was less than three minutes for new patients and less than two minutes for return visits (Acheson et al., Genet Med 2000).

Some of the barriers to enhancing family history taking in primary care include time constraints, current reimbursement policies, practice organization and resources, patient expectations, and knowledge and information applicability. Currently, family history tools are available to primary care practitioners, such as paper family history tools, computer-based examples, Internet tools, and electronic medical records. In conclusion, Dr. Rich predicted that family history aids would consist of electronic tools for patient entry and interact with a portable electronic family history and electronic medical record. Point-of-care tools would then provide advice about predictive genetic testing and medical management.

The final speaker, Dr. James Evans, Clinical Associate Professor of Medicine and Director of the Clinical Cancer Genetics Program at the University of North Carolina, discussed genetics in primary care and the spectrum of utility in predictive genetic testing. Dr. Evans stated that genetics is already present in primary care. For example, an emergency room physician would consider the family history of a patient with chest pains in the diagnostic work-up and adjust the treatment accordingly. Other common diseases with a confirmed genetic component encountered in primary care include cancer, infectious diseases, Alzheimer disease, and schizophrenia.

Dr. Evans stated that genetics has evolved from a field focused on the past to one of the future. Predictive genetic tests provide a potential risk or probability of developing a future disease or condition. A predictive genetic test is determined useful if it has a high predictive power and provides an opportunity to intervene in a disease process and affect outcome, however imperfect. The utility of a test is constantly changing and hinges on such factors as the availability of an

effective treatment. Today's genetic tests span the spectrum of utility from high utility (characterized as highly effective, acceptable and with a presymptomatic intervention; e.g., multiple endocrine neoplasia type 2) to harmful (no effective intervention; e.g., Alzheimer disease). Dr. Evans reviewed two genetic tests of intermediate utility – colorectal cancer (HNPCC) and breast/ovarian cancer (BRCA1 and 2) – as well as the test for Alzheimer disease (ApoE).

In conclusion, Dr. Evans reiterated that the position of a test on the spectrum of utility is subject to change. The dynamic character of the spectrum of utility will be influenced by progress in treatment of disease (decreased testing), improved presymptomatic diagnosis of disease (increased testing), efficacious options for prevention with adverse side effects (increased testing), and options for prevention with minimal side effects (decreased testing). In addition, the position of a test along the spectrum will differ among individuals based on personal and familial experiences.

In the next session, a panel of health professionals of different disciplines participated in a discussion on the various roles of healthcare providers in the provision of genetics services and the differing levels of genetics education and competencies. Dr. Reed Tuckson, SACGT member and Vice-President for Consumer Health and Medical Care Advancement at United Health Group, moderated the panel discussion. The panel was composed of seven individuals: Dr. Allan Bombard, Vice President and Chair of the Department of Obstetrics and Gynecology, Lutheran Medical Center, New York; Dr. Caryl Heaton, Director of Graduate Medical Education and Family Practice Residency Director, UMDNJ-Robert Wood Johnson School of Medicine, New Jersey; Dr. Marilyn Dumont-Driscoll, Associate Professor of Pediatrics, University of Florida; Dr. Lemmietta McNeilly, Associate Professor and founding Chair of the Department of Communication Sciences and Disorders, Florida International University; Dr. Kenneth Offit, Chief of Clinical Genetics Service and Associate Attending Physician, Memorial Hospital, New York; Dr. Reed Pyeritz, Chief of the Division of Medical Genetics at the Hospital of the University of Pennsylvania and Professor of Medicine at the University of Pennsylvania; and Ms. Paula Rieger, a nurse practitioner, University of Texas M.D. Anderson Cancer Center.

To understand how different professions are trained to think about genetic issues and how they define their roles in the provision of genetics services, Dr. Tuckson began the discussion with a hypothetical case study. A newborn presents with an oro-maxillo-facial malformation at birth following a normal pregnancy with no family history of birth defects. After significant study and work-up, it is determined that the child has SACGT Syndrome, an autosomal dominant condition with a known causative gene for which a genetic test is available. SACGT syndrome is pleiotropic and typically presents with craniofacial abnormalities at birth resulting in speech/language impairment and swallowing difficulties. Affected individuals can undergo corrective surgery to repair the oro-maxillo-facial malformations. The syndrome is also characterized by variable expression and a high incidence of developing head and neck cancer in the second to third decade of life. With close surveillance and treatment, affected individuals are able to live to their 50s and 60s.

Dr. Tuckson first asked Dr. Pyeritz to respond to the case, describe how he was trained, and what it means to be a medical geneticist. Dr. Pyeritz stated that since virtually all disease is genetic and that genetics crosscuts most medical specialties, all health professionals should be familiar with genetics to some degree. Genetics is a primary medical specialty that requires specialized training and board certification similar to internal medicine and surgery. However, geneticists have avoided appearing self-serving and therefore, have neglected to define the field of genetics and when a referral should be made to a geneticist. The case study emphasizes the importance of the multidisciplinary nature of genetic conditions since SACGT syndrome is pleiotropic. Furthermore, even though a test is available for the hypothetical syndrome, the analytical validity, clinical validity, and clinical utility of the test were not described to determine the appropriate use of the test.

As a pediatrician, Dr. Driscoll stated that her role would be to serve as the medical home and coordinator of care for the child and family. Due to the variable expressivity of the syndrome, other family members may also be at risk to develop cancer and require evaluation. The pediatrician would also provide support to the family who are likely to be overwhelmed with medical information about the diagnosis of the child. In response to a question from Dr. Tuckson about who should provide information regarding reproductive decisions, Dr. Driscoll stated that the pediatrician plays an important role in exploring those issues with the family. For the general pediatrician, Dr. Driscoll stated that there are likely significant deficiencies in knowledge about the diagnosis and care for children with genetic conditions such as described in the case study due to the explosion of genetics information. Also, there is sometimes a lack of recognition that a genetic component is relevant in certain conditions.

As a family practitioner, Dr. Heaton responded that she would focus on the immediate need to bring the family together to support the parents and child. The scope of services provided by a family practitioner depends on a number of factors including location of practice (urban vs. rural) and whether the parents and/or the child are the patients of the family practitioner (the child may see a pediatrician). Dr. Heaton would likely refer the child and family to a specialist. In response to a question of how well prepared family practitioners are to provide care for the wide range of needs of the child and family in this case, Dr. Heaton responded that if a practitioner knows nothing, the first step would be to consult with a professional about the case and find resources to guide the medical management of the child and family. Family practitioners should recognize their limits of knowledge and know when to consult or refer a case to a specialist.

As a nurse practitioner, Ms. Rieger stated that nurses often serve as case coordinators in bringing together the necessary specialists to care for the patient and family. However, wherever the patient receives his/her healthcare services, it is highly likely that a nurse(s) will be involved in the provision of that care. Nurses assist in assessing the case, making diagnoses collaboratively with the physician, and deciding how to manage the case. In particular, nurses would be involved in health teaching for a lifelong condition as described in the case study, including education for the patient and family about how this disease is going to impact their life, potential risk factors for certain types of cancer associated with the syndrome, how to best manage the

disease and steps they can take to diminish their risk, and the psychosocial implications of the disease. Advanced practice nurses may also provide genetic counseling to the patient and family.

Before moving on to the next panelist, Dr. Tuckson commented that the last three panelists each stated that they could serve as case coordinators or the medical home of the patient and family and asked who decides which health professional should coordinate. Dr. Heaton responded that it doesn't matter who coordinates as much as whether it gets done. Dr. Tuckson asked Dr. Driscoll whether pediatricians have the necessary knowledge to provide care and counseling for the child and family. Dr. Driscoll responded that the level of genetics knowledge would depend on when and where they were educated. Genetics is often combined with neonatology on the pediatrics board examinations, and therefore, there isn't an incentive to have a strong genetics background. Pediatrics sub-specialists in genetics can sit for a separate board examination in medical genetics.

Turning to the speech-language pathologist, Dr. Lemmietta McNeilly stated that speech-language pathologists should be aware of the genetic etiologies of certain syndromes that require their services. Traditionally, speech-language pathologists have significant training and knowledge of single-gene disorders, but have lesser knowledge about multi-factorial issues and how it affects their clinical assessment practices and intervention. A speech-language pathologist can see a child and their family several times per week and are able to establish a good rapport with the family. Often, they are presented with a variety of questions that most speech/language pathologists would have difficulty answering. Although the pathologist is not equipped or trained to provide statistical or risk probabilities regarding the recurrence of a genetic condition in future offspring or the development of clinical symptoms outside of their medical specialty, they do have the opportunity to provide information about the various resources that are available to that individual and where to find that information, and to make a referral.

As an oncologist, Dr. Offit sees his role as two-fold: 1) to identify/diagnose these syndromes; and 2) to treat patients if effective treatments are available or suggest participation in appropriate clinical cancer trials. Dr. Offit commented that professional organizations such as the American Society of Clinical Oncology have made great strides in promulgating cancer genetics to the larger healthcare community and has developed a cancer genetics curriculum. Dr. Tuckson inquired about the importance of understanding the penetrance of a gene leading to cancer and how it affects the clinical management of the patient. Dr. Offit responded that this question could be asked of all cancers and that the spectrum of decisions is based in large part on the efficacy of the intervention. For example, if an effective chemoprevention option were available for SACGT syndrome, then the issue of penetrance would not be a critical issue. In contrast, patients at risk for ovarian cancer, where the penetrance is variable and prevention and detection measures are unproven, the decision to undergo risk-reducing surgical intervention becomes difficult and requires multi-disciplinary interface.

The last panelist, Dr. Bombard, was asked to comment from the perspective of his previous position in the health insurance industry about the reimbursement of the provision of genetic

services. In his experience, insurers refer to relevant professional colleges and organizations for guidance regarding the appropriate care and testing of patients with genetic conditions. Insurers then decide which management or treatment option would be most cost-effective and beneficial since they have a fiduciary duty to the employers that are paying for the healthcare to provide coverage of services that would benefit their employees. But issues in genetics extend beyond the patient/member to the entire family, which creates tremendous challenges for insurers. For example, with respect to cancer genetics, the proband is usually over 65 years of age and therefore, Medicare would provide some coverage of services. However, if a physician wished to determine if the affected individual in the family carries a genetic marker that could be used to screen other family members, Medicare does not pay for presymptomatic tests.

In light of the rising costs of healthcare, Dr. Tuckson noted that the same services could be paid multiple times since many of the panelists indicated that they would provide similar or overlapping services. Dr. Bombard agreed that this is a critical concern to health insurers but stated that patients will request to see specialists even though a primary care practitioner could handle their needs.

In closing, Dr. Tuckson asked each panelist to briefly state what policy recommendation(s) they would suggest SACGT propose to the Secretary in terms of genetics education and training.

- Dr. Pyeritz made two recommendations: 1) to raise genetic awareness and genetic education for all health professionals; recognition by SACGT would greatly help spread the message of the importance of genetics to specialty societies, boards, and other relevant groups; and 2) to recommend that it become standard practice for pretest and post-test counseling in every aspect of genetic testing, and therefore acknowledge key role of the genetic counselor.
- Dr. Driscoll recommended federal funding for faculty development programs in order to have protected time for faculty not only to be involved in the development of these educational programs but also in the development of the testing for the accreditation.
- Dr. Heaton recommended more funds be allocated for educational resources; increased governmental support for faculty development projects and continuing education for the practicing physicians; support for coverage of family history-taking, counseling, and a standardized online family history system to assist health professionals; and most importantly, support for translational research.
- Ms. Rieger agreed with Dr. Pyeritz's recommendations and suggested that SACGT think outside the traditional boxes and to encourage the multitude of specialty societies to provide more specialty education to those professionals who will encounter patients with genetic conditions.
- Dr. McNeilly recommended that health care professionals from a variety of disciplines who are not necessarily MDs be included in groups that are providing education as well as serving as members of research teams that can assess the actual

clinical implications for the infusion of genomics.

- Dr. Offit emphasized the importance of multidisciplinary training as a major priority and a commitment to access of genetics services across the board, independent of means, and recommended the need to re-examine Medicare and Medicaid coverage policies of genetics services.
- Dr. Bombard recommended that preventive presymptomatic testing be covered under Medicare, because all of the health plans use Medicare as the default coverage policy and significant efforts would be required within the different health plans to cover something that's not covered by Medicare.

Professor Diane Hoffman, Director of the Law & Health Care Program and Associate Dean at the University of Maryland School of Law, delivered the luncheon address. Professor Hoffman spoke about laws regarding genetic testing and discussed how and in what context genetic testing is currently used, lawsuits pertaining to genetic testing, and how future legal issues. In the employment setting, there have been three major legal cases involving the use of genetic testing in the employment setting: Norman-Bloodsaw v. Lawrence Berkeley Laboratory (1998), Terri Seargent (2000), and EEOC v. Burlington Northern Santa Fe Railroad (2001). Other legal cases involving genetic testing have been related to paternity, adoption, criminal cases, torts, malpractice, and products liability.

In the afternoon, participants divided into one of four focus groups to concentrate in depth on the issues raised in the morning session and the different areas of genetics education, training, and integration and to develop recommendations for consideration by SACGT to address problem areas or gaps in these areas. The four focus groups concentrated on one of the following topics:

- I. Genetics Content and Curricula in Health Education
- II. Inclusion of Genetics in Training, Examinations, and Accreditation Requirements
- III. Development of Genetic Tools and Resources
- IV. Implementation of New Developments in Genetics
  - a. Cystic Fibrosis Screening Guidelines
  - b. Pharmacogenetics
  - c. Genetic Guidelines for Congenital Hearing Loss

Following a break after the two-hour focus group sessions, the meeting was reconvened in plenary session to hear the reports of each focus group. The moderators of each group were asked to briefly discuss the major points of the group's discussion and provide a summary of the group's recommendations.

Dr. Bruce Korf, co-moderator of **Group I** with Dr. Jean Jenkins, reported on their group's discussion and recommendations. The first question considered by the group is whether genetics should be taught as a separate course or integrated into the curriculum. Before addressing that question, it was noted that there is a general need to excite students and colleagues about



genetics. No firm conclusion was reached on how genetics should be included in the curriculum, but the group strongly felt that, at the very least, genetics should be integrated. The group recognized some value in highlighting the fundamental principles of genetics in order to sensitize students to the integration that they're likely to see from that point forward in the curriculum. The term "guerrilla genetics" was used to describe a stealth approach to inserting genetics into the curriculum wherever the opportunity arises.

Some of the obstacles identified by the group included the need for reinforcement of genetics knowledge on board and other qualifying examinations, access to time in the curriculum, lack of resources to meet demand for knowledge (particularly the number of faculty who feel qualified to teach genetics), and the lack of faculty recognition of the importance of genetics in their practices. The point was also made that there should be a focus not on the genetic disease but on genetics and its contribution to biology and health. However, the group recognized the challenges in advocating the importance of genetics when most of the current-day applications are relatively limited.

Several issues raised during the discussion included how much genetics education is needed for general health professionals and the difference between consultation and empowerment. An important point was made that the more health professionals know, the more engaging they will be with genetic specialists, which will result in more consultations by genetic specialists.

Genetics is in the midst of an identity crisis as a profession. Traditionally, genetics has been considered a specialty dealing with rare diseases – single-gene disorders, chromosomal disorders, inborn errors of metabolism, and congenital anomalies. Until geneticists recognize the need to expand their own horizons and to be involved in the genetics of more common disorders, they will risk marginalization. Genetics will eventually be integrated into medical practice and the question was raised whether geneticists will help lead the charge or be left behind clinging the traditional view and icons of genetics.

Considerable time was devoted to discussing the value of the NCHPEG core competencies and there was a general consensus that the core competencies were a good starting point, although it isn't realistic to expect all health professionals to become mini-geneticists. The phrase "genetics vital signs" was used to describe the critical elements that a clinician should recognize as an alert to provide genetics services (e.g., testing or counseling) or make a referral. The NCHPEG core competencies have pulled the basic genetic concepts out of medical education, but now there is a need to reintegrate those concepts back into practice and to recognize the potential for using these as a basis for continuing education as well as undergraduate education.

The need for increased funding was quickly identified as an action item for SACGT to consider, but several questions were raised— specifically what types of projects should be funded and what would be the value-added for supporting those projects? The idea for a Human Genome Implementation Project was discussed and the need to develop a diverse body of clinical educators in genetics. Above all, further development of a set of educational approaches may be

needed to advocate the importance of partnerships with consumers, the use of genetics as an information science, the need to use varied technologies and approaches in education, and a recognition of the role of the commercial sector in genetics knowledge dissemination.

Dr. Monsen, co-moderator of **Group II** with Dr. Glenn Davis, reported on their group's discussion and recommendations. Dr. Monsen described how the three topics assigned to Group II – training, examinations, and accreditation – are inter-dependent. Education drives testing, which includes licensure, certification and accreditation, because accreditation standards are often developed from educational programs, and that those testing and accreditation standards again contribute to patterns in education, new developments, and curricula foci.

The group identified some of the pressures that educators, test developers, certification item writers, and accreditation bodies face, including state and federal certification and licensure requirements, Medicare and Medicaid programs, and practice guidelines. Payers and the commercial sector have a major influence in determining what material is included in health professional curricula. The Food and Drug Administration is influential in terms of how medical products, diagnostic tests, and laboratory procedures are perceived. In addition, litigation and the general public (consumers, media, watchdog organizations, etc.) are extremely important in calling attention to how healthcare providers are prepared.

Several major challenges were identified regarding the training of health professionals, including time priorities and priority-setting challenges. Another major issue is the lack of institutionalization of genetics knowledge. Faculty development is important and train-the-trainer models are helpful to increase faculty knowledge of genetics. The group also encouraged the establishment of interdisciplinary training programs.

Genetics competencies should be applicable at both the general practice level for all providers as well as be profession-specific. Competencies should be performance-based and medical organizations and professional societies should work to focus on the translation of discipline-specific competencies into delivery and practice, behaviors, and attitudes for their members. The group also suggested that genetics competencies be provided to specialty testing organizations since many test writers and groups don't possess genetics expertise. As a way to measure provider competency, performance indicators or measures for clinical genetics providers and specific behavioral measures for the delivery of clinical genetics services should be developed by professional and specialty societies.

It was recognized that the biotech industry plays a large role in provider education. Exploration of joint partnerships that would include industry as well as academics and direct care providers and clinicians should be encouraged. Since Medicare and Medicaid coverage policies largely define which services private health insurers reimburse, the group recommended that a national assessment of the provision and quality of genetics services be conducted. The group also recommended the inclusion of genetic testing for rare diseases under federal legislation for orphan drugs and orphan products to specifically evaluate the effectiveness and quality

assurances associated with rare disease genetic testing.

Dr. Bonnie Pagon, co-moderator of **Group III** with Ms. Robin Bennett, reported on their group's discussion and recommendations. The group was asked to consider what resources and tools were needed and who should develop them, fund them, and approve them. The group first considered what role SACGT, professional societies, or other groups potentially have regarding the development of genetic tools and resources. The group was very supportive of the SACGT process and felt that SACGT should consider focusing on the issues related to educational materials and education itself as a future area of study. Two key standards should be considered when reviewing genetic resources and tools: quality and currency of the information. The group was very complementary of SACGT's outreach efforts to bring together diverse groups and to listen and consider different perspectives regarding oversight and other issues in genetic testing. Collaboration is a key element in the development of any educational resources and perspectives of diverse audiences need to be considered during the process.

It was suggested that SACGT could play a similar to the U.S. Preventive Services Task Force in reviewing educational resources and guidelines regarding genetic testing. In this role, SACGT could help set a research agenda to fund projects on the validity and utility of genetic testing which would help in the development of evidence-based guidelines and information. If SACGT were able to review educational resources, they could also help identify gaps and promote multi-agency funding.

One of the major needs identified was the need for a common language in genetics that can be understood with a shared vocabulary by scientists, consumers, policymakers, media, legislators, and by our accrediting and certification bodies. However, time did not permit definition of a process for developing a common language.

Ms. Bennett reported on the group's discussion regarding genetic tools. In light of the morning presentations, the group first looked at family history tools as a resource, but found that more funding is needed to provide an evidence-base for the efficacy such tools. There was some discussion about transportable medical records in terms of family history, where a patient brings their own family history. In order for this to be successful, it will be necessary for the public to be educated about the importance of genetics and to understand how to update their family history. In particular, the group was strongly supportive for teaching genetics at a very early stage. Equally, the health provider needs to be educated on how to interpret family history information. Faculty development projects could help increase awareness about family history and genetics in general for health professionals.

Regarding the dissemination of genetics tools, the group concurred that continuing medical education was a useful approach to reach the practicing healthcare workforce. In addition, promotion of multigenerational teams would help health professionals that were trained years ago to become aware of new advances and tools in genetics.

Dr. Carole Kenner, co-moderator of **Group IV** with Dr. Rodney Howell, reported on their group's discussion and recommendations. The group concentrated on the steps involved in the implementation of new developments in genetics. The development of guidelines is incredibly important in terms of moving genetics forward. However, in discussing the cystic fibrosis guidelines, it was suggested that more attention should have been given to how the guidelines would be implemented after they were developed. The cystic fibrosis guidelines consist of two parts, the clinical guidelines and the laboratory guidelines, and it was not clear how the laboratory guidelines should be implemented and by whom. Nor was it clear what groups were involved in the development of the guidelines and whether certain groups that have been affected by the guidelines were included, which would contribute to the confusion about implementation. One commenter noted that help was needed in translating developments in pharmacogenetics and that perhaps SACGT could play a role in bringing together the relevant groups and organizations to develop clinical guidelines.

Another important issue that was raised was informed consent – specifically, who needs it and how should the consent be formatted. Some institutions use very long, detailed consent forms that may take up to 20 minutes to review. The group asked whether a balance could be reached, where a shorter version for informed consent could be used and whether it could be in the format of a brochure or other type that would be very explainable and usable, yet still address the issues of informed consent.

The group recommended that increased resources be devoted to the implementation of new developments in genetics, but noted that more consideration should be given to how, when, and what populations should be involved in these processes. Similar to Group III, the group recommended the need for practice guidelines, but stated that more translational research needs to occur first with more focus on outcomes, evidence of utility, and cost-effectiveness. One of the major obstacles to the implementation of new developments in genetics is reimbursement of new services and tests.

Dr. Howell reiterated that the group found practice guidelines to be extremely helpful although sometimes difficult to implement. He also repeated the need for help in developing resources and guidelines for pharmacogenetic testing.

In summary, Dr. Boughman highlighted the major recommendations put forth by the four focus groups. Several groups mentioned the need for integration and to bring together professional organizations from different professions to carry new developments in genetics forward. The institutionalization of genetics and genetics learning is also extremely important to adequately educate students and health practitioners. A common genetics language could facilitate communication between health professionals of different disciplines, consumers, policy-makers, and accrediting and certification bodies.

Diverse inclusion and collaboration are essential to forwarding the research agenda, translating new research findings into clinical applications, and developing evidence-based practice

guidelines and implementation strategies. SACGT should review and challenge the Secretary and the HHS agencies to fund some of these studies, to develop new models for faculty development and to develop new mechanisms to fund teaching and training of genetics to health professionals in general as well as providing funds for the training of genetic specialists to expand that workforce.

Some of the major policy issues that were raised by the focus groups included informed consent issues, access to genetics services and underserved populations, and rare disease genetic testing. The need for collaboration was emphasized several times and SACGT was encouraged to continue fostering a public dialogue on genetics issues and being inclusive and attentive to all perspectives.

In closing, Dr. Boughman invited participants to attend SACGT's public meeting on May 14-15 where she would present an overview and lead a discussion of the outcomes of the education conference. She encouraged participants to continue today's dialogue with colleagues, students, and administrators to advance genetics education of health professionals and the public. Dr. Ed McCabe, Chair of SACGT, also thanked the audience for their participation in the education conference and reiterated that the Committee always welcomes public comments.