

## 2004 ASHG PRESIDENTIAL ADDRESS What Is Special about the “Human” in “Human Genetics”?

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I would like to begin where all of my predecessors as President have begun, by thanking the membership for placing their trust and confidence in me to serve as your President this past year. Serving in this role has given me a superb bird's eye view of the organization and the dedicated and knowledgeable people who work in the administrative office. I have also had an opportunity to share the thoughts and wisdom of a remarkable group of people on the Board of Directors and on the committees who give their time and energy as volunteers to our Society. To all of you, I want to express my gratitude and admiration.

When I began to compose my presidential address, I

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went to the texts of previous presidential addresses and discovered that it was déjà vu all over again: most of my predecessors had done the same thing and had read through the comments of previous Presidents to get some idea of what was expected of them. Most commented that this review of the “sacred texts” was not as helpful as they hoped. Why? They found, as did I, that there was no “formula.” H. J. Muller, Alfred Knudson, Janet Rowley, Arthur Beaudet, and David Valle gave scholarly dissertations on scientific areas of personal interest to them and, they hoped, more broadly to the membership. John Hamerton, Margery Shaw, and Charlie Epstein spoke eloquently about important social issues of the day, such as the controversy over the 47,XYY karyotype and anti-social behavior, the concepts of wrongful life and wrongful birth, and, most famously, Charlie Epstein's forceful reminder that “Not everyone loves human genetics.” Still others gave personal remembrances, such as Judy Hall and Hunt Willard, who spoke of their personal experiences and relationships with mentors and colleagues over the years. I did receive some advice from my adult children to “Keep it short and try to be entertaining.” I at least accomplished one of those goals (i.e., to keep it short). I donated a good portion of the time usually allotted to the presidential address to Brendan Lee and the Program Committee so they could increase the number of presentations at the plenary session.

I had to face the dilemma of being just one in a long line of presidents, many of whom were very distinguished geneticists, true giants in the field. What could I say that had not already been said? In this dilemma, I was not alone. There is a wonderful erudite set of essays on 18th century literature by a college professor of mine, Walter Jackson Bate, called “The Burden of the Past and the English Poet,” in which he discusses the problem faced by poets who had to ply their craft in the footsteps of the likes of Shakespeare and Dryden. In the introduction to his essay, he quotes an Egyptian scribe who wrote 4,000 years ago: “Would I had phrases that are not known, utterances that are strange, in new language that has not been used, free from repetition, not an utterance which was spoken by men of old and already grown stale.”

Don't we all! So, in trying to find something new to

say, free from repetition, I decided to go back to the beginning and review what is written in the charter of our Society when it was launched, and there wasn't a long past history of presidential addresses. The chartered mission of the Society is

1. to encourage and integrate research, scholarship, and education in all areas of human genetics,
2. to bring into close contact investigators in the many general fields of research that involve human genetics, and
3. to encourage discourse on applications of human genetics to society at large.

The founders of our Society had very ambitious goals, and continuing to strive toward these goals remains critical for the continued vitality and relevance of our Society. They are, however, goals that cannot be achieved without your help and involvement. I will be giving a report on how well we, the members of the Society, are fulfilling the charge and responsibility laid out by its founders.

Broadly speaking, each component of our mission can be mapped onto three major activities of our Society:

1. to encourage and integrate research, scholarship, and education in all areas of human genetics—that's our journal,
2. to bring into close contact investigators in the many general fields of research that involve human genetics—that's our meeting—and
3. to encourage discourse on applications of human genetics to society at large—that's our Policy and Education Programs, as embodied in the work of our permanent Executive Vice President, our Mentor Network and our Social Issues and Information and Education Committees.

Each of these activities is healthy, but there is much still to be done as we face a changing terrain with major challenges.

First, the journal.

As I am sure you have heard, the entire field of scientific publishing was awoken from its dogmatic slumber by the launch of the Open Access movement. At a meeting of its proponents held in the Spring of 2003, two basic principles were enunciated:

1. the author(s) and copyright holder(s) grant(s) to all users a free, irrevocable, worldwide, perpetual right of access to, and a license to copy, use, distribute, transmit, and display the work publicly, and
2. a complete version of the work and all supplemental materials is deposited immediately upon initial publication in at least one appropriate online repository, to enable open access and unrestricted distribution.

The real push, however, came this summer, when, in mid-July, the House Appropriations Committee approved legislation funding the Department of Health and Human Services for fiscal year 2005. Accompanying the bill was a report, with language by Congressman Istook, ordering the National Institutes of Health to develop a plan placing an electronic copy of any paper resulting from NIH-funded research on PubMed Central, the free digital library maintained by the National Library of Medicine. As proposed, copies would be available 6 months after their journal publication date or—if the publication costs were partially paid for by NIH—the initial manuscript would be available immediately on PubMed. Soon thereafter, an open letter from 25 Nobel laureates was sent to Congress in support of the Open Access Initiative.

The underlying motive of making peer-reviewed scientific literature available electronically, online, without subscription cost is a laudable goal. The devil is, of course, in the details.

One issue is financial. The Open Access Initiative says optimistically, “We can be confident that OA journals are economically sustainable because the true costs of peer review, manuscript preparation, and OA dissemination are considerably lower than the prices we currently pay for subscription-based journals. Moreover, as OA spreads, libraries will realize large savings from the conversion, cancellation, or demise of subscription-based journals.”

Unfortunately, this statement was more a leap of faith than a hard-eyed look at the publishing balance sheet. Our journal already operates successfully with open electronic access 6 months after publication. If even this small delay were eliminated, what would happen to paid subscriptions? We should not lose sight of the fact that the journal is hardly a profit-making operation that is lining the pockets of fat-cat owners and shareholders. What profit we make from the journal helps support the activities of the Society, including the office, the work of our committees, the outreach efforts, and all the rest.

The second issue, of where the online electronic versions are to reside, is also an important one. The Open Access model makes a strong scientific argument that “... barrier-free access to the literature also means barrier-free access for the *software* that facilitates full-text searching, indexing, mining, summarizing, querying, linking, alerting, and other forms of processing and analysis.”

Just as we have open access to genomic data, expression array data, polymorphism data—just so, we need open access to the far more complicated but rich data in the published literature, to allow data mining to support the computer-assisted analysis that can bring novel relationships and correlations to light.

We are concerned that PubMed Central may not be

up to the task of hosting the entire biomedical literature in electronic form as the sole repository. The National Library of Medicine claims that it is. *The American Journal of Human Genetics* and its publisher, the University of Chicago Press, have actually had some experience trying to get PubMed Central to post full-text articles from our journal in a timely manner—our experience did not build confidence. However, that was in the past, and PubMed Central's functioning could always improve.

In a notice published last month, the NIH announced that it was seeking public comments regarding its plans to facilitate open access for publications of work supported by the NIH.

The intent will be for accepted manuscripts to go into PubMed Central for open access as e-publications immediately on acceptance. This version would be replaced by the final copy-edited version from the publisher no more than 6 months after acceptance, with a link to the publisher's Web site. The NIH "trusts" that the up-to-6-month delay will not result in such disruption of current journal revenue streams and business models that unreasonable or disproportionate charges will be made to grantees.

Open access is an issue to which we all—as authors, peer reviewers, and members of a society with a valued journal—must pay attention. The leadership of ASHG is working to reach a reasonable accommodation that will serve the public interest without destroying a whole raft of scholarly journals, including our own.

Now, the meeting. Here, I would like to turn to the main theme of my address and provide some personal answers to the question I posed in the title of my talk: What is special about the "human" in "human genetics"?

Let me remind you of component 2 of the Society's mission:

2. to bring into close contact investigators in the many general fields of research that involve human genetics.

In his 1977 presidential address, Arno Motulsky wrote, "The boundaries of human genetics are indistinct and blurred. It is sometimes said that future progress in the sciences, particularly in those areas of importance for human health and welfare, will increasingly come from interdisciplinary fields by applying concepts and techniques from one field to another area. We must therefore continue to be on the lookout for methods from other fields...."

I have been coming to the meetings of the ASHG for many years because it is the one place where not only are novel and exciting findings in human genetics presented, but the attendees care about what these results mean to patients, their families, society at large, and our understanding of who we are as a species. The scope of the meeting is expansive, not narrow. However, new ar-

reas of research important to human genetics continue to develop and flourish, often through the efforts of many of our members, yet often largely outside the boundaries of our meeting. Are we in a position to share in this information and benefit from it? As a society, we all have much to learn in areas of inquiry, such as genomics, gene-replacement technology, stem-cell research, social science and behavioral research, communication research, functional physiological imaging, real-time microscopy, and translational research in multifactorial risk assessment and intervention. This is not to say that these fields are entirely neglected at the meeting or that the meeting is overly occupied with research that is irrelevant to the field of human genetics. No, my greatest fear is that of parochialism, in which we continue a system that is simply a self-perpetuating status quo. If we get set in our ways, become known for being interested in certain areas and not others with relevance to human genetics, we will attract members with research interests that simply reinforce such conceptions. The result? A self-fulfilling prophecy that narrows our membership base and scientific scope.

The Board of Directors discussed this issue among themselves and with the Program Committee and decided on an experiment that we call "Strategic Outreach." We have convened a group of ASHG members with strong ties to other organizations and fields, to be headed by Dr. Diana Bianchi from your Board of Directors. The Strategic Outreach Committee is charged with organizing a special symposium at the annual meeting, designed to highlight a topic that is pertinent and relevant but unusual compared with what is currently presented and discussed at the meeting. The Strategic Outreach Symposium will be a plenary session and should highlight unusual areas that are off the beaten path. What makes this different from the very valuable interdisciplinary symposia that our members suggest to the Program Committee is that we want the symposium to be primarily, if not exclusively, composed of people from outside our Society, who may never have attended an ASHG meeting and yet are doing research that may prove very important to future progress in our field. Our success in this endeavor will be measured in a number of ways: first, by how successful we are in putting together symposia here that the members find thought provoking and illuminating; second, by whether we encourage attendance at our annual meeting by individuals from these other societies once they encounter the power and beauty of the field of human genetics; and, third, by whether we are asked to participate in other annual meetings to spread the word about human genetics.

Strategic Outreach is doomed to fail, however, without the active involvement of our ASHG membership. Be active—get in touch with the Strategic Outreach group so we can take advantage of your knowledge and

contacts within other societies to help us form partnerships. I set this as a challenge to you, the members, to work with your leadership to make the meeting a place where we continue to emphasize that the “human” in “human genetics” is special, because it requires us to be broad and interdisciplinary if we are going to truly encompass the many ways that genetics is informed by and is applied to human biology, medicine, and society.

How about the third component of our mission?

3. to encourage discourse on applications of human genetics to society at large.

This aspect of our mission was addressed by Lee Rosenberg in his eloquent presidential address in 1980. He chose to talk about the American Society of Human Genetics itself, how it was organized, how it was functioning, who were its members. What I found particularly interesting was that his talk occurred at a pivotal moment in our Society’s history, when we were outgrowing the stage of being a cottage industry run out of the academic Office of the President and of the journal publisher. In a memorably succinct understatement, Lee said, “Our organization is anything but a household word.... [We face a] fundamental problem—namely, that of increasing the likelihood that our opinion will be sought about those matters in the domain of our knowledge and experience.”

He went on to recommend that the ASHG establish a permanent office in Washington, D.C., and join forces with other organizations with clout. The Society took him at his word, although we took our own sweet time in doing it! In 1983, ASHG/GSA established its own independent office in the Washington, D.C., area with its first executive director, Gerry Gurvich, who was then succeeded by Elaine Strass 9 years later. Nineteen years after Lee’s speech, we joined FASEB to add our voice to the strength of all the other members of the Federation. Twenty-one years after Lee’s talk, Joann Boughman began as executive vice president to provide a full-time presence on policy issues in Washington.

Thus, after nearly 20 years of cautious development and maturation, the Society began to carry out some of the recommendations first proposed by Lee Rosenberg.

The final point I would like to make is that the third component of our mission, to encourage discourse on applications of human genetics to society at large, requires an informed, knowledgeable public. Thus, I want to emphasize the role we need to play in education, particularly what we can do to encourage discourse on applications of human genetics to society at large.

A commitment to education is a duty. The public is clearly supportive of research. Consistently, polls show that, when asked if they supported federal funding for research, over 80% said they did, even among those who

scored highly on a questionnaire designed to measure levels of reservation about science and technology.

Consistently over the past three decades, when asked to weigh the benefits and risks of scientific research, between 70% and 80% of the American public felt that benefits outweighed risks.

Yet, in the recent NSF survey of public attitudes toward science and technology, half of all adults expressed an interest in science but did not feel well informed, whereas only 10% were considered attentive (i.e., they felt informed and read at least a monthly magazine relevant to some area of science). Society is clearly in favor of research, yet most of them know very little about what we do and how we do it.

Informing the public is not, however, a hopeless task. I draw your attention to a poll, released in February of last year, of the American public’s knowledge of something very basic: “What is DNA?”

#### **New Poll Shows Dramatic Rise in Americans’ “DNA I.Q.”: Threefold Increase Delights Educators, But Is It Enough?**

Richmond, VA, Feb. 27, 2003—A new Harris poll of a nationally representative sample of 1,031 Americans released today showed that the “DNA I.Q.” of American adults is much higher than expected. The poll found that 60% of U.S. adults got the right answer when asked “What is DNA?” When given the multiple choice question, “What does DNA stand for?” two thirds chose deoxyribonucleic acid. The findings show a dramatic rise in genetics awareness since 1996, when a National Science Foundation survey showed only 21% of adults could define DNA.

I would love to think that the increasing importance and visibility of human genetics is responsible for this rise in what the *Richmond Times Dispatch* called the “DNA I.Q.” Is it, perhaps, the Human Genome Project and all of the attendant publicity? However, according to the Pew Research Center for the People and the Press, as reported in the National Science Foundation Report on Science and Technology: Public Attitudes and Public Understanding, only 16% of the public claimed to be following that story very closely. Thus, there may be other places where people are getting their information. My favorite candidate is the DNA forensics shown on the television show *Crime Scene Investigation*, or *CSI*.

How can we get involved in education of children below the undergraduate level, in grades K–12, where the vast majority of our school-aged children will receive essentially all the classroom instruction in human genetics they will get for the rest of their lives? We hold our regular local high school program that precedes our annual meeting. This all-day workshop is an all-volunteer effort by a very dedicated group of individuals, people like Paula Gregory, Loraine Omen-Gaines, Susanne Haga, for whom education is a passion and a calling.

We also have participated on a regular basis, giving workshops and lectures to teachers at national conferences of the High School Biology Teachers and the National Science Teachers Association. However, we think we can do more; in particular, we can contribute to improving the genetics education. I would like to highlight the “Outreach to Teach” or Mentor Program, which the Society established and is working to expand.

The mentor program, sponsored jointly by The American Society of Human Genetics, The National Human Genome Research Institute, The Genetic Alliance, and the Genetics Society of America, encompasses roughly 1,200 geneticists in the United States, Canada, Mexico, and 37 other countries who have expressed a willingness to participate in local educational efforts, such as DNA Day for the kindergarten-through-the-12th-grade level. We have worked to inform superintendents, science coordinators, and science and biology teachers of the mentor network and educational resources available for use in the classroom. The ASHG organized a meeting last month that brought together representatives of major science teachers’ groups, organizations that provide online educational material for genetics, experts involved in the Biological Science Curriculum Study textbooks, and a number of other interested parties. Loraine Omen-Gaines, of the Information and Education Committee, was the prime mover and organizer. These discussions, which have now just begun and are in their infancy, are designed to establish partnerships between the Society and educators. There are a number of ways we have explored involving our Society in the education effort, including developing a “clearing house” on our Web site for already-existing age-appropriate, well-vetted educational material for our mentors to use when they volunteer in classrooms, joining state and local networks established by the National Science Teachers dedicated to this effort, to even getting involved in consulting with

textbook companies that put biology textbooks into thousands and thousands of classrooms around the country.

This effort is an important part of our mission, but I also recognize that we are a small organization whose members volunteer their time and energy to the cause. We all have full-time jobs already. We have a well-run meeting and a presence in the hearing rooms and offices where important public policy is made because we have wonderful people on our administrative staff who can focus their energy on the Society’s business. The Board of Directors has been very supportive in setting aside a small fund for developing pilot projects in education, as well as for hiring an additional ASHG staff member to spearhead an education initiative. If we are to move out beyond the confines of our meeting and our journal, to encourage discourse on *applications of human genetics to society at large*, as our charter puts it, we need financial resources beyond what we currently can muster. Foundations who might support our efforts will want us to show that the members are willing to pitch in and support our efforts by contributing. I am launching a fund-raising campaign, to form the basis of an effort in the near future to highlight our initiatives in education to potential donors, either individual or corporate. To start this campaign off, I have a pledge from the Board of Directors to kick-start our campaign. My hope is that the membership will join in when we come knocking.

The members of this Society are dedicated to exploring the many ramifications of human genetics research and informing the public about what we do and why.

Our willingness to embrace interdisciplinary approaches and our commitment to have an ongoing dialogue with policy makers and the public is what’s special about the “human” in “human genetics.”

Thank you for your attention, and enjoy the rest of the meeting. I certainly will!