2017 Presidential Address: Checking, Balancing, and Celebrating Diversity: Celebrating Some of the Women Who Paved the Way[1]

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Given that the verbal presentation and slides of the 2017 ASHG presidential address are available on the ASHG website, it seemed appropriate to take the opportunity in this article to provide more detailed information on some of the women—highlighted in the presentation—who have made contributions to the science of human genetics. Among those highlighted, I will focus on the subset of women who I have known personally. Human genetics has a long history of strong female scientists who not only contributed greatly to science but also paved—and smoothed—the way for those who came after. My goal here is to celebrate that legacy by documenting some of it for the younger generations who might not have had direct knowledge of the contributions of these women. The generosity of these women—in their official mentoring of students and postdocs and in their consistent efforts to keep female scientists in the forefront of the field of human genetics—is well worth celebrating!

Human Cytogenetics

Human cytogenetics was a rapidly moving and tremendously exciting field as its technologies evolved—comparable, perhaps, to today's excitement in genomics given the revolution in interrogating genome variation. Many of the field's leaders were women, and of them, Pat Jacobs, Janet Rowley, Dorothy Warburton, and Uta Francke were all former ASHG Allan Award winners who I have been fortunate to know.

Patricia Jacobs

Born in London in 1934, Pat Jacobs moved to Scotland as a child and was educated at the University of St. Andrews in Fife, Scotland, where she earned both BSc and DSc degrees. She came into human genetics through zoology and cytogenetics, and her early human genetics research career focused on population cytogenetics. She continues to conduct research today (in her mid-80s) on both the nature and basis of human chromosomal variation and on population studies of human chromosomal variations. Pat Jacobs described the first chromosomal anomaly characterized in humans with colleague John Strong in 1959[1] as a 47 XXY, already known as Klinefelter syndrome because of the recognition of the symptom cluster associated with this disorder in 1942 by Harry Klinefelter. Her long association with the Medical Research Council's Edinburgh unit led her to value a team approach (including clinicians, epidemiologists, and cytogeneticists) to scientific investigation long before it became as widely used in human genetics as it is today. She was an active leader of human cytogenetics in its early days, when several major discoveries were reported every year for the first decade or so, and remained in the forefront of large-scale investigations of human cytogenetics over her entire career. Her lab made seminal contributions to our understanding of widely disparate parts of this science, including not only cytogenetic anomalies associated with a variety of human diseases[2–4] but also general investigations of the sex chromosomes, including the single best-titled paper[5] ever. She was among the first to tackle larger-scale epidemiological investigations by using cytogenetics[6,7] and did seminal work on many aspects of fragile X syndrome.[8–17]

[1]This article is based on the address given by the author at the meeting of the American Society of Human Genetics (ASHG) on October 17, 2017, in Orlando, FL, USA. The video of the original address can be found at the ASHG website.

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Dr. Jacobs was the first woman to be awarded the Allan Award from the ASHG in 1981, which was also the first and (so far) only year the society had both a female president and a female president-elect (Barbara Bowman and Marge Shaw, respectively).

Dr. Jacobs has won many additional awards over her long career. To elaborate on just a few, she was elected as a fellow of the Royal Society of Edinburgh in 1977, received the University of Hawaii Regent's Medal for Excellence in Research in 1983, was elected as a fellow of the Royal College of Pathologists in 1987, became an elected fellow of the Royal Society in 1993, won the Mauro Baschirotto Award from the European Society of Human Genetics in 1999, was elected as a foreign associate of the National Academy of Sciences in 2009, and was awarded the March of Dimes Prize in Developmental Biology in 2011.

Pat Jacobs was a pioneering scientist in human genetics and was described by her colleagues as vivid, vibrant, genuine, and genuinely fun to work with. One of her former trainees noted that she could have been the originator of "rigor and reproducibility" but that she would have referred to it simply as "doing science." She married Newton Morton, the first Allan Award winner (1962), in 1972 and joined him at the University of Hawaii in creating a unique research environment that attracted scientists from far and wide. Many prominent human geneticists were students and/or postdocs there, but scientists at all levels came for sabbaticals and other extended visits to learn new aspects of human genetics being created there in both human cytogenetics and human population and quantitative genetics. What could be better than doing great science in paradise? Newton Morton once characterized Pat's personality by describing a long, meandering walk that the two of them had taken with Charlie MacLean along volcanic rocks. They had been talking about science the whole way, and after a lengthy side discussion with Charlie, Newton realized not only that they were completely lost but also that they had long since lost Pat. Newton said in describing the incident many years later that although Pat had been angry, only a woman of truly expansive good humor would have still been married to him. I love that story because I have played both the role of Pat and the role of Newton in similar scenarios. Talking about science while stationary has always been much safer for me.

In addition to students and postdocs who they mentored individually, a number of young people, such as Stephanie Sherman, did research with both. Dr. Jacobs continues to conduct research and publish both on her own and with long-time colleagues, including Stephanie Sherman and Terry Hassold, and is currently co-director of research at the Wessex Regional Genetics Laboratory of the University of Southampton.

Janet Rowley

Janet Davison Rowley was born in New York in 1925 and moved to Chicago at 15 to attend a special advanced program through the University of Chicago Laboratory Schools, where she completed her last years of high school. She ultimately earned an undergraduate degree at the University of Chicago and then went to medical school there. Although she had to wait until the second year after she first applied to the medical school to be officially admitted (because they already had the one woman allowed in the medical school class in the first year she applied), she received her MD degree in 1948 and her medical license in 1951. Dr. Rowley began her faculty career at the University of Chicago in 1962, although she worked only part time when her children were young. By 1973 she had developed the ability to identify a translocation between chromosomes 9 and 22 as the cause of the Philadelphia chromosome seen in chronic myelogenous leukemia.[18] Through her continued research and discovery of chromosomal translocations associated with different forms of cancer, Dr. Rowley became convinced that these observations were not mere bystanders, or even a consequence of the cancer, but rather a primary driver of cancer biology. This view was, of course, eventually vindicated, and Dr. Rowley was a hugely influential figure in cancer cytogenetics and cancer biology.

Janet was a colleague and friend at the University of Chicago. Although she was recognized as a force of nature even by the time I arrived there in 1987, she was completely approachable at seminars or journal clubs and, as I was surprised to learn, as the lecturer on cancer genetics in a human genetics course I taught as a young faculty member. I remember being a little nervous when I first met her, but she was so down to earth that it was impossible to be ill at ease. But she really was a force of nature, and it was due in part to her constant pressure that the University of Chicago eventually established their Department of Human Genetics. She was an advocate for quality science and the resources necessary to support it for her entire career, and she was a great friend to new faculty at the University of Chicago. She would always ask, "Do you have everything you need to do your best science?" And she freely shared her own story by emphasizing not her amazingly accelerated undergraduate and medical education but the many years that she worked only part time because of her young children, the slow acceleration of her research program, and her conviction that persistence in quality science was the key to real success—and that real success is centered on the pleasure of learning what you set out to learn and on the journey to that knowledge. She was a generous mentor and extended her mentoring role to many outside her immediate lab. Her trainees are a who's who of cancer genetics and biology, and she knew absolutely everyone and had an amazing ability to connect people in just the right way by recommending the right person to contact if you had a question or wanted to start a project.

Dr. Rowley also won many prestigious awards in addition to her 1991 Allan Award, shared with Alfred Knudson, Jr. She won the Lasker-DeBakey Clinical Medicine Research Award and National Medal of Science in 1998, the Gruber

Prize in Genetics and National Medal of Freedom in 2009, the Pearl Meister Greengard Prize in 2010, and the Japan Prize in 2012. It was not widely known, but Janet Rowley contributed any awarded prize money to the University of Chicago. She said that all of her success was attributable to the education and the opportunities that the university had afforded her and that she really valued the time they gave to build her early career. She did whatever university work she was asked to do but still conducted research nearly to the end of her life. She scored in the top few percentiles on an NIH grant well into her 80s. Life in science is filled with memorable people. Everyone who knew Janet Rowley felt grateful for that privilege.

Dorothy Warburton

Dorothy DeMontmerency Warburton was born in Galt, Ontario, Canada, in 1936 and, focused on the more quantitative aspects of human genetics, earned both her undergraduate (1957) and graduate (1961) degrees at McGill University in Montreal. She became a faculty member at Barnard College of Columbia University and learned human cytogenetics at that time to allow her to further her research interests in the causes of human miscarriage. She recognized early the value of creating a clinical laboratory for conducting human cytogenetic studies and founded the Cytogenetics Laboratory at the NewYork-Presbyterian Hospital and Babies' Hospital (affiliated with the Columbia University College of Physicians and Surgeons) in 1969. She directed the laboratory for 37 years and was associate director until she died. She conducted state-of-the-art research in human cytogenetics from the time she learned it.

Dr. Warburton was outspoken, disarmingly funny, and a terrific dinner partner. I had the good fortune to serve with her on a number of study sections and learned a great deal from her on how to evaluate study designs even for research not immediately in my area. She was also ferocious in the defense of science she believed was worth pursuing. It is easy to find the weaknesses in any grant, and everyone at the table will appreciate your intelligence in recognizing and pointing out those weaknesses. Advocating effectively for a grant requires a different kind of argument and a genuine confidence in your scientific views and your ability to articulate them. I wish it were possible to bottle the kind of wisdom and passion she demonstrated in those meetings and pass them out to new members of study sections.

Dr. Warburton's speech for her 2006 Allan Award is well worth reviewing. Entitling it "Having it All," she used the term to refer to many different aspects of her career and life that she valued highly. She reveled in being able to skate on the edge of several different disciplines—quantitative genetics, genetic epidemiology, and human cytogenetics, for example, were all part of the research she conducted over most of her career. But she loved having both the basic science and translational perspectives in her day-to-day research life as well. Running a clinical laboratory enabled her to accumulate knowledge on unusual patients, which sometimes spurred new research ideas and always stimulated her to learn more for the sake of the patient. She also used the term to note that she felt that she had it all with respect to having, and deeply enjoying having, a family throughout much of her career and, at the same time, a career that was intellectually satisfying to her. Although she valued her career and the opportunities to pursue the science she loved, she noted that she was precluded from becoming tenured for many years because only one member of a married couple could be considered for tenure at Columbia, and when that anachronism was finally ended, she was nevertheless denied tenure. It is still astonishing to me, knowing her research and career, that such an outcome would be possible. It is, perhaps, a sober reminder that improvements are often incremental and sometimes come with lingering headwinds. Her Allan Award presentation also reminds us that our current concerns about the ability of modern data generation to far outstrip our ability to provide confident clinical interpretation of that data are not really new at all. Her well-articulated concerns are difficult to improve upon and would fit in any of the current discussions on that topic.

Dr. Warburton also expressed the hope that the dearth of female Allan Award winners was largely attributable to the tendency of the award to be presented as a lifetime achievement and that as more women spent a lifetime doing human genetics research, more women would win such awards. After Pat Jacobs won the award in 1981, three women received it over the next 10 years, but not a single woman won the award over the 10 years that followed. Over the next 10, there were but two women, Dorothy Warburton in 2006 and Uta Francke in 2012. In the 5 years since 2012, only one woman, Kay Davies, has been an Allan Award winner (2015). Is it really true that fewer than 20% of the scientists worthy of the Allan Award are women? The Curt Stern Award was first awarded in 2001, and over its life, only one woman, Vivian Cheung (2010), has achieved this honor. The statistics for the Curt Stern Award are particularly grim, because to some extent they might presage the male/female ratio of future Allan Award nominees. At this point, I'm afraid these statistics say more about the processes for nominations and awards at ASHG than about the quality of female scientists doing research in human genetics. The ASHG Board of Directors has recently voted to establish a more straightforward nomination procedure that will be available for use in 2018, as well as a larger awards committee to ensure diverse representation and a good quorum for all calls. Dorothy Warburton died in April 2016. I am anxious to see Dr. Warburton's hopes for the representation of women among Allan Award winners—indeed all ASHG award winners—come to fruition.

Uta Francke

Uta Francke is a physician scientist originally from Germany (MD from Munich in 1967) and has been working in the United States since 1970. She completed fellowships in medical genetics and genomics at the University of

California, Los Angeles (UCLA; 1971) and University of California, San Diego (1973) and joined the faculty at Yale's new Department of Human Genetics in 1978. Dr. Francke was also a pioneer in human cytogenetics and used both somatic cell genetics and high-resolution cytogenetics to drive the field into new areas. I had the pleasure of doing a rotation in Dr. Francke's lab as a first-year graduate student in human genetics at Yale. Although I had done some mosquito cytogenetics as an undergraduate and a clinical cytogenetics rotation with Dr. Roy Breg, the Francke lab was pursuing a host of new methods in cytogenetics. There were elaborate setups for making high-resolution chromosome spreads and dozens (hundreds?) of somatic cell hybrids that could be used for mapping genes to chromosomal regions through the clever use of translocations,[19] so as the hybrids shed human chromosomes, higher resolution mapping was possible. Though I had chosen Yale specifically to work with Ken Kidd in quantitative human genetics, the experience in Dr. Francke's lab was memorable. Even though I did not ultimately work in human cytogenetics, I always kept up with Dr. Francke. She was very encouraging throughout my graduate studies at Yale and always made time to talk with me about human genetics research that I wanted to discuss. Even after I finished at Yale, I remember receiving a note in which she reminded me of science we had discussed, and I always caught up with her at ASHG meetings. She would sometimes rent a plane and fly herself to the meeting, which seems like an even better idea these days. After I served with her on a study section that she chaired, I was so impressed with how accurately she summarized the discussions that I've always tried to model her approach when I chair a study section; I am still working on getting that right. I doubt that she ever realized what an important role model she was, but to this day when I get to spend time with human genetics graduate students who are not in my own lab but who want some advice or just to hang out and talk about science, I think of her and her generosity in sharing her enthusiasm for human genetics.

Dr. Francke moved from Yale University to Stanford University, where she spent most of her career and is a professor emerita in genetics and pediatrics. Her innovative cytogenetics led to the precise mapping of hundreds of human and mouse genes, and she created much of the early knowledge on how regions of mouse and human chromosomes share syntenic regions. She contributed to knowledge in many human diseases[20,21] and was among the first to create mouse models of human microdeletion syndromes.[22] Her research was patiently iterative—it went back and forth between different kinds of somatic cell hybrids to very finely map a disease gene or went back and forth between human and mouse models of disease to establish mechanisms of disease at both the mutation and gene levels. And her research was widely recognized for its creativity and impact in both the basic science and clinical arenas. She was elected to the Institute of Medicine (now the National Academy of Medicine) in 1990 and as a fellow in the American Association for the Advancement of Science in 1995. She won the Antione Marfan Award from the National Marfan Association in 1996 and was elected as a fellow of the American Academy of Arts and Sciences in 1996. She was the recipient of the Colonel Harland Sanders Lifetime Achievement Award from the National March of Dimes Birth Defects Foundation in 2001 and of the William Alan Award from the ASHG in 2012. Dr. Francke was a very early and passionate advocate of open science and remains so to this day. She even anticipated in her 1999 ASHG presidential address the growth of large-scale direct-to-consumer genomics and the use of biobanks for genomic research. She was also a vocal supporter of team science and the diversification of human genetics and other graduate education given the diverse skills likely to be needed in science in the future. We are indeed lucky to have such visionary and generous scientists in our society.

Quantitative Human Genetics

Although human cytogenetics was a fertile ground for human genetics research that seemed to disproportionately attract women, quantitative human genetics was from its early days also a draw for a disproportionate number of women. Among the women who helped to drive and popularize this area of science were Jean MacCluer, M. Anne Spence, and Cathy Falk in the US and Françoise Clerget-Darpoux in France. They were greatly aided in this effort by visionary and effective NIH program officers such as Irene Eckstrand, who recognized early that population and quantitative genetics, genetic epidemiology, and statistical genetics would be linchpins for the success of genetic and genomic sciences. Beneficiaries of the environment these pioneers helped to create included Gruber Prize winner (2004) Mary Claire King and Curt Stern Award winner (2010) Vivian Cheung, not to mention all the rest of us.

Jean MacCluer

Jean MacCluer received her PhD in human genetics at the University of Michigan with William Schull. Dr. MacCluer and long-time partner Dr. Bennett Dyke were the first geneticists to be recruited to the Southwest Foundation for Biomedical Research in 1981, and Dr. MacCluer led the population genetics group there until she retired in 2008. Dr. MacCluer's research was meticulously designed to maximize her ability to map and subsequently identify genetic risk factors for diseases of major (and often disproportionate) impact in US minority populations (Mexican Americans and Native Americans), as well as the associated quantitative traits that might be better-powered targets for gene mapping and identification. A testimony to the elegance of the original design of those studies is that many continue today, decades after their origin, under the guidance of one or another of her former trainees.

As noted above, Dr. MacCluer directly mentored many human quantitative geneticists throughout her career,

but she also developed the concept for a genetic analysis workshop that allowed her to contribute to the growth of the entire field. The genetic analysis workshops (GAWs as they are known) began in 1982 with an unusual format: they were actual workshops. Participants had to do at least an "iota" of work on the analysis of datasets that were provided in advance of the workshop. The intent was to deter exclusively theoretical contributions in favor of actual applications of new or existing methods to real (or simulated) data. In either type of data, the intent was to learn what we could about the performance of the methods by scrutinizing the similarities and differences among results from applications of different methods to data. When the data had been simulated, there was an additional element of testing how well the methods recovered information about the generating model, but it was always recognized that simulations are never likely to be as devilishly complex as reality. The proposal for a workshop was first suggested by Dr. MacCluer as she was refereeing an argument between Newton Morton and Robert Elston on the relative merits of their different approaches to segregation analysis. The workshops rapidly became a tradition—20 genetic analysis workshops have been held over the past 35 years.

It is hard to overstate the value of the workshops in creating a more cohesive and interactive community in quantitative human genetics. The activities attracted an international group of participants, and GAWs have been held in the US and Canada, as well as in Europe. At an early stage of the workshops—between the first and second, perhaps, or the second and third—I remember a meeting whose goal was to assess how much participation there might be for the next workshop. Not only was participation well in excess of what Dr. MacCluer had hoped, but Anne Spence, who was tallying the total who said they would be likely to participate and attend, also reported that the headcount of likely participants was exactly evenly split between males and females. This was cause for great celebration among the organizers, who were, at least at that meeting, all women. Over the years, Rich Spielman and Max Bauer were among the workshop leaders with Y chromosomes, but Jean MacCluer, Cathy Falk, and Diane Waggoner did much of the heavy lifting for the organization of early meetings.

Over many years in science, I observed Jean MacCluer to be one of the most effective boosters of young scientists I have ever known. Whether she was recommending someone for a study section or as a reviewer for a manuscript submitted to a prominent journal or recommending that someone attend NIH-sponsored scientific meetings or chair a scientific session, Dr. MacCluer was a selfless promoter of the next generations. But it is hard to thank her enough for the collegial landscape we enjoy in quantitative human genetics. If you do the "Jimmy Stewart – *It's a Wonderful Life*" experiment, it is difficult to imagine another scientist who has had such a positive impact on her community. Human genetics without the environment that Jean MacCluer created and fostered in human quantitative genetics would be a very different, and much poorer, science.

M. Anne Spence

M. Anne Spence earned a BA in biology from Grinnell College in 1966. She completed a PhD at the University of Hawaii (1969) and then did postdoctoral work at the University of North Carolina with Robert Elston. Dr. Spence spent many years as a faculty member at UCLA and then moved to the University of California, Irvine, to continue her research, as well as service in some academic leadership roles. Although Dr. Spence had a long-standing research focus on autism and neurological and neuro-psychiatric disorders, she was a quantitative scientist with extraordinarily broad interests in human genetics. Few human geneticists had such extensive publications both in mathematical and computational genetics and in clinical genetics, particularly in the consideration of bias in probability calculations used for genetic inference and genetic counseling. Dr. Spence directly educated and mentored a long string of distinguished human geneticists and also collaborated widely in the field in ways that were a great benefit to many additional scientists. Clinician collaborators benefited from her clear thinking in probability and the subtle biases that can creep into what otherwise seem like straightforward clinical genetics questions. Collaborators from mathematics and statistics benefited from her encyclopedic knowledge of human genetics and her unerring instinct for the problems that were worthy of their focused attentions.

Dr. Spence received the Woman of Science Award in 1979, and Grinnell College awarded her an honorary degree in 1999 and an alumni award in 1990. She served as a member of the board of directors for the American Board of Medical Genetics and for Grinnell College and received the 2001 Leadership Award from the International Genetic Epidemiology Society. Throughout her career, she was a champion for quantitative science in human genetics, particularly for women in human genetics quantitative sciences. She cajoled, prodded, exhorted, and just plain led everyone around her to be better scientists, to be more engaged in their science and educational activities, and to do what they could to improve their scientific environment both locally and for the broader scientific community.

Cathy Falk

Catherine T. Falk conducted her PhD research with C.C. Li at the University of Pittsburgh, which she finished in 1968, and spent much of her career conducting research in human quantitative genetics at the Lindsley F. Kimball Research Institute at the New York Blood Center. She had extensive collaborations with the population and statistical genetics groups at the Rockefeller University and generally knew all of the population and quantitative geneticists in the New York City area. Courtesy of the field trips that Jeff Powell organized to enable Yale graduate students interested in population and quantitative genetics to attend a monthly lecture in this area of science at

Columbia University in New York City, she was the first woman I ever met who did research in quantitative human genetics. Dr. Falk always attended the lectures and joined the big group who went to dinner at local restaurants after the lectures. She taught me how to use chopsticks and much about linkage and linkage disequilibrium. She was another of the hugely supportive female quantitative human geneticists who nurtured the careers of several generations of the women who followed her.

Dr. Falk published on a wide variety of topics and made contributions to the field of human quantitative genetics on assortative mating[23–25] in linkage mapping and association[26] and was among the first to seriously investigate the use of neural networks in human genetics.[27] Her development of the haplotype relative risk method[28] substantially influenced the field and stimulated many extensions and a great deal of downstream method development, including the eventual development of the transmission/disequilibrium test.

Françoise Clerget-Darpoux

Françoise Clerget-Darpoux earned a MSc in mathematics in 1970 and a PhD in genetics in 1980. She did postdoctoral research with Elliot Gershon at the National Institutes of Mental Health and then accepted a permanent position at the French National Institute of Medical and Health Research (INSERM) in genetic epidemiology in 1982, after which she became research director in 1986. From 2000 to 2009, she headed a program at INSERM in genetic epidemiology and human population structure and from 2010 to 2011 was Directeur de Recherches de Classe Exceptionnelle. She is currently an emeritus research director with INSERM.

Dr. Clerget-Darpoux was a leader in the fields of genetic epidemiology and statistical genetics for her entire career. She published on a wide variety of research topics and made major contributions to our understanding of linkage analysis[29,30] and the characterization of the consequences of linkage disequilibrium in linkage mapping[30,31] and to the development of novel methods for identifying genetic risk factors for disease.[32] She contributed substantively to genetic studies in founder populations[33] and of neuropsychiatric disorders and autoimmune diseases, including multiple sclerosis, celiac disease, and type 1 diabetes, as well as to genetic studies in cardiomyopathy and Alzheimer disease. She and now many of her former trainees are internationally renowned for their rigorous science and elevation of scientific discourse. Over many years and many scientific meetings, I have never observed anyone to behave badly in word or deed in her presence; people are simply too charmed to behave badly. Consequently, meetings hosted by Dr. Clerget-Darpoux in France were the most civilized it has been my pleasure to attend. Even the most passionate differences of scientific opinion can be fruitfully discussed over a fine Bordeaux in France. Dr. Clerget-Darpoux has directly mentored many of the top quantitative human geneticists currently working in France and had a major impact on many more through the courses she taught in genetic epidemiology and statistical genetics. She received the Leadership Award from the International Society of Genetic Epidemiology in 1998 and was awarded the Beauperthuy Prize from the French Académie des Sciences in 2008. She remains an articulate advocate for rigor at all levels of human genetics science and is particularly eloquent in her reminders of how much we do not know about the non-genetic factors contributing to multi-factorial disease, as well as how wrong we might be about the genetic prediction of liability to disease in the absence of this knowledge.

Irene Eckstrand

Irene Eckstrand studied biology at Earlham College and went on to receive a PhD in biology from Wright State University. After conducting research for a number of years at the University of Texas at Austin, Dr. Eckstrand became a program officer at the National Institutes of General Medical Sciences. Dr. Eckstrand recognized early how critical human population and quantitative genetics would be for the nascent field of genomics and how cross-cutting NIH research investments in this field might be. Over many years, she helped to develop NIH meetings around this science and was the program officer overseeing the funding for the GAWs. Her work is emblematic of the contributions that NIH program staff members make to science and our scientific community. For her long-term contributions to population and quantitative genetics, Dr. Eckstrand was elected as a fellow of the American Association for the Advancement of Science in 2014.

Vivian Cheung

Vivian Cheung is a physician scientist who joined the faculty in the Department of Pediatrics at the Children's Hospital of the University of Pennsylvania in 1998 after notably creative discoveries[34,35] at UCLA with Stan Nelson. The Curt Stern Award honors outstanding scientific achievements that have occurred in human genetics during the last 10 years of the award winner's career. Vivian Cheung won the Curt Stern Award in 2010, and the award covers a remarkable period of productive investigation of the genetic basis of gene expression. From early papers offering the first evidence for potential heritability of gene expression phenotypes[36] through subsequent studies characterizing *cis*- and *trans*-expression quantitative trait loci discovered through linkage mapping,[37] Dr. Cheung initiated hugely influential studies that continue to resonate today in recent publications from the Genotype-Tissue Expression Consortium,[38] for example. The delineation of gene expression as heritable, whereby common genetic variants have sufficiently large effects to be detected in even relatively modest sample sizes, has driven a great deal of additional discovery into the contribution of such regulatory variation to common disease, as well as the large-scale mapping of functional elements in which DNA variants can influence gene expression. Dr. Cheung continues to do exciting research, now at the University of Michigan, where she is the Frederick G.L. Huetwell Professor of Pediatrics, a professor of genetics, and a member

of the Howard Hughes Medical Institute. Dr. Cheung, a card-carrying member of the -omics generation, is able to marry large-scale investigations using novel technologies with superb experimental design to create a research portfolio of unusually high impact and remains the sole woman to have received the ASHG Curt Stern Award.

Mary-Claire King

Mary-Claire King was born in Evanston, Illinois, in 1946 and studied mathematics at Carleton College in Minnesota, where she received her BA in mathematics in 1966. She was persuaded to continue her studies in genetics rather than mathematics by Allan Wilson and earned a PhD in genetics from the University of California, Berkeley (UC-Berkeley) in 1973. After conducting postdoctoral research at the University of California, San Francisco, Dr. King joined the faculty at UC-Berkeley and conducted genetic and epidemiology research there from 1976 to 1995. In 1990, Dr. King reported the linkage mapping of a breast-cancer-related gene that was ultimately known as *BRCA1*.[39] Dr. King and colleagues had used a clever approach capitalizing on the likelihood that subjects with a young age of onset of a common disease are more likely to be heavily loaded with genetic liability for that disease. Although the idea used by Dr. King and her colleagues is by now well established in not just linkage studies but also association mapping and the analysis of sequence data, the methods were quite novel at that time.

Dr. King is also world renowned for her service to the Argentinean community in using genetics to identify the children of the "disappeared," more than 50 of whom were returned to their biological families. She has subsequently led humanitarian efforts to use genetics to identify missing people from all over the world. Dr. King continues to lead stellar research in her laboratory at the University of Washington Genome Sciences Center and has been the recipient of many prestigious scientific awards, including the Gruber Prize in Human Genetics in 2004, the Weizmann Award in 2006, the Pearl Meister Greengard Prize in 2010, and the Lasker Award in 2014. A complete listing of her awards would put me over the word limit for this article, and it will come as no surprise to learn that Dr. King is an exceedingly popular invited guest of young scientists from both the basic and clinical areas of human genetics and cancer biology. In these venues, Dr. King is an unusually attentive listener and active communicator, and these young scientists are inevitably elated to have had such a genuine and engaging interaction with a scientist of her caliber. Of course, Dr. King appears to be just as elated about her interactions with young scientists—a reciprocity that is humbling to watch unfold and deserves emulation.

Conclusion

In closing, I want to note explicitly that the few women whose contributions I was able to highlight in this article are but a small drop in a very large bucket and reflect my own idiosyncratic history in the field. These are all women who won major ASHG awards and whose contributions I could more personally articulate because of my interactions with them or whose contributions I personally prize and believe helped to cement a place for women in human quantitative genetics. Many other woman who have contributed hugely to our field were not included simply because I did not know them as well, and still many others who are my contemporaries, or younger, have similarly contributed hugely to our field and benefited in the same way I did from the support of those who came before us. I also freely acknowledge having enjoyed tremendous support and mentoring from a host of male mentors and colleagues over the years, and many of them were also generous mentors to countless other women in human genetics. But given that more than 80% of our society's awards have celebrated deserving male scientists, I am grateful for the opportunity to bring some additional attention to—to truly celebrate—women who have helped to shape our science and the culture of our field. I also want to reiterate my invitation for women who have been made to feel unwelcome and/or poorly supported in any quantitative field to join us in human genetics. We certainly will be working for the foreseeable future to ensure true equity in opportunity across the entire diversity of our membership, and I can promise a genuinely supportive community and spectacular scientific opportunities.

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