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My Year as 1999 ASHG President

Uta Francke

It has been a pleasure and a challenge to serve as ASHG President during these times of rapid change. Since its inception in 1948, the Society's main purpose has been to serve its members by bringing together investigators in many areas of human genetics and to encourage and integrate their efforts by providing a forum for sharing research findings. This is accomplished primarily through our annual meeting and our journal, the AJHG. The ASHG also plays an increasingly important public role. Numerous current public policy issues and debates are relevant to human genetics, and the Society is called upon to offer advice and take a stand. We clearly need to strengthen our efforts to inform legislators and their staff, health policy makers, and the general public about all aspects of human genetics.

Who Are We?

Our Society has over 6,700 members, a 3% increase from last year. It is truly international, with more than a thousand members living overseas. Demographic information, collected over the last two years, reveals great diversity. About half of the members are engaged primarily in research, 30% in clinical work, 10% in counseling, followed by administration and teaching. This level of diversity sets our Society apart from other learned societies. Our meetings and our journal provide opportunities for learning and interaction across a wide range of human genetics research and practice. Organizational details can be found at http://www.faseb.org/genetics/ashg/org.htm.

What Happens at Our Annual Meeting?

The Program Committee, under Lynn Jorde, has followed directions by the Board to reorganize the program of the annual meeting by spreading out the invited speaker sessions, previously clustered on the first day. This change serves to balance the broader educational perspectives with the reporting of focussed new research results. Platform presentations of selected abstracts are considered a valuable component of our meeting and will be continued. To counteract the trend towards subspecialization, however, we enlarged the plenary sessions in which the best work from different subdisciplines is presented to the entire group of attendees. Late-breaking research reports will be integrated in these sessions. To reach out to the public, Paula Gregory, chair of the Information and Education Committee, organized educational sessions for high school science teachers, and Jane Salomon workshops for science writers.

For my Presidential Address, entitled "Human Genetics in the Information Age," I considered how the revolution in information technology is changing the way we do our work—and even the type of work we will be doing—in research, laboratory diagnostics, as well as in clinical genetics and counseling. In particular, I assessed the impact of the information revolution on three areas that are tightly linked to each other: the acquisition, dissemination, and interpretation of new knowledge. The full text is available at the ASHG website: http://www.faseb.org/genetics/ashg/ann-meet/99-pres/pres-addr-1999.htm. The main points are summarized in the following paragraphs.

To successfully mine the human DNA sequence for genes that are responsible for or predispose to diseases and encode potential drug targets, we will need an organized interdisciplinary effort that includes mathematicians and software engineers to further develop the informatics tools; biologists to formulate the questions, to interpret the output, and to annotate the sequence; and clinicians and pathologists, who understand disease processes, to suggest possible links between genes and diseases. Data by themselves are useless, unless they are turned into knowledge by the interaction of all these components. Communication between researchers working with model organisms and human geneticists will illuminate the connections. Cross-species workshops, focusing on specific developmental or signaling pathways, are planned to facilitate the dialogue.

To disseminate the rapidly growing new knowledge, we are moving from exclusively paper journals to electronic versions of the paper journals, with new electronic-only journals also being launched. The ASHG-owned journal (AJHG) under the new editor, Stephen Warren, is shortening the time from submission of articles to publication in electronic format. For optimal

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dissemination of new knowledge, however, we also need free and unlimited access to the electronic literature (i.e., to the complete articles, not just the MedLine abstracts that are currently available through PubMed). Towards this goal, PubMed Central (PMC), at the National Library of Medicine on the NIH campus, will be an archive of primary research papers, in all areas of life sciences, that offers easy search and retrieval across a wide range of scientific literature. It is envisioned that all journals will eventually participate and deposit their issues after print publication and that new Web-based journals will be created as part of PMC. The Board of Directors of ASHG has had intense discussions as to whether and when the AJHG would be contributed to PMC. No decision has been reached. The reasons in favor of this proposal are detailed in my Presidential Address. Without any doubt, PMC will benefit the community of scientists by accelerating the discovery of new knowledge and its worldwide dissemination.

The availability of health-related Web information to the general public has a significant effect on our practice of medical genetics and genetic counseling. Although resources for the genetics professional are growing on the Web, there is a paucity of online genetic information, tailored to consumers, which is accurate, up-to-date, and accessible. Web-based tools for genetic education for health care professionals and the lay public are needed. As a professional society, we are concerned about the discrepancy between the explosion of new knowledge and the shortage of trained clinical geneticists and counselors who can convey this information and provide services on a one-on-one basis. The Internet offers a solution. Web-based genetic information will increase the awareness of genetic conditions and of the role of genetic factors in common diseases among the public. Awareness creates more requests for services. Interactive Web-based systems will be developed that can provide accurate, timely, and individualized genetic information.

What Role Do We Play in the Public and Science Policy Arena?

Upon our request, the Society has been admitted to full membership in FASEB (the Federation of American Societies of Experimental Biology), the nation's largest association of biomedical scientists. Made up of 19 societies, with a combined total of 63,000 members, FASEB has a strong voice on matters of legislative and public interest and has campaigned successfully for increased biomedical research funding. We have appointed ASHG representatives to the FASEB Board of Directors and to seven different committees dealing with research funding, public affairs, and science policy.

In national debates about guidelines and regulations, many issues have come to the forefront and will continue to be addressed by the Society, its committees, and its members. These include the overhaul of the NIH peer-review system, the establishment of PubMed Central, the derivation and use of human pluripotent stem cells in research, the use of archived human tissues for research, federal oversight of genetic testing, and patenting of human genes and of genetic diagnostic tests. The protection of human subjects from research risks—including the protection of the privacy of genetic information collected as part of family studies—and issues of informed consent procedures for mentally disabled persons have caught public attention. With the formation of the Office of Human Research Protection (OHRP) under the Secretary of HHS—replacing the Office of Protection from Research Risks (OPRR) under the NIH Director—there will be increased scrutiny of informed-consent procedures and of the role of Institutional Review Boards. These developments are likely to have considerable impact on researchers in human genetics.

Under John Carey, an Ad Hoc Committee on Consumer Issues has been formed of researchers and leaders of advocacy groups. With the support of ASHG, a workshop was held to address consumer issues surrounding participation in clinical genetic research. The combined voice of researchers and consumer advocates will be needed to ensure that the progress of research will not be impeded by—and will be conducted within the framework of—existing federal regulations. The ASHG Social Issues Committee, under Mark Rothstein, has developed a statement on Genetic Testing in Adoption that will soon be released. As Social Issue Committees of other genetics organizations (ACMG, NSGC, and ISONG) are dealing with similar topics, the Board strongly encouraged joint sessions and activities of the ASHG committee with these committees.

To improve communication between the ASHG officers, the membership, and the public, a Communications Committee has been established by the Board to redesign our Web site, making it more visible, informative, and interactive. This effort should enable ASHG members to be informed of critical issues in a timely fashion and to act in influencing public policy. Active participation of the ASHG membership will be essential for some of these complex issues.

With increasing globalization, the communication between human genetics societies around the world is facilitated by the formation of the International Federation of Human Genetics Societies (IFHGS). The Federation currently has three full members—the regional societies ASHG, ESHG (European Society of Human Genetics), and HGSA (Human Genetics Society of Australasia)—and over 30 national societies as corresponding members. As I am beginning a three-year term as President of the IFHGS, I look forward to expanding the activities of this global organization in the information age. The new IFHGS web site is http://www.faseb.org/genetics/ifhgs/index.html. Elaine Strass, the ASHG Executive Director, deserves recognition for her expert management of the Society's office in Bethesda and special gratitude for serving as IFHGS Executive Secretary as well.