



NEWSLETTER
Spring 2013

A Letter from Center Director Kathryn A. Phillips, PhD

Dear Colleagues,

Spring is a time of growth and change, and so it seems fitting that the TRANSPERS Center is also experiencing exciting new advancement and expansion this season.

We have many new developments from the field to share, including news of new publications and projects that will advance the science of personalized medicine and the work of our Center. Our new funding from NHGRI in the field of Whole Genome Sequencing shows that TRANSPERS continues to grow and meet the ever-dynamic changes and needs for new research in the field.

As always, we welcome your thoughts, comments, and ideas for collaboration.

Best,

Kathryn Phillips, PhD
TRANSPERS Center Director

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NIH Common Fund Conference on Economics of Personalized Medicine Inspires New TRANSPERS Publication

A new **TRANSPERS publication is in press** and will be featured in the *Forum for Health Economics and Policy*: **Phillips, K.A., Sakowski, J.A., Liang, S.Y., and Ponce, N.A. Economic Perspectives on Personalized Health Care and Prevention, 2013.**

This publication arose out of the NIH Health Economics Common Fund's conference entitled "Economics of Personalized Health Care & Prevention" held in July 2012. The conference brought together health economists and researchers specializing in personalized health care and prevention. The purpose was to discuss and provide clarity on the current state of the field of personalized medicine, and to highlight current gaps in knowledge. Six presentations were given – including one by **TRANSPERS Director Kathryn Phillips** – followed by discussion and closing statements by invited participants.

Dr. Phillips's presentation was the inspiration for this new publication in the *Forum for Health Economics and Policy*. This publication provides an overview of economic evaluation of personalized medicine, focusing particularly on the use of cost-effectiveness analysis and other methods of valuation, drawing on insights from work conducted by the TRANSPERS Center. The paper also discusses six areas for future research:

1. Developing and Applying Multiple Methods of Assessing Value
2. Identifying Key Factors in Determining the Value of Personalized Medicine
3. Using Real World Perspectives in Economic Analyses
4. Considering Patient Heterogeneity and Diverse Populations in Economic Analyses
5. Preparing for Upcoming Challenges of Assessing Value of Emerging Technologies
6. Incorporating Behavioral Economics into Value Assessments

To request a copy of the article, please contact Claire Richardson, Program Manager for the TRANSPERS Center, via [email](#).

New Guidelines on Incidental Findings and TRANSPERS Research Highlighted at the American College of Medical Genetics Annual Meeting



Source: www.acmg.net

March 2013's ACMG Annual Clinical Genetics Meeting in Phoenix was an exciting event in the world of exome and genome sequencing (WES/WGS).

Dr. Phillips presented at the ACMG closing plenary session. Her talk focused on the present and future of reimbursement for Genomic Medicine, with a special presentation entitled: *"Clinical and Laboratory Genomics: What to expect for coverage, reimbursement, and policy decisions by payers."*

TRANSPERS collaborator and Principal Investigator of the [MedSeq™ Project](#) Dr. Robert Green was co-chair of the ACMG Working Group and lead author of the highly-anticipated report released at the meeting: **"Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing."**

Incidental findings, or secondary findings that are revealed through the testing process but are not the actual reason for the testing itself, are a prominent issue in WES/WGS. The ACMG recommendations put forward a minimum list of findings recommended to be returned by laboratories to clinicians on conditions, genes, and variants whenever clinical sequencing is performed. This is the first time recommendations for handling incidental findings in the clinical setting have been put forth related to WES/WGS.

In a [recent press release from Brigham and Women's Hospital](#), Dr. Green said:

"As clinical sequencing becomes more widespread, laboratories are looking for guidance on how and what should be communicated to clinicians when results are analyzed. These recommendations will allow a small percentage of families to learn unexpected but potentially life-saving information about an illness they may have never suspected they were at risk for."

Click [HERE](#) to find out more information about the ACMG recommendations and [HERE](#) for the ACMG conference.

Kathryn Phillips Leads National Study of Benefit/Risk in Emergent Whole Genome Sequencing

By David Jacobson, web editor, UCSF School of Pharmacy
Article first appeared on pharmacy.ucsf.edu on March 11, 2013

Improving technologies are rapidly cutting the cost of whole genome sequencing, a process that reveals the complete library of a patient's genetic information. Indeed, the era of the \$1,000 genome—a catchphrase for the test's relative affordability—appears imminent.

But will the wider application of this encyclopedic option in personalized medicine help patients and health care providers prevent and more effectively treat diseases, or will it open a Pandora's Box of confusion, fears, and costly, unnecessary treatments?



UCSF School of Pharmacy faculty member Kathryn Phillips, PhD, will lead the first national study to analyze how physicians and patients in the general population, as well as those given whole genome sequencing results in a clinical trial, evaluate the benefits and risks posed by this profusion of genetic information. The project will address questions such as:

- How much do patients want to know?
- How do patients and physicians assess the significance and usefulness of these tests' myriad potential findings?
- Which findings call for medical intervention versus monitoring?
- What about likely future conditions that currently cannot be treated?

The four-year, \$2.4 million project, "Benefit-Risk Tradeoffs for Whole Genome Sequencing," recently funded by the National Human Genome Research Institute (NHGRI), will also be the first to systematically examine the overall implications of such testing for the health care system and for society by considering, for example:

- When should complete genome sequencing be recommended by health care providers and covered by insurers as clinically useful?
- Will the economic value of preventing disease or more effectively targeting treatments outweigh the costs of the initial whole genome sequencing testing, plus additional testing and treatments its results may generate?
- How can whole genome sequencing findings be most appropriately and effectively applied?

A health economist and founding director of the first-of-its-kind UCSF [Center for Translational and Policy Research on Personalized Medicine](#) (TRANSPERS) in the School's [Department of Clinical Pharmacy](#), Phillips and her Center colleagues have spent recent years establishing the evidence base for evaluating existing condition- or treatment-specific personalized medicine.

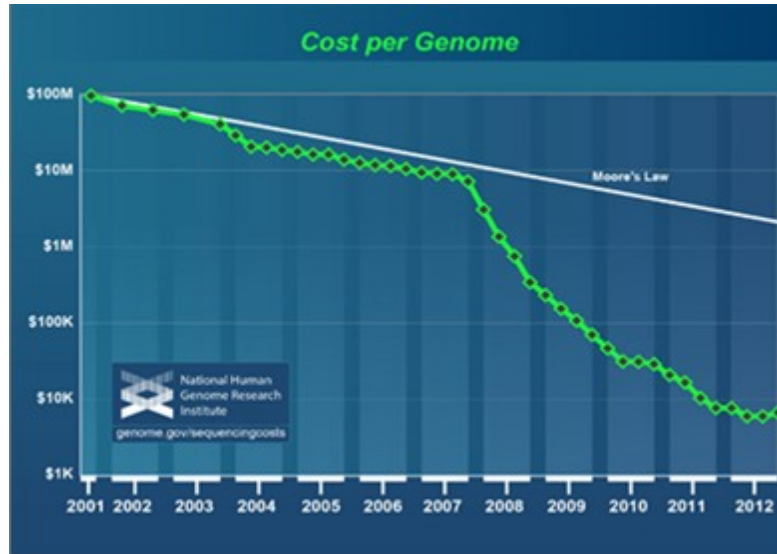


Image credit: © <http://www.genome.gov/sequencingcosts/>

For example, TRANSPERS researchers have studied patient and physician preferences and uses of testing that matches genetic sub-types in certain breast and colorectal cancers with supposedly more effective treatments or interventions, and how such testing is—or is not—consistently translated into clinical practice, insurance coverage, cost savings, and health policies.

The new project will build on the [Medical Sequencing Project](#) (or MedSeq) led by Harvard Medical School researchers, in which physicians and patients are being randomly assigned to two groups: Half will receive “clinically meaningful information” drawn from whole genome sequencing analysis and interpretation; the rest will receive standard medical care without complete genetic mapping. The goal of MedSeq is to assess the understanding, behavior, medical consequences, and health care costs associated with such information.

The Benefit-Risk project will conduct additional preference surveys of MedSeq patients and physicians (including before and after they receive sequencing findings), as well as for a national sample, broadening the scope of the findings and examining diverse populations.

The TRANSPERS-based study will also analyze how whole genome sequencing tradeoffs are weighed and what evidence will be used by health insurers making coverage decisions and by clinicians developing guidelines for use of the testing.

In addition, the work will identify data needed to assess the value of whole genome sequencing. Working with colleagues from several institutions, Phillips will examine how to balance the benefits and costs of this new technology so that it is both efficient and equitable in its use.

As the NHGRI’s director told the Wall Street Journal last year: “We can sequence the genome for dirt cheap, but we don’t know how to deal with the data. We’ve got to work on that.” Phillips’ project will be doing just that.

TRANSPERS Welcomes New Executive Director, Heidi Auman, PhD



Heidi joins UCSF from the Canary Foundation where she was the Scientific Program Manager. Canary Foundation is the world's first non-profit organization dedicated solely to the funding, discovery and development of tests for early cancer detection. Heidi was previously a Postdoctoral Research Fellow at New York University School of Medicine's Skirball Institute of Biomolecular Medicine in New York City, where her research focused on studying the genetics and cell biology of cardiovascular development and disease. During her doctoral studies, she was awarded a Christine Mirzayan Science and Technology Policy Fellowship at the National Academies in Washington, DC. There she worked on issues at the intersection of policymakers, scientists, and the public with the Committee on Science, Engineering, and Public Policy. She earned her Ph.D., M.S., and M.Phil. in Molecular, Cellular and Developmental Biology at Yale University.

Our previous Executive Director, Julie Sakowski, has relocated with her family to Chapel Hill, North Carolina. Please help us wish Julie well and welcome Heidi to our team!

TRANSPERS Research Highlighted at Scripps Symposium

TRANSPERS research was highlighted at the Future of Genomic Medicine conference sponsored by the Scripps Translational Science Institute Symposium in March in San Diego. This annual conference brings together the leading experts in genomic medicine with human geneticists, scientists, physicians and health-care professionals of all disciplines. This year's event included a dynamic presentation, "Toward Precision Medicine," by **UCSF's Chancellor, Dr. Susan Desmond-Hellmann**, highlighting exciting new advances in research and medicine at UCSF.

TRANSPERS' contribution to the field of personalized medicine and cancer was highlighted when two key Center publications - on Pharmacogenomics ([Phillips et al. 2001](#)) and Genomic Testing and Therapies for Breast Cancer ([Haas et al. 2011](#)) - were cited by speakers.