



Is Personalized Medicine in Your Future?

*Penn Center for the Integration of Genetic
Healthcare Technologies
(Penn CIGHT)
Second Annual Symposium*

*Friday, October 9, 2009
8:00a.m.-3:00p.m.*

*Harold Prince Theatre, Annenberg Center
(3680 Walnut St, Philadelphia)*

The completion of both the Human Genome and the Human HapMap Projects has enabled studies associating human genetic variation with the occurrence of complex diseases such as various cancers, coronary artery disease, and diabetes. The concept that a person's health care provider could potentially tailor care based on a patient's personal genomic profile is a practice that has been referred to as "personalized medicine" (Feero, Guttmacher, and Collins 2008). Although personalized medicine is gaining clinical and policy attention and appears to be technically feasible, major challenges still exist to using this technology to improve health. This symposium will focus on the history of personalized medicine; public attitude, knowledge, and use of personalized medicine; and future implications and strategies for integrating personalized medicine into healthcare.



Center for the Integration of Genetic Healthcare Technologies

- 8:00am to 8:30am: Registration and Continental Breakfast
- 8:30am to 8:45am: Introductions and Welcoming Remarks
Reed Pyeritz, MD, PhD, Penn CIGHT Director
- 8:45am to 9:15am: “Archibald Garrod and the Origins of Personalized Medicine”
Ruth Schwartz Cowan, PhD, History and Sociology of Science, Penn

Personalized medicine – consumer knowledge and attitudes

- 9:15am to 9:35am: “Public attitudes about personalized medicine: Online deliberations in a national sample”
Joseph Cappella, PhD, Annenberg School, Penn
- 9:35am to 9:55am: “Motivations and Perceptions of Early Adopters of Personalized Medicine: Perspectives from Research Participants”
Sarah Gollust, PhD, Leonard Davis Institute, Penn
- 9:55am to 10:15am: “The Navigenics experience: perspectives from consumers and providers”
Elissa Levin, MS, CGC, Navigenics Corporation
- 10:15am to 10:35am: Discussion
- 10:35am to 10:50am: Coffee Break, Prince Lobby (Lower Level)

Examples of Personalized Medicine

- 10:50am to 11:10am: “Pharmacogenetics and Personalized Medicine for Nicotine Addiction”
Caryn Lerman, PhD, Department of Psychiatry, Penn
- 11:10am to 11:30am: “Using somatic genetics to personalize cancer therapy”
Kate Nathanson, MD, Division of Medical Genetics, Penn
- 11:30am to 11:50am: “Genetic Counseling and Testing for Inherited Breast Cancer Risk in African American Women”
Chanita Hughes-Halbert, PhD, Department of Psychiatry, Penn
- 11:50am to 12:10pm: Discussion
- 12:10pm to 1:10pm: Lunch, Prince Lobby (Lower Level)

The future of personalized medicine

- 1:10pm to 1:30pm: “The Intersection of Personalized Medicine and Health Insurance”
I. Steven Udvarhelyi, MD, Independence Blue Cross
- 1:30pm to 2:15pm: “Personalized Medicine: Education of Health Professionals”
Joseph McInerney, MS, CGC, National Coalition for Health Professional Education in Genetics
- 2:15pm to 2:45pm: “Genomics, clinical uncertainty and racial disparities in health care”
Katrina Armstrong, MD, MSCE, General Internal Medicine, Penn
- 2:45pm to 3:00pm: Discussion / Closing Remarks



Reed E. Pyeritz, M.D., Ph.D., F.A.C.P., F.A.C.M.G.
Chief, Division of Medical Genetics
Vice-Chair for Academic Affairs, Department of
Medicine
University of Pennsylvania School of Medicine

In 2001, he became chief of the Division of Medical Genetics at the Hospital of the University of Pennsylvania and Professor of Medicine and Genetics at the University of Pennsylvania. In 2008 he became Vice-chair for Academic Affairs of the Department of Medicine. He is board-certified in internal medicine and clinical genetics, and is an elected member of the American Society of Clinical Investigation and the Association of American Physicians. He has served on the editorial boards of the American Journal of Medical Genetics, the New England Journal of Medicine, Journal of the American Medical Society and Circulation and is a corresponding editor for Human Mutation. Pyeritz is the co-editor of the standard textbook in the field, Principles and Practice of Medical Genetics, the 5th edition of which was published in 2007. In 1991, he was one of the founders of the American College of Medical Genetics and became its 2nd president. He also served as the president of the Association of Professors of Human and Medical Genetics, as an elected member of the Board of Directors of the American Society of Human Genetics, and as a Council Delegate of the American Association for the Advancement of Science. Currently he is the Secretary-Treasurer of the American College of Medical Genetics Foundation.

He has published over 400 scientific articles, reviews and chapters in textbooks. In 2008 he was awarded a \$5.5 million grant from the National Human Genome Research Institute to establish the Center for the Integration of Genetic Healthcare Technologies at the University of Pennsylvania. This multidisciplinary effort brings together faculty and students from the Department of History and Sociology of Science, the Leonard Davis Institute for of Health Economics, the Wharton School, the Nursing School, the Annenberg School for Communication, the Bioethics Institute, the Children's Hospital of Philadelphia, and the School of Medicine. The principal theme that guides the investigations of the Center is uncertainty. As the ability to analyze a person's genes at increasingly refined levels, the resulting information often leads to unclear utility, indecision on the part of both healthcare professionals and patients, and novel ethical dilemmas.



Ruth Schwartz Cowan, Ph.D.

Dr. Ruth Schwartz Cowan is Janice and Julian Bers Professor of the History and Sociology of Science at the University of Pennsylvania. Dr. Cowan received her A.B. in zoology from Barnard College in 1961, her masters in history from UC Berkeley in 1964, and her Ph.D. in the history of science from Johns Hopkins University in 1969. Her current research investigates the intersection between gender studies and the histories of modern science, technology, and medicine. Her most recent book is *Heredity and Hope: The Case for Genetic Screening* (Harvard University Press, 2008). She has been the President of the Society for the History of Technology, the Chair of the National Academy of Science's U.S. National Committee for the International Union for the History and Philosophy of Science, a member of the Smithsonian Council and of the Humanities Visiting Committee of MIT. Dr. Cowan now serves as the Chair of the Community Advisory Board for the BioGene Bank, Feinstein Research Institute, North Shore/Long Island Jewish Health System.



Joseph N. Cappella, Ph.D.

Joseph N. Cappella, Ph.D., is Professor of Communication and holds the Gerald R. Miller Chair at the Annenberg School for Communication at the University of Pennsylvania. Dr. Cappella's research has resulted in more than 100 articles and book chapters and three co-authored books in areas of health and political communication, social interaction, nonverbal behavior, media effects, and statistical methods. The articles have appeared in journals in psychology, communication, health, and politics. His research has been supported by grants from NIMH, NIDA, NSF, NCI, NHGRI, The Twentieth Century Fund, and from the Markle, Ford, Carnegie, Pew, and Robert Wood Johnson foundations. He has served on the editorial boards of 15 different journals. He is a Fellow of the International Communication Association and its past president, a distinguished scholar of the National Communication Association, and recipient of the B. Aubrey Fisher Mentorship Award.



Sarah Gollust, Ph.D.

Sarah Gollust is a postdoctoral researcher in the Robert Wood Johnson Foundation Health & Society Scholars Program and a Senior Fellow at the Leonard Davis Institute of Health Economics at the University of Pennsylvania. She received her Ph.D. in Health Services Organization and Policy from the University of Michigan in 2008. From 2001-2003 she completed a two-year fellowship in bioethics at the National Institutes of Health, where she researched issues related to social and policy implications of new genetic technologies. Her early work on direct-to-consumer marketing of genetic testing was published in the *Journal of the American Medical Association* and *Genetics in Medicine*. Her current research focuses on the news media's influence on public opinion regarding type 2 diabetes, obesity, and health disparities. Her broad research interests include public perceptions of health issues (such as personalized medicine), the role of the media, and ethical issues in public health and health policy.



Elissa Levin, M.S., C.G.C.

Ms. Levin is a board-certified Genetic Counselor whose experience ranges from the research lab to medical centers to the Internet. She has provided genetic counseling in many settings, including directly to consumers via telephone and the Web.

Ms. Levin earned a master of science degree in genetic counseling from Arcadia University in Philadelphia and is certified by the American Board of Genetic Counseling. She began her career focused on the genetics of heart disease, providing genetic counseling and education to families and staff in the cardiology division of the Children's Hospital of Philadelphia. At the University of California, San Francisco Medical Center, Ms. Levin counseled clients of all ages about general and metabolic genetics and coordinated clinical trials for enzyme replacement therapy. Most recently, she helped pioneer DNA Direct, one of the first direct-to-consumer genetic testing companies, providing testing, education and counseling services for specific medical conditions. Ms. Levin's broader efforts to improve health care by setting high standards for providing reliable, quality genetics services through the Internet have led to nationwide lectures, workshops and training sessions.



Caryn Lerman, Ph.D.

Caryn Lerman, PhD, is Mary W. Calkins Professor in the Department of Psychiatry and the Annenberg Public Policy Center. She also is the Deputy Director of the Abramson Cancer Center. Her research focuses on the pharmacogenetics research on nicotine dependence treatment. Dr. Lerman is the Principal Investigator (PI) of an NIH Transdisciplinary Tobacco Use Research Center, Co-PI of an NIH Center of Excellence in Cancer Communication Research, as well as PI for grants supporting pharmacogenetic investigations of nicotine-dependence treatment, medication development, and neuroimaging research. She has over 275 peer-reviewed publications in these areas. Dr. Lerman has been a member of the NCI Board of Scientific Advisors and is a current member of the National Human Genome Research Institute Advisory Council. She is the recipient of numerous awards including the American Psychological Association Award for Outstanding Contributions to Health Psychology, the American Society of Preventive Oncology Cullen Award for Contributions to Tobacco Research, and the Alton Ochsner Award for Research Relating Tobacco and Health.



Katherine Nathanson, M.D., F.A.C.M.G., F.A.C.P.

Katherine L. Nathanson, MD, is an Assistant Professor of Medicine at the University of Pennsylvania. Her research focuses on the genetics of human cancer, both germline changes which confer susceptibility to cancer and somatic genetic changes associated with outcome. Dr. Nathanson is the PI of federally funded grants investigating the genetics of breast and testicular germ cell tumors for which she identified most significant susceptibility gene, as well as the somatic genetics of melanoma and renal cancers as determinants of response to therapies. Dr. Nathanson has been the recipient of the Maesey Research Fellowship Award and was elected to the American Society of Clinical Investigation in 2009.



Chanita Hughes Halbert, Ph.D.

Chanita Hughes Halbert, Ph.D., is an Associate Professor and Director of the Center for Community-Based Research and Health Disparities in the Department of Psychiatry at the University of Pennsylvania. The goal of Dr. Hughes-Halbert's research program is to identify sociocultural, psychological, and environmental determinants of cancer disparities and to translate this information into interventions to reduce cancer disparities in African Americans. Her research is supported by the National Human Genome Research Institute, the National Center on Minority Health and Health Disparities, and the National Cancer Institute. Dr. Hughes-Halbert is nationally recognized for her research on genetic counseling and testing for inherited breast cancer risk in African Americans and has published widely on minority participation in cancer genetics research, responses to genetic risk information in racially diverse populations, as well as the contribution of cultural beliefs and values to outcomes for cancer prevention and control.



I. Steven Udvarhelyi, M.D.

Dr. Udvarhelyi is Senior Vice President and Chief Medical Officer for Independence Blue Cross (IBC). In this role, Dr. Udvarhelyi has overall responsibility for medical management programs and policies, provider contracting and provider relations, pharmacy services, and informatics. Informatics comprises analytic and reporting support for all functional areas, and includes customer reporting. Pharmacy services include IBC's pharmacy benefit management subsidiaries, FutureScripts and FutureScripts Secure.

Dr. Udvarhelyi is a board certified internist and has almost 20 years of experience in the managed care industry. He received an A.B. degree from Harvard College, an M.D. degree from the Johns Hopkins University School of Medicine, and a Master of Science degree in Health Services Administration from the Harvard School of Public Health.

Prior to his career in the managed care and insurance industry, Dr. Udvarhelyi was a faculty member at Harvard Medical School and has published numerous articles on quality in health care.

He currently serves on the Board of Directors of the National Committee for Quality Assurance (NCQA); the National Council of Physician Executives of the Blue Cross Blue Shield Association, and on the Chief Medical Officers Committee of America’s Health Insurance Plans (AHIP). He is also a member of the Institute of Medicine (IOM) Roundtable on Evidence-Based Medicine and the IOM Committee on Comparative Effectiveness Research Priorities; and he has served on other IOM committees in the past.



Joseph McInerney, M.S., C.G.C.

Joseph McInerney, M.S., C.G.C., is the Executive Director of the National Coalition for Health Professional Education in Genetics (NCHPEG). Established in 1996 by the American Medical Association, the American Nurses Association, and the National Human Genome Research Institute, NCHPEG is an “organization of organizations”

committed to a national effort to promote health professional education and access to information about advances in human genetics. NCHPEG works to integrate genetics content into the knowledge base of health professionals and students of the health professions by developing educational tools and information resources that are accessible and useful to the target audience. Trained as a genetic counselor, Mr. McInerney has been involved in genetics education for more than three decades. For many years he worked at the Biological Sciences Curriculum Study, and directed the program from 1985-99.



Katrina Armstrong, M.D., M.S.C.E.

Dr. Armstrong is Chief of the Division of General Internal Medicine, Professor of Medicine, and co-Director of the Robert Wood Johnson Clinical Scholars Program at the University of Pennsylvania. She is also Senior Fellow and Director of Research at the Leonard Davis Institute of Health Economics, Senior Scholar in the Center for Clinical Epidemiology and Biostatistics and the Associate Director of the Abramson Cancer

Center. Dr. Armstrong was the recipient of the 2009 American Federation for Medical Research Outstanding Investigator Award and was elected to the American Society of Clinical Investigation in 2006. Dr. Armstrong's research seeks to elucidate the complex relationship among the social environment, health care, and cancer outcomes. Her federally funded research program has concentrated on several areas of critical policy importance including genetic testing for cancer susceptibility and racial disparities in cancer outcomes. Her current projects explore the relationship between segregation and prostate cancer outcomes and the contribution of health care related distrust to health care cost and quality. Dr. Armstrong currently serves as Director of the Training Core at Penn CIGHT.

About Penn CIGHT:

The University of Pennsylvania Center for the Integration of Genetic Healthcare Technologies (Penn CIGHT) is one of six national Centers of Excellence for research on the Ethical, Legal, and Social Implications (ELSI) of genetics and the human genome. The goal of the Penn CIGHT is to develop tools that will help consumers, professionals, policy makers and insurers understand and cope with the scope of certainty and uncertainty that the application of genetic technologies engender. The work of the center is conducted by teams of experts representing a broad range of disciplines, such as medicine, bioethics, law, behavioral and social sciences, public policy, economics, and research in genetics, genomics, and health services. The Center is directed by Dr. Reed Pyeritz and Barbara Bernhardt.

Ongoing Projects at Penn CIGHT:

- **Historical, Ethnographic and Public Opinion Studies of Prenatal Screening and Risk Assessment:** an historical and sociological analysis of the ethical, legal, and social consequences of prenatal testing for cystic fibrosis and Down syndrome, led by Dr. Ruth Schwartz Cowan.
- **Utilization and Impact of a New Diagnostic Test for Hereditary Hemorrhagic Telangiectasia (HHT):** an assessment from the perspectives of patients, providers and third party payers of the utility of a genetic test to identify the likelihood of future disease susceptibility, using HHT, a single gene disorder, as an example, led by Barbara Bernhardt, Clinical Associate Professor of Medicine.
- **Long-term Behavioral Impact of Genetic Counseling and Testing for BRCA1/2:** an evaluation of the long-term psychological, social, and medical effects of genetic testing and counseling for breast cancer on African American women and their families, led by Dr. Chanita Hughes-Halbert, Associate Professor of Psychiatry.
- **Understanding the Meaning and Impact of Genome-wide, Array-based Testing for Children and Families:** An exploration of the implications of receiving array-based cytogenomic genetic test results and the uncertainty that the results create for patients, their families, and their healthcare providers. This project is led by Dr. Marian Reiff.
- **Assessing Attitudes about the Utility of Personal Genomics:** A collaboration between researchers at the University of Pennsylvania and the Coriell Institute's Personalized Medicine Collaborative (CPMC) to evaluate the public's perceptions of and responses to personalized risk assessment. Investigators on this project include Barbara Bernhardt, Co-Director of Penn CIGHT; Dr. Sarah Gollust, Postdoctoral Researcher in the Robert Wood Johnson Foundation Health & Society Scholars Program; and Erynn Gordon, Director of Genetic Counseling at Coriell Institute.

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