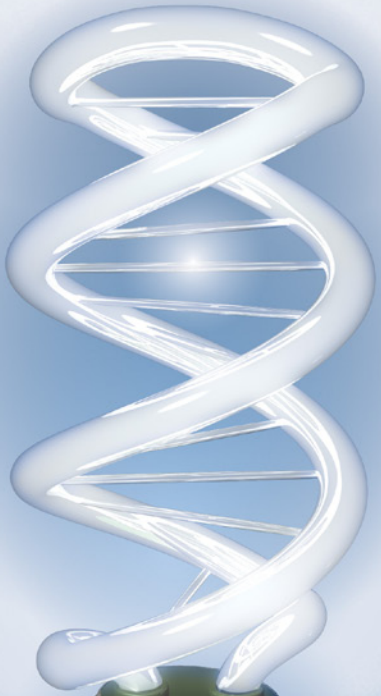
*An NHGRI Symposium*

# A Decade with the Human Genome Sequence

## *Charting a Course for Genomic Medicine*




February 11, 2011
Ruth L. Kirschstein Auditorium, Natcher Conference Center
National Institutes of Health


DEPARTMENT OF HEALTH & HUMAN SERVICES · USA
NATIONAL INSTITUTES OF HEALTH


Welcome to "A Decade with the Human Genome Sequence: Charting a Course for Genomic Medicine," a scientific symposium brought to you by the National Human Genome Research Institute (NHGRI). This symposium aims to provide a glimpse into contemporary genomics research, illustrating how this scientific field is accelerating medical discoveries and how it relates to individuals, communities, and societies. I am delighted that you can join us for this exciting day of talks and discussion.

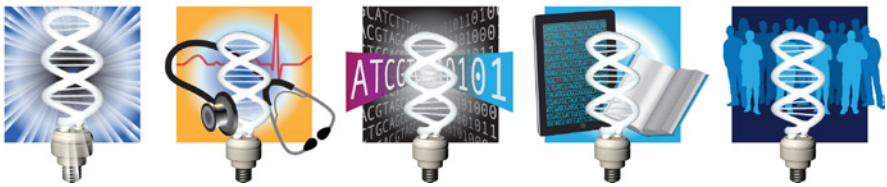
There are a number of historic events surrounding this symposium to which I would like to draw your attention. Last year marked the 20th anniversary of the start of the Human Genome Project. February 15th of this year marks the 10th anniversary of the publication in the journal *Nature* reporting the draft sequence of the human genome produced by the Human Genome Project. Also, this moment in time marks the end of a multi-year strategic planning process for NHGRI, the output of which was published yesterday (February 10th) in *Nature*. This new strategic plan — entitled "Charting a course for genomic medicine from base pairs to bedside" — is a 2011 vision for the future of genomics, which describes how genomics is building on its foundational knowledge of genome structure and function to tackle increasingly complex problems in biomedicine, including those aiming to improve human health. Gazing across the scientific horizon, it is apparent that genomics will be one of the most vital biomedical disciplines of the 21st century.

I hope that today's presentations will illustrate how far the field of genomics has come since the publication of the draft human genome sequence and, more importantly, where the field will take us in the coming decade. Thank you for attending our celebratory symposium, and I hope you enjoy today's program.

Finally, please note that a video archive of the entire symposium will be available at http://genome.gov/Symposium2011 for future viewing.

Sincerely,

Eric Green, M.D., Ph.D.
Director, National Human Genome Research Institute



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| *8:30 A* | *Welcome*<br>Eric Green, M.D., Ph.D.<br>National Human Genome Research Institute |
| *8:45 A* | *Anticipating the Next Decade of the Genome*<br>Francis Collins, M.D., Ph.D.<br>National Institutes of Health |
| *9:15 AM* | *The Human Genome at 10: An Overview*<br>Eric Lander, Ph.D.<br>Broad Institute |
| *10:00 AM* | *Reading Genomes Bit by Bit*<br>Sean Eddy, Ph.D.<br>Howard Hughes Medical Institute |
| *10:30 AM* | *Break* |
| *10:45 AM* | *Sex Chromosome Evolution and Medicine*<br>David Page, M.D.<br>Whitehead Institute |
| *11:15 AM* | *Genes, Genomes, and the Future of Medicine*<br>Richard Lifton, M.D., Ph.D.<br>Yale University |
| *11:45 AM* | *Fevers, Genes, and Targeted Therapies*<br>*Adventures in the Genomics of Inflammation*<br>Dan Kastner, M.D., Ph.D.<br>National Human Genome Research Institute |
| *12:15 PM* | *Lunch* |

*1:00 PM* — *Exploring Your Genetic Blueprint: A Panel Discussion*

**Moderator: Sharon Terry, M.A.**
Genetic Alliance

**Misha Angrist, Ph.D.**
Duke University

**The Honorable Michael Burgess, M.D.**
United States Congress

**Stephen Sherry, Ph.D.**
National Library of Medicine

**Rick Del Sontro**
ClinSeq Participant

**James Watson, Ph.D.**
Cold Spring Harbor Laboratory

*2:00 PM* — *Systematic Surveys of Human Epigenomes*

**Bradley Bernstein, M.D., Ph.D.**
Harvard Medical School

*2:30 PM* — *Functionalizing the Cancer Genome*

**Lynda Chin, M.D.**
Harvard Medical School

*3:00 PM* — *Break*

*3:15 PM* — *Ethical, Legal, and Social Issues in Genomics: Reflecting Back, Planning Ahead*

**Amy McGuire, J.D., Ph.D.**
Baylor College of Medicine

*3:45 PM* — *The Public Place in Personal Genomics*

**Amy Harmon**
New York Times

*4:15 PM* — *The Path Ahead*

**Maynard Olson, Ph.D.**
University of Washington

*4:45 PM* — *Closing Remarks*

**Eric Green, M.D., Ph.D.**
National Human Genome Research Institute

*5:00 PM* — *Adjourn*

**Misha Angrist, Ph.D.**, is Assistant Professor in Duke University's Institute for Genome Sciences & Policy and a Visiting Lecturer at the Sanford School of Public Policy. In 2007 he became the fourth subject in Harvard geneticist George Church's Personal Genome Project and committed to make his genetic and health information public. In 2009 he had his complete genome sequenced. His narrative nonfiction book about these experiences, *Here is a Human Being: At the Dawn of Personal Genomics*, was published recently by HarperCollins.

**Bradley Bernstein, M.D., Ph.D.**, is an Associate Professor at Massachusetts General Hospital, Harvard Medical School, and the Broad Institute in Cambridge, MA. He is also an early career scientist at the Howard Hughes Medical Institute in Chevy Chase, MD. He received an M.D. and Ph.D. from the University of Washington in Seattle, and subsequently completed a clinical pathology residency at Brigham and Women's Hospital in Boston and a postdoctoral fellowship at Harvard University. Dr. Bernstein's laboratory studies chromatin structure and epigenetic regulation of mammalian genomes, with a particular focus on how these entities influence developmental fate decisions and disease. He currently serves as principal investigator (PI) of the National Human Genome Research Institute 'ENCODE' project and a National Institutes of Health Roadmap Epigenome Mapping Center.

**U.S. Congressman Michael Burgess, M.D.**, was first elected to Congress in 2002 after spending nearly three decades practicing medicine in North Texas. Dr. Burgess serves on the prestigious House Energy and Commerce Committee and is the Vice Chairman of the Subcommittee on Health. He is also a member of two other subcommittees: Oversight and Investigations and Energy & Power. He is a member of the bicameral Joint Economic Committee and in 2009, Dr. Burgess founded, and currently serves as Chairman, of the Congressional Health Care Caucus. Because of his medical background, Dr. Burgess has been a strong advocate for health care legislation. Dr. Burgess graduated with both an undergraduate and a Masters degree from North Texas State University, now the University of North Texas. He received his M.D. from the University of Texas Medical School in Houston and completed his residency programs at Parkland Hospital in Dallas. Dr. Burgess also received a Masters degree in Medical Management from the University of Texas at Dallas, and in May of 2009 was awarded an honorary Doctorate of Public Service from the University of North Texas Health Sciences Center.

**Lynda Chin, M.D.**, is a Professor of Dermatology at Harvard Medical School, the Department of Medical Oncology at the Dana-Farber Cancer Institute, and a senior associate member of the Broad Institute in Cambridge, MA. At Dana-Farber and Harvard, Dr. Chin serves as the scientific director of the Belfer Institute for Applied Cancer Science, and co-leads the Dana-Farber/Harvard Cancer Center's Melanoma Program and the Harvard Skin SPORE program. At the Broad Institute, Dr. Chin is PI of The Cancer Genome Atlas (TCGA) Genome Data Analysis Center. Dr. Chin received her M.D. from the Albert Einstein College of Medicine in the Bronx, NY. Dr. Chin's research program focuses on mining and translating complex multi-dimensional genomic data through comparative oncogenomics – the comparison of mouse and human cancers – and the integration with functional genomics to identify novel cancer targets and diagnostic biomarkers.

**Francis Collins, M.D., Ph.D.**, is the 16th Director of the National Institutes of Health. Dr. Collins, a physician-geneticist noted for his landmark discoveries of disease genes and his leadership of the Human Genome Project, served as Director of the National Human Genome Research Institute at NIH from 1993-2008. He is an elected member of the Institute of Medicine and the National Academy of Sciences. He is the 2007 recipient of the Presidential Medal of Freedom and the 2009 National Medal of Science. On April 22, 2010, Dr. Collins was a co-recipient of the Albany Medical Center Prize in Medicine and Biomedical Research.

**Rick Del Sontro** is currently the President of Zippy Shell USA, a nationwide mobile self storage franchise company. He is part of a group of investors that has secured the master franchise rights to Zippy Shell in the United States. Over the past eight years, Mr. Del Sontro has been the Founder and CEO of two non-profit organizations and has been responsible for their dramatic growth. In January of 2002 he became the CEO of the Rainy Day Foundation. During the same time, Mr. Del Sontro served as the Founder and CEO of the Home Downpayment Gift Foundation. Additionally he has ownership interests in a number of businesses ranging from restaurants, to an apparel company and being a partner in Centripetal Capital Partners, a venture capital firm. Mr. Del Sontro is the first participant to have his whole genome sequenced in ClinSeq, a large-scale medical sequencing clinical research pilot study sponsored by the National Human Genome Research Institute at the National Institutes of Health.

**Sean Eddy, Ph.D.**, is a group leader at the Janelia Farm Research Campus, the Howard Hughes Medical Institute's new research center near Washington, DC. His research focuses on deciphering the evolutionary history of life by comparing genomic DNA sequences. He developed computational algorithms and software tools for biological sequence analysis including a package called HMMER. He coauthored the Pfam database of protein domains and the book *Biological Sequence Analysis: Probabilistic Models of Proteins and Nucleic Acids* (Cambridge University Press, 1998).

**Eric Green, M.D., Ph.D.**, is the Director of the National Human Genome Research Institute (NHGRI) at the National Institutes of Health, a position he has held since late 2009. NHGRI is the largest organization in the world dedicated to genomics research. Previously, Dr. Green was the NHGRI Scientific Director, Chief of the NHGRI Genome Technology Branch, and Director of the NIH Intramural Sequencing Center. Prior to becoming NHGRI Director, his own research focused on using large-scale DNA sequencing to address important problems in genomics, genetics, and biomedicine. Dr. Green received his B.S. degree in bacteriology from the University of Wisconsin-Madison, and M.D. and Ph.D. degrees from the Washington University in St. Louis, MO.

**Amy Harmon** is a national correspondent for the New York Times who has reported on the impact of science and technology on American life since 1997. She won the 2008 Pulitzer for explanatory reporting for her series, "The DNA Age," which explored the personal dilemmas posed by new genetic technology and the 2001 Pulitzer for a series on race relations, which she coauthored with a team of reporters. Recently, Ms. Harmon has written about a genetically targeted therapy for melanoma through the lens of the patients and doctors involved in its first clinical trials.

**Dan Kastner, M.D., Ph.D.**, is the Scientific Director of the National Human Genome Research Institute's Division of Intramural Research. His laboratory has studied the genetic basis of inflammation in a number of inherited disorders, including Mendelian diseases such as Familial Mediterranean Fever (FMF) and complex disorders such as Behcet's disease. His group has described new diseases, uncovered novel gene families, proposed a now-commonly used classification of immune-mediated illnesses and pioneered novel therapies. In 2010, he was elected to the National Academy of Sciences.

**Eric Lander, Ph.D.**, is President and Founding Director of the Broad Institute of MIT and Harvard, as well as a Professor of Biology at MIT and Professor of Systems Biology at Harvard Medical School in Boston, MA. In 2008, President Obama appointed Dr. Lander co-chair of the President's Council of Advisors on Science and Technology. Dr. Lander was a principal leader of the Human Genome Project from 1990-2003. A geneticist, molecular biologist and mathematician, his research has focused on population genetics, medical genetics, cancer genomics, mammalian genetics, epigenomics, evolution, and computational biology.

**Richard Lifton, M.D., Ph.D.**, is Chairman of the Department of Genetics, Sterling Professor of Genetics and Internal Medicine, Director of the Yale Center for Human Genetics and Genomics, and investigator at the Howard Hughes Medical Institute at Yale University School of Medicine in New Haven, CT. He received his B.A. from Dartmouth College in Hanover, NH and M.D. and Ph.D. degrees from Stanford University in Palo Alto, CA. Dr. Lifton's laboratory has used human genetics and genomics to identify causes of heart, kidney, and bone disease. By investigating thousands of families from around the world, his group has identified more than 25 human disease genes including key genes and pathways that are critical to the risk of hypertension, stroke, heart attack, and osteoporosis.

**Amy McGuire, J.D., Ph.D.**, is an Associate Professor of Medicine and Medical Ethics and Associate Director of Research for the Center for Medical Ethics and Health Policy at Baylor College of Medicine (BCM) in Houston, TX. Her research focuses on legal and ethical issues in genomics. She is currently studying participant attitudes toward genomic data sharing, investigators' practices and perspectives on the return of genetic research results, ethical issues in human microbiome research, and ethical and policy implications of direct-to-consumer genetic testing. Her research is funded by the National Human Genome Research Institute, the Cancer Prevention and Research Institute of Texas in Austin, and the BCM Clinical and Translational Research Program.

**Maynard Olson, Ph.D.**, is Professor Emeritus of Genome Sciences and Medicine at the University of Washington. Trained as a chemist at California Institute of Technology in Pasadena and Stanford University, Olson pursued a career in genetics starting in the 1970s when he was involved in early applications of recombinant-DNA techniques to study yeast genes. During the 1980s and 1990s, he developed basic techniques for analyzing complex genomes such as large-scale physical mapping based on clone fingerprints, yeast-artificial-chromosome vectors, and the use of sequence-tagged sites as mapping landmarks. More recently, his research has focused on natural genetic variation in bacteria and humans.

**David Page, M.D.**, is Director of the Whitehead Institute, Professor of Biology at the Massachusetts Institute of Technology, and investigator at the Howard Hughes Medical Institute. Beginning in 1979, as a student with David Botstein and Ray White, Dr. Page worked on a forerunner to the Human Genome Project. Immediately after receiving an M.D. from the Harvard-MIT Health Sciences and Technology Program, Dr. Page joined the newly founded Whitehead Institute as the first Whitehead Fellow, and he has remained there since. Dr. Page's laboratory studies the foundations of mammalian reproduction, with particular attention to the X and Y chromosomes, infertility, and the fetal origins of sexual dimorphism in the germline.

**Stephen Sherry, Ph.D.**, is the Reference Collection Section Chief within the Information Engineering Branch of the National Center for Biotechnology Information (NCBI), an intramural component of the National Library of Medicine in Bethesda, MD. Dr. Sherry supervised the design and deployment of several variation archives at NCBI including the dbSNP database of genetic variation, and the dbGaP database of genotypes and phenotypes. His group works on variation-related software resources in forensics (data quality assurance), genome wide association studies (GWAS), whole genome sequence analysis, and polymorphism discovery.

**Sharon Terry, M.A.**, is President and CEO of the Genetic Alliance, a network of 10,000 health related organizations, based in Washington, DC. She is the founding CEO of PXE International, a research advocacy organization for the genetic condition pseudoxanthoma elasticum (PXE). Following the diagnosis of their two children with PXE in 1994, Sharon, a former college chaplain, and her husband, Patrick, founded and built a dynamic organization that enables ethical research and policies, and provides support and information to members and the public. Ms. Terry is also a co-founder of the Genetic Alliance Biobank.

**James Watson, Ph.D.**, was born in Chicago, IL in 1928 and educated at the University of Chicago. In 1953, while at Cambridge University, he and Francis Crick successfully proposed the double helical structure for DNA, described by Sir Peter Medawar as "the greatest achievement of science in the twentieth century." Drs. Watson, Crick, and Maurice Wilkins were awarded the Nobel Prize in Physiology or Medicine in 1962. At Harvard, Dr. Watson commenced a writing career generating *The Molecular Biology of the Gene, The Double Helix, and Avoid Boring People.* He was elected to the National Academy of Sciences in 1962, received the Medal of Freedom in 1977, and was a driving force behind the Human Genome Project that led to his receiving the Copley Medal from the Royal Society in 1993. In addition to many honorary degrees, he received the National Medal of Science in 1997, the City of Philadelphia Liberty Medal in 2000, and the Benjamin Franklin Medal in 2001. Queen Elizabeth II proclaimed him an honorary Knight of the British Empire on January 1, 2002. Dr. Watson is currently Chancellor Emeritus of the Cold Spring Harbor Laboratory.