

Center for Translational and Policy Research on Personalized Medicine

NEWS

NEWSLETTER
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A Letter from Center Director Kathryn A. Phillips, PhD

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Dear Colleagues,

We at TRANSPERS are thrilled to collaborate with and glean insights from our colleagues all around the world. Most recently, two TRANSPERS members were honored to participate on a panel with Dr. Francis Collins, NIH Director. On the global level, below we describe how TRANSPERS researchers are participating in international activities.

This kind of cross-national communication and collaboration is one of the hallmarks of TRANSPERS, where we strive to foster an open dialogue and share perspectives as we seek to help move scientific discoveries from the bench and bedside to improve population outcomes and health policies.

Best,

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NIH Director Collins Speaks on Panel with TRANSPERS Members



Photo credit: Lisa Woldin

Shown in the photo (left to right) are Kathryn Phillips, Val Sheffield, Miriam Kupperman, and Francis Collins. Not shown are Robert Nussbaum, Greg Barsh, and Kate Rauen.

TRANSPERS director Kathryn Phillips and TRANSPERS collaborator Miriam Kupperman (UCSF) participated on a panel with **National Institutes of Health Director Francis Collins** during a UCSF symposium to honor the late Charles Epstein in early October.

The overarching theme of the panel discussion was **establishing pathways for genetic discoveries to more readily move into clinical settings**. This neatly coincides with TRANSPERS' main goal: to advance the translational continuum for personalized medicine to ensure that new personalized medicine technologies can be used beneficially and efficiently in clinical care.

Key takeaway points from the panel discussion include that whole genome sequencing (WGS) is expected to be commonplace in cancer within the next five years; that the NIH test registry will be up and running by the end of the year and that gene patenting issues will only get more complicated as WGS becomes more widely practiced.

Leading International Health Policy Expert Robyn Ward at TRANSPERS

Australian health policy expert Dr. Robyn Ward visited TRANSPERS in October to talk about health technology assessments in Australia, focusing on cancer co-dependent technologies for breast and colorectal cancer.

Dr. Ward's talk focused on health technology assessment (HTA) challenges for co-dependent technologies in personalized medicine. Although she described Australia's unique HTA requirements for drugs and diagnostics she highlighted four challenges for personalized medicine technologies that can be considered global: oversimplification of molecular biology, diagnostic inaccuracy, real world use and burden of statistical proof.

Australia is a leader in the use of rigorous, evidence-based assessments and in the integration of stakeholder interests. The audience was excited about Dr. Ward's insights into Australian processes on everything from the uses of whole genome sequencing to policies regarding adoption of health technologies.

TRANSPERS' International Impact



TRANSPERS' reach extends beyond national and even continental borders. TRANSPERS collaborators continue to travel overseas to disseminate research and the Center continues to bring in international speakers to broaden our perspective.

TRANSPERS director **Kathryn Phillips** has traveled to Australia twice this year, most recently as the highlighted international plenary speaker and session chair for a conference sponsored by Australian health care agencies on "**Health Care Technology Assessment for Future Generations.**" Australia is a leader in the use of rigorous,

evidence based assessments and in the integration of stakeholder interests across academia, industry, and government - and many issues they face parallel those in the US.

Another TRANSPERS collaborator, **Jennifer Haas** (Harvard), recently presented at **The International Society for Quality in Healthcare conference in Hong Kong**, on the Impact of Gene Expression Profile Testing on Chemotherapy Use, Costs, and Treatment-Related Serious Adverse Effects for Women with Breast Cancer. That study provides some of the first evidence about the value of GEP testing for women with early stage breast cancer in clinical practice, and demonstrates that GEP testing is being used to tailor decision-making about the use of adjuvant chemotherapy.

Robert Nussbaum wins Zülch Prize

Parkinson's disease discoveries by TRANSPERS Board Member Robert Nussbaum, MD, a leading human geneticist and neuroscientist at UCSF, have led to his being named the recipient of the **2011 Klaus Joachim Zülch Neuroscience Prize by the Gertrud Reemtsma Foundation in Germany.**



Nussbaum and colleagues determined that a mutant form of a protein called alpha synuclein can cause a rare, inherited form of Parkinson's. Soon afterward, the same protein was found to accumulate abnormally in the brains of patients with the common, sporadically occurring form of the disease. Nussbaum's studies have led to a realization that the neurological symptoms of tremor and rigidity seen in Parkinson's are late-stage impacts of a disease that affects not only the brain, but also the nervous system throughout the body. Parkinson's may begin decades before the most familiar neurological symptoms become evident, Nussbaum and others have discovered.

The Zülch Prize has been awarded for outstanding achievements in basic neurological research every year since 1990. The prize is endowed with 50,000 Euro.

TRANSPERS Collaborators Publish High-Impact Paper

It's not every day that researchers find out about the direct impact of their work on policy.

Such was the case when TRANSPERS collaborator Uri Ladabaum (Stanford University) found out that as a result of a paper on which he was lead author, a hospital in Boise, Idaho would be changing its policy to test all colon tumors in order to identify families with the heritable Lynch Syndrome. Approximately three to five percent of colorectal tumors are caused by Lynch Syndrome, which greatly increases the odds of colon and other cancers in a person's lifetime.

The paper, **Strategies to Identify the Lynch Syndrome Among Patients With Colorectal Cancer**, was [published](#) in the *Annals of Internal Medicine* on July 19, 2011. The article described results of a study that found that screening every new colon cancer patient for a particular for the Lynch syndrome extends lives at a reasonable cost.

TRANSPERS to Extend Research into Whole Genome Sequencing

The increasing ability to rapidly and inexpensively sequence entire genomes is poised to be the next paradigm shift in the application of genetic information to clinical care and health policy. Kathryn Phillips is a member of the Scientific Advisory Board on a newly funded U01 Research Proposal on "**Integration of Whole Genome Sequencing into Clinical Medicine**", with funding to Harvard from NHGRI at \$10.5 Million over 4 years.

This will position TRANSPERS to become an active participant in translational research on whole genome sequencing, which is expected to be commonplace in the cancer field within the next

TRANSPERS Center Recent Research

Ponce NA, Tsui J, Knight S, Aafable-Munsuz A, Ladabaum U, Hiatt RA, Haas JS. **Disparities in Cancer Screening in Individuals with a Family History of Breast or Colorectal Cancers.** *Cancer*. In press. [[Abstract](#)]

Lorizio W, Rugo H, Beattie MS, Tchu S, Melese T, Melisko M, Wu A, Lawrence J, Nikoloff M, Ziv E. **Pharmacogenetic testing affects choice of therapy among women considering tamoxifen treatment.** *Journal of Genome Medicine*. In press. [[Abstract](#)]

Ladabaum U, Wang G, Terdiman J, Blanco A, Kuppermann M, Boland CR, Ford J, Elkin E, Phillips KA. **Strategies to Identify the Lynch Syndrome Among Patients With Colorectal Cancer.** *Annals of Internal Medicine*. July 19, 2011;155(2):69-79. [[Abstract](#)]

Haas JS, Liang SY, Hassett MJ, Shiboski S, Elkin EB, Phillips KA. **Gene expression profile testing for breast cancer and the use of chemotherapy, serious adverse effects, and costs of care.** *Breast Cancer Res Treat*. Jun 17 2011. [[Abstract](#)]

Elkin EB, Marshall DA, Kulin NA, Ferrusi IL, Hassett MJ, Ladabaum U, Phillips KA. **Economic evaluation of targeted cancer interventions: Critical review and recommendations.** *Genet Med*. Jun 1 2011. [[Abstract](#)]

To learn more about TRANSPERS Center collaborators and our research, please visit our website at
<http://transpers.ucsf.edu>