The Editors' Recollections on the Occasion of the 60th Anniversary of The American Journal of Human Genetics

Volume 1, Number 1 of The American Journal of Human Genetics was published in September 1949. The first paper was an 18-page preface to the journal by H. J. Muller, president of The American Society of Human Genetics, entitled "Progress and Prospects in Human Genetics." Charles W. Cotterman served as the first editor, and since that time a dozen other human geneticists have shared that distinction. In recognition of the 60th anniversary of AJHG, recollections of five editors are recorded here.

Arno G. Motulsky: 1970-1975

When I became the editor for the January 1970 issue, taking over from H. Eldon Sutton, our journal had already been published for 22 years. The masthead of The American Journal of Human Genetics indicated that genetics articles from four different areas—medicine, anthropology, psychology, and social sciences-were invited, but few articles from the behavioral and social sciences were submitted.

Editorial policy was not to publish single-case reports. For papers on new statistical methods, we encouraged authors to provide specific applications to real data. Occasional annotations were welcomed. Our rejection rate of scientific articles was around 50%.

Eleven of 359 articles published during my six years as editor received more than 50 citations. Six of these papers dealt with statistical methodology, two with population differences, and three with clinical genetics and genetic counseling.

Even though research in medical and human genetics represents only a small proportion of all biomedical research, there were 12 journals of human and medical genetics in September 1970, including two journals with social and eugenic content—a large number for a small field.

Jim Neel, celebrating the 25th anniversary of The American Society of Human Genetics, using the number of papers presented at the 1974 ASHG meeting, as an indicator of research favored by ASHG members, pointed out that these papers represented only 1-2% of the total content of human and medical genetics. A similar assessment now-35 years later-would be interesting. Does current research cover a larger proportion of our field?

I elaborated on my views of human and medical genetics in the 1971 William Allan Memorial Award Lecture. 1 Among a variety of topics discussed, I expressed the belief that control of common, complex diseases by genetics would not be achievable by 1996. This prediction was expressed while our research team, with future Nobel laureate Joseph Goldstein as a postdoctoral fellow, was working on the genetics of coronary heart disease in the

hyperlipidemias. In another area, my assessment of the ethical problems raised by the then current and future approaches to prevention, treatment, and research in genetic diseases was published in Science² and covered genetic disorders, including birth defects, intrauterine diagnosis, abortion, artificial insemination, and in vitro fertilization.

Jerome Lejeune, in his 1969 Allen Award address, expressed his deeply felt rejection of abortion in a discussion of Down syndrome and other chromosomal aberrations in the 1970 publication.³ His presentation was long remembered by the audience, most of whom probably did not share his views on abortion.

David E. Comings: 1979-1986

At the 1977 San Diego meeting of The American Society of Human Genetics, Arno Motulsky cornered me at the door and asked whether I would consider taking over as editor of The American Journal of Human Genetics. Bill Mellman was having health problems and wanted to pass the responsibilities on to a new editor. After recovering from the shock, I agreed to the great honor of editing this esteemed journal.

Ever since its inception, AJHG featured on the front cover the name "The American Journal of Human Genetics" in black text on a white cloud. This was sometimes referred to as the "chicklets" cover. The chewing gum had to go. I felt my first task was to give our journal a sexier, more modern cover. We combined three figures—the DNA helix, a family pedigree, and $p^2 + 2pq + q^2$ —to illustrate its breadth of coverage from molecular biology to clinical and population genetics. I note that it has since become a ritual for almost every subsequent editor to announce his or her arrival with a redesigned cover.

It is easy for me to remember one of the greatest highlights of my tenure of 1979 to 1986. With the advent of high-throughput DNA sequencing, revolutionary events now seem standard fare for the field of human genetics, but in the late 1970s, the previous decades had seen mostly the explosive effect of chromosome banding. This advance was tapering off by the time I took over the helm, and

DOI 10.1016/j.ajhg.2009.08.008. ©2009 by The American Society of Human Genetics. All rights reserved.

despite the increased resolution that banding provided, the field was still struggling with working out how to identify the chromosomal location of individual human genes. *Drosophila* genetics, with its giant chromosome bands, was still leaving human genetics wallowing in the comparative doldrums. I still remember my personal excitement when I received a call from David Botstein, urging rapid handling of a paper that he and his colleagues were submitting, entitled "Construction of a genetic linkage map in man using restriction fragment length polymorphisms." For those readers interested in the history of our field, I reproduce the following abstract of this paper:

We describe a new basis for the construction of a genetic linkage map of the human genome. The basic principle of the mapping scheme is to develop, by recombinant DNA techniques, random singlecopy DNA probes capable of detecting DNA sequence polymorphisms, when hybridized to restriction digests of an individual's DNA. Each of these probes will define a locus. Loci can be expanded or contracted to include more or less polymorphism by further application of recombinant DNA technology. Suitably polymorphic loci can be tested for linkage relationships in human pedigrees by established methods; and loci can be arranged into linkage groups to form a true genetic map of "DNA marker loci." Pedigrees in which inherited traits are known to be segregating can then be analyzed, making possible the mapping of the gene(s) responsible for the trait with respect to the DNA marker loci, without requiring direct access to a specified gene's DNA. For inherited diseases mapped in this way, linked DNA marker loci can be used predictively for genetic counseling.

I immediately sent the paper for review, urging a quick return. To my astonishment, one of the esteemed reviewers recommended rejection. I passed this on to the authors, using my editorial prerogative to let them know that despite this curmudgeon's opinion, their paper was still accepted. They could add theirs to the long list of groundbreaking ideas, papers, or grants that were initially confronted with critical reviews. For me, the paper was sufficiently exciting to stimulate me to wax poetically in an editorial, calling it "the birth of the age of 'the new genetics,'" and so it was. Mapping the Huntington disease gene was one of the first of many successes with this new technique.

Those of my generation remember the refrain "Old soldiers never die, they just fade away." I end this with a personal note of where this old editor has faded. After 37 years of work, I retired from the City of Hope Medical Center in 2002. One of the first books that I read in my leisure time claimed that mutation rates were too low for evolution to occur, along with other Intelligent Design nonsense. I knew from our own research with microsatellite polymorphisms that the mutation rates in some important gene regulatory segments of DNA could be very rapid. This stimulated me to write a series of rebuttals. Once I

started, it was hard to stop, and two years and 694 pages later, this resulted in a book, entitled *Did Man Create God?*—a review of the interaction of genetics, evolution, brain science, and religion. This was followed by my taking up golf (a sport I had long thought was a waste of time), traveling, bird photography, and, five years after becoming a widower, marriage. Fading into retirement can be totally wonderful. And to quote another famous man, but relative to grant writing, "Free at last, free at last."

Charles J. Epstein: 1987-1993

"Seven Momentous Years." That was the title of the editorial that I wrote in December 1993, in the last issue of *The American Journal of Human Genetics* that I edited.⁵ They were indeed momentous years, and, as I phrased it then, "much has happened—to the journal, to human genetics as a science, to medical genetics as a profession, and to me personally." Rather than my trying to summarize it all here, I invite the reader to take a few minutes to read the editorial.

When I assumed editorship of AIHG in 1988, I immediately decided that if the journal was to take its place among the major publications covering human genetics, it would have to look the part. Therefore, it was reconfigured with a larger page size, new typography, and a glossy cover that was mostly black to show off a new depiction of the molecular structure of DNA. But this was merely a matter of cosmetics; the more important decision to be made was what AJHG should be. There was no question that its primary goal, as it has always been, was to publish highquality research in human genetics. However, perceptions at that time, and even now, about what constituted the "best" journals in which to publish work in the molecular biology area dictated that AJHG would rarely receive what would be considered the must cutting-edge and paradigmshifting articles. By contrast, when it came to statistical and population genetics and related areas, AJHG was the place to publish, and all of the ten most cited AJHG articles, each with over a thousand citations, fall into this category. Highest on the list is the article published by the late Richard Spielman and his colleagues, during my editorship, on the transmission test for linkage disequilibrium.⁶

At the time that I became the editor, *The American Journal of Human Genetics*, as the journal of the world's leading human genetics research organization, was, and still is, unique among all of the journals publishing in the area of human genetics. As such, I believed that it had broader responsibilities beyond publishing outstanding scientific research articles. Therefore, in addition to the traditional book reviews, the Allan Award and Presidential addresses, and the occasional obituary, there could be found newly added invited editorials commenting on articles of particular interest, opinions, reviews, a Human Genetics Education section (complete with its own mortar board logo), statements and committee reports from The American

Society of Human Genetics, and even a 30-year history of the Bar Harbor Short Course in Human Genetics, complete with over 100 of Victor McKusick's inimitable snapshots of many of us when we were much younger. There were also articles that could not be strictly considered as scientific research per se—perhaps the most noted of these being the one by Billings et al.⁷ on genetic discrimination (no. 201 on the list of most cited *AJHG* articles).

With all of this newly broadened content, the Letters to the Editor section became much more lively. The most spirited exchange of letters came after publication of Eric Lander's invited editorial on the forensic applications of DNA typing.⁸ "Spirited" is perhaps too mild a word, since the intensity of the letters from a variety of prosecutors and geneticists necessitated the writing of a "real" editorial⁹ in response to the accusation that the peer-reviewed journal had, in one correspondent's opinion, permitted the publication of an invited editorial that was flawed and potentially injurious. While this contretemps may seem quaint in retrospect, it was deadly (no pun intended) serious back in 1991 when major efforts were being made to use DNA typing for forensic purposes. In one other editorial worthy of note-there were only five in all-I used the "bully pulpit" that editors have to advocate for the then-proposed splitting of genetic counselors from the American Board of Medical Genetics so that the board might be admitted to the American Board of Medical Specialties as a recognized medical specialty. 10 This editorial marked a very traumatic time in the history of organized medical genetics, one from which the profession has fortunately recovered.

At this time, when we are celebrating the 200th anniversary of the birth of Charles Darwin and are thinking a lot about evolution, it is worth recognizing that sciences and organizations, like living organisms, evolve, and human and medical genetics are no exceptions. Human genetics didn't crawl out of the primordial ooze until the nineteenth century and medical genetics until considerably later, and their early progress was slow. However, the evolution of both accelerated greatly after the middle of the 20th century-sometimes with incremental changes, sometimes with saltatory jumps. The American Journal of Human Genetics, which started in 1949, has borne witness to virtually the entire course of this accelerating evolution, and each of its editors has had the opportunity to monitor its progress. In my case, it was the wide-scale introduction of molecular genetics and recombinant DNA technology into human genetics research, just after the invention of PCR, and the maturation of the professional institutions of medical genetics that were in the spotlight. What a wonderful time to have been editor!

Peter H. Byers: 1993-1999

After being chosen to succeed Charlie Epstein as editor of The American Journal of Human Genetics (I would be the fourth editor with University of Washington ties), I visited the University of Chicago Press (UCP) Journals Division office, apparently the first such visit by an editor in their recent memory. The choice of editor had been made at the March 1993 ASHG spring board meeting, I had found a managing editor, Roberta Wilkes, who would stay for my full tenure as editor, and by early June, we were prepared to begin accepting new manuscripts in July. When I walked into the press office, the then-head of the Journals Division, Bob Shirrel, asked me whether I had heard the news about Charlie and showed me a page of The New York Times, bearing Charlie's picture after the attack by the Unabomber. So, rather than the leisurely beginning that we had anticipated, with a slow ramp-up to full speed, all of the editorial responsibilities for AJHG shifted immediately from San Francisco to Seattle. Shelley Diamond, Charlie's managing editor, joined us for about six weeks, primarily to handle revised manuscripts from the preceding weeks. Controlled chaos presided for about two months, as we made a very rapid transition to what soon became a relatively smooth and growing operation.

I had three major objectives when I applied for the position of editor: to bring AJHG into the electronic age, to speed manuscript processing, and to introduce a series of review and assessment papers that highlighted new advances and also commented on current papers. I discovered that the UCP had published The Astrophysical Journal as both a paper and an online effort for the previous couple of years, and they were eager to extend this practice to their portfolio of biological and medical journals. It took a couple of years, but with the hard work of Evan Owens and his informatics group, we made the transition to an electronic journal with the then-unique feature (I think) of rapid electronic publication of both HTML and PDF versions prior to the assembly of the paper journal, now almost universally followed. I think that this process revived interest in publishing in AJHG; our submissions grew from just under 400 to almost 1200 per year by 1999, my last year as editor.

Although this innovation sped manuscript publication, it did little for the hands-on, labor-intensive process of handling the paper that we inherited. It was only toward the end of my tenure that we started the process of electronic submission that was then rapidly implemented by Steve Warren.

Commentary was largely lacking from *AJHG*, with the exception of the rare editorial and occasional letter to the editor prompted by a scientific disagreement. We started a monthly summary of papers that we thought interesting in each issue, a task that fell to our first editorial assistant, who would later become our associate editor, John Ashkenas. John had the rare ability to succinctly summarize the key points of a paper, place it into context, and suggest new directions, all in the space of a couple hundred words. He extended his innovation to commissioning a set of reviews of topics that he and I—mostly John—thought

would be interesting to the human genetics community and to finding people to comment on papers in each edition. John was devoted to this task and worked very well with authors, gently reminding them of deadlines, providing references, and in some cases writing so much of the commentaries that he was invited to join the list of authors.

The process of going from typed submission to published paper was more complex than I had imagined, and we ultimately hired Patty Baskin to direct the technical processes. She, too, stayed with *AJHG* until it moved to Atlanta.

At the beginning of my editorship, papers appeared in the printed issue largely in the order in which they had been accepted for publication. Although this might have increased the chance that a reader would encounter, and perhaps even glance through or read, a paper far from his or her center of interest, it seemed ungainly to me. So we ordered the papers by what seemed an impeccable logic, starting with those that dealt with human genetics at the nucleotide level and ending with those that dealt with it at the population level, followed by more theoretical papers or methods papers. I am not sure that anyone outside of our office realized that this was a conscious decision, but for me it gave structure to the journal contents and organized the diversity of the field.

I had thought that this strategy might put the more influential papers near the top of the table of contents. However, it is clear from review of highly cited papers published during the period of 1994 to 1999 that nothing could be further from the truth. It was the "back of the bus" papers from the methods and population sections that received the most interest, consistent with the recognized principle that successful methods papers are usually highly cited. In addition, this result reflected the rapidly emerging interest in common disorders and the drive to find efficient methods for detecting the underlying genetic contributions. This process bloomed later on, but many of the methodologic strategies that underlay it were published in *AJHG* during that period.

I saw every submission as it came through the door, read every abstract, and read the introduction and discussion of most papers before deciding whether to return a paper immediately or send it out for review. We reviewed about half of the submissions and published just under a quarter. Parsing this task reveals annual numbers of about 1000 first readings, 500 rejections without review, 500 readings of external reviews, 200 postreview rejections, 300 revisions, 50 papers sent out for a second review, 50 subsequently rejected, and about 250 published. At a minimum, I reviewed each accepted manuscript three times. This translates into roughly 10-12 decisions per day during the 200 or so working days at The Journal each year. Such a process draws heavily on the experience and interest of a sole editor, so each edition of AJHG perforce reflects that editor's views. We had relatively few vituperative responses to our decisions, but one paper that I rejected was published in Nature Genetics, perhaps a benefit to both journals.

Although *AJHG* recently moved from the UCP to Cell Press, my experience with the group at the UCP was enormously rewarding. Bob Sherril, Everett Conner, Evan Owens, and Jim Searle (the single sharp-eyed, Chicago style devotee of a copyeditor who was ours alone) welcomed my visits, taught me about the publication business and process, visited us in Seattle, and started the tradition of attending ASHG board meetings. They were committed to the success of *AJHG* and managed the electronic transition to bring us to the forefront of electronic publishing.

When I assumed the job, my closest association with statistical and population genetics was that I could spell the words. Indeed, word was around the community that AJHG would devolve into a clinical journal in no time. My frequent consultant during the first three years, and periodically thereafter, was Lynn Jorde, soon-to-be president-elect of ASHG. I asked Lynn to extend his tenure on the editorial board by three years, and once a month or more, I would send him a paper, read it, try to explain to him what I thought it meant, listen to his assessment, and then, together with him, come to a decision about publication. Lynn had the ability to make me think I could understand these papers and move me over the rough spots when they inevitably appeared. Reading parts of all the papers, physically blue pencilling a number of them, and discussing them with Lynn made this a period of intense learning, during which I distilled my understanding of human and medical genetics. Difficult sometimes but always rewarding, it gave me a deeper sense of what we as human and medical geneticists do and of the important questions that we ask.

The focus of human and medical genetics evolved during the six years that I was editor of our journal, and I think that with the help of our staff, our editorial board, and our reviewers, we helped to both shape and reflect those changes.

Stephen T. Warren: 1999-2005

The period that I was editor was an exciting time for human genetics, with major changes in methodologies and approaches, marked by the declared completion of the human genome sequence. The genome sequence put the field into a fast-forward mode and resulted in a windfall of gene discoveries. This period also saw the dawn of the era of microarrays, and the resulting genome scale data were both exciting from a scientific standpoint and a challenge from an editorial standpoint. How would we as journals manage the massive data sets generated by microarrays? The American Journal of Human Genetics had to adjust its policies regarding access to full data sets. We were one of the first journals to make frequent use of supplemental data and had to work with our publisher, unfamiliar with this aspect of publishing, to develop procedures to house the data and make it available to readers.

The genome era also changed the way that we look for genes. When we took over AJHG, highly penetrant loci influencing disease phenotypes were largely found by linkage analysis. For example, in the January 2000 issue, we published nine papers on linkage, including one single-family report. However, this approach was quickly becoming overtaken by association studies with a focus on complex disease. This was somewhat uncharted territory in terms of evaluation of significance and led to several heated discussions at editorial board meetings. Although the standards were dynamic, we strived to lead the field in terms of requiring correction for multiple testing (commonplace now, but not then) and replication. But it was a learning experience. In hindsight, some of papers that we published then would not even come close to the standards of today—for example, papers of association studies with fewer than 100 cases or controls.

Not only were there scientific revolutions during my tenure as editor, but scientific publishing saw some major changes as well. One that generated very heated debates was the "open access" movement. Although not philosophically opposed to the concept, I strongly disagreed with the initial demands. For example, early on it was proposed that all papers be published without peer review and that readers should be allowed to post comments online as sort of a post hoc peer review. While this may work in some fields, in medically relevant areas I viewed this as potentially dangerous, allowing the pseudoscience views of charlatans to be legitimatized by publishing under our banner. I've seen too many instances of megavitamins being sold as cures for Down syndrome to believe that such a policy would work. The other aspect of open access that was poorly understood by its proponents was the actual cost of running the editorial process. The initial estimates wildly under estimated the cost. While we ran a tight ship at The Journal, our costs would not be covered by charges being discussed. Indeed, the online journals that emerged from this movement, although highly successful from a scientific viewpoint, still require subsidizing grants and gifts to maintain operations. Regardless of these operational disagreements, the concept of open access was something I embraced. I believe that our society can be proud of the fact that in 2002, AJHG was among the very first journals to release its contents freely after six months of the publication date. Seven years later, we still remain ahead of the curve; for example, the current NIH policy requires open access 12 months after publication.

During my tenure as editor, we also finished up the move to fully electronic submissions and reviews, initiated by Peter Byers. This was quite a change—no more faxes and FedExing of envelopes. Thinking back, this move by *The Journal*, saving reams of paper, certainly made it a green journal in more ways than one!

Overall, my time as editor was driven by our quest to improve the "cutting edge" nature of *AJHG*. While *AJHG* has had a long history of publishing very high-quality

papers, it sometimes took quite some time to eventually publish a paper. Publication time became somewhat the coin of the realm among journals publishing the latest and greatest. While electronic publishing certainly made substantial improvement, we also initiated a rapid review track for exceptionally timely papers, often turning around reviews in 24 hours or less. Word got out that AJHG could now compete with any journal in terms of rapid publication time, and we enjoyed a marked increase in the number of "gene discoveries." When we took over AJHG in 1999, only an occasional paper reporting the discovery of a gene associated with a disease found its way into The Journal, but by 2005, each issue usually had at least two such papers. Besides this area, we also tried to be strong stewards of what many saw as the "bread and butter" of The Journal, statistical and population genetics. One of the biggest personal surprises of being editor was my awakened interest in these areas; obviously not as a practitioner but as a reader. All of these changes paid off with annual increases in AJHG's impact factor, rising over two and half full points during my tenure.

The final accomplishment of my time as editor is the introduction of the Cotterman Award in 2000 to highlight the best paper to be published over the past year in *AJHG* with a student or trainee member as first author. I am happy that this tradition has continued under my successor as a small way to foster the careers of our youngest members.

People often ask me whether I am glad that I rotated off being editor and are surprised to learn that, in general, I miss it. It was a singular educational experience for me, and I would highly recommend it. It was an honor and a privilege to be your editor and an experience that I shall forever value.

Cynthia C. Morton: 2006-

The path to my tenure as editor began during a coffee break at a board of directors' meeting of ASHG, held in Honolulu in 2004. Peter Byers and Steve Warren encouraged me to apply for the post, and I was simultaneously enormously flattered and frightened at the prospect. That meeting was five years ago now. I remain flattered, and the fright has dissipated. I have deep gratitude to both of them for sending me on this journey, and to The Society for entrusting me with the care of this precious written record of our field. Without a doubt, being the editor of *The American Journal of Human Genetics* will always be one of the highlights of my career as a human geneticist.

I was very fortunate to have Robin Williamson join me in the office as the deputy editor, and then to add Kathryn Bungartz as science editor. One of the first tasks that Robin and I took on was to give *The Journal* a facelift with a new cover. We endeavored to use the cover as a teachable moment in human genetics, beginning with Mendel's peas, and returned the background color to a shade of

green. Cover illustrations have included images representing color blindness, Hardy-Weinberg equilibrium, copy number variants, and inherited breast cancer, and we invite suggestions and contributions to provide monthly genetics brain teasers. Some new features have been added, including a series of articles titled Perspectives, edited by a former AJHG editor, Arno Motulsky, and short summaries of papers of interest to human geneticists, titled This Month in Genetics, edited by a former AJHG deputy editor, Kate Garber. Most recently, Kate has expanded the summaries to include papers from Genetics and Genetics in Medicine, representing a paper swap with our sister societies, in a section titled This Month in Our Sister Journals. In a further outreach effort to our colleagues, beyond those in our sister societies represented by the paper swap (Genetics Society of America and The American College of Medical Genetics), in 2008 I sought the board of directors' approval to open up the editorial board to human geneticists residing outside of North America. Recognizing the global origin of the submitted and published manuscripts, and the wonderfully global nature of the scientific community, the number of associate editors was increased by three. This addition of associate editors, each serving terms of three years, represented the first increase since 1970, under Arno Motulsky's editorship. As with the new cover, it was time for a change!

No doubt the biggest change that we undertook was a publisher competition that resulted in a transition in January 2008 to Cell Press after a 40-year relationship with the University of Chicago Press (UCP). We certainly learned a lot about our journal through nine publishers' proposals, and we remain grateful for the many new ideas that we received about how to make AJHG even better! With the able assistance of publishing consultant Cara Kaufman, and a task force of Society members, we met in Phoenix prior to the spring 2007 board of directors' meeting and made the business decision to leave UCP. UCP staff, especially Everett Connor and Alec Dinwoodie, had become cherished friends of ASHG over the many years, and the parting was not without remorse for this loss. Nonetheless, it was a decision that I and the members of the task force believed important to move The Journal forward to better reach the global community and to enhance its position financially to support many worthy activities of The Society. The transition to Cell Press went smoothly, with much gratitude due to Robin Williamson and to the superlative efforts of individuals at Cell Press, including especially Keith Wollman, and with appreciation also to Jim Krosschell and Emilie Marcus. The change in publisher meant a change in our editorial processing system from the home-grown system at UCP, known as WPR ("whipper"), to Aries Editorial Manager. We had a few hiccups once the switch was flipped, but this was another change for which the time had really come, and the new editorial processing capabilities were much welcomed. Further developments in manuscript processing took place with this year's migration to the Elsevier Editorial System, and the future will be one of continuous evaluation and improvements to the process. From my perspective, AJHG has benefitted in a number of ways from being a Cell Press journal, supported by the larger infrastructure of Elsevier. A new and enhanced web site was launched and various resources were provided, such as open access to a monthly featured AJHG article as well as an article selected from the Cell Press family of journals. Papers are published ahead of the print version on a weekly basis, and AJHG papers continue to be open access at six months, an atypical arrangement for a commercial publisher but an ASHG core value that was critical in the publisher evaluations. Podcasts by Drs. Williamson and Bungartz now frequent the web site, providing another venue to get the word out about advances in human genetics. As a side note, in a separate effort, all of the back archives of AJHG became available through special efforts of staff at the National Library of Medicine. So, you may now proceed directly after reading these recollections to PubMed Central to download President Muller's inaugural paper in AJHG!

But...what has been going on scientifically in human genetics during this time? AJHG continues to be a highly regarded, if arguably not the most highly coveted, publication for papers concerning statistical genetics methods, with its most frequently cited publication, already mentioned above, being a paper by Richard Spielman and colleagues, about a transmission test for linkage disequilibrium, fondly known as TDT.6 Recognizing this paper is certainly bittersweet at this time because of the recent untimely death of Rich, whose future contributions to AJHG will be sorely missed! During my editorship, the top cited paper is another methods paper, by Shaun Purcell and coauthors, 11 concerning a free, open-source, wholegenome association analysis toolset, designed to perform a range of basic, large-scale analyses in a computationally efficient manner, known as PLINK. Gene discovery papers still figure prominently in AJHG, although citation rates for some of these rarer disorders are often not high, which has an impact on our impact factor. Lastly, it goes without saying that this has really been the time of genome-wide association studies. Although perhaps not the top genetics journal for publishing the most cutting edge GWAS papers, AJHG receives a number of these manuscripts and has been rigorous in its review of them, being at the table with the community of leading investigators trying to sort out the criteria necessary for significance, replication, and validation of the findings.

So, happy 60th birthday, *AJHG*! My wish is that you will go forward in the next 60 years to be so cherished as you have been by the six editors who have written about you here. And, for the *AJHG* editors to come, may you enjoy your editorships as much as we have and welcome the opportunity to serve a society of human geneticists that we have all so loved.

References

- 1. Motulsky, A.G. (1971). The William Allan Memorial Award Lecture. Human and medical genetics: a scientific discipline and an expanding horizon. Am. J. Hum. Genet. 23, 107-123.
- 2. Motulsky, A.G. (1974). Brave New World? Science 185, 653-663.
- 3. Lejeune, J. (1970). The William Allan Memorial Award Lecture. On the nature of men. Am. J. Hum. Genet. 22, 121-128.
- 4. Botstein, D., White, R.L., Skolnick, M., and Davis, R.W. (1980). Construction of a genetic linkage map in man using restriction fragment length polymorphisms. Am. J. Hum. Genet. *32*, 314–331.
- 5. Epstein, C.J. (1993). Seven momentous years. Am. J. Hum. Genet. 53, 1163-1166.
- 6. Spielman, R.S., McGinnis, R.E., and Ewens, W.J. (1993). Transmission test for linkage disequilibrium: The insulin gene

- region and insulin-dependent diabetes mellitus (IDDM). Am. J. Hum. Genet. 52, 506-516.
- 7. Billings, P.R., Kohn, M.A., de Cuevas, M., Beckwith, J., Alper, J.S., and Natowicz, M.R. (1992). Discrimination as a consequence of genetic testing. Am. J. Hum. Genet. 50, 476-482.
- 8. Lander, E.S. (1991). Research on DNA typing catching up with courtroom application. Am. J. Hum. Genet. 48, 819-823.
- 9. Epstein, C.J. (1991). The forensic applications of molecular genetics-the journal's responsibility. Am. J. Hum. Genet. 49,
- 10. Epstein, C.J. (1992). Organized medical genetics at a crossroad. Am. J. Hum. Genet. 51, 231-234.
- 11. Purcell, S., Neale, B., Todd-Brown, K., Thomas, L., Ferreira, M.A., Bender, D., Maller, J., Sklar, P., de Bakker, P.I., Daly, M.J., et al. (2007). PLINK: a tool set for whole-genome association and population-based linkage analyses. Am. J. Hum. Genet. 81, 559-575.