

Personalized Medicine

Fall 2008 • Volume 3, Number 2



Inside

From the Director 2

In Memoriam:

Tom Berger 2

Genetic research,
testing likely to get
boost from new genetic
non-discrimination law . . . 3

National Institutes of Health policy will facilitate data- sharing, aid PMRP in its work

Sharing data with researchers worldwide, while at the same time protecting individual privacy, is the key to advancing the public health benefit of personalized medicine.

Jeff Struewing, M.D., Personalized Medicine Research Project (PMRP) Program Officer with the National Human Genome Research Institute, gave a presentation this past spring to the PMRP Community Advisory Group about the updated data-sharing policy implemented by the National Institutes of Health (NIH).

Sharing data from genome-wide association studies (GWAS) is done under terms and conditions consistent with the informed consent document that you signed when you enrolled in the PMRP.

A number of genome-wide association studies are underway as part of the Personalized Medicine Research Project. The PMRP will deposit data from one of the studies – on cataract and low-HDL (good cholesterol) – into a repository that can be accessed by researchers from other institutions who have been approved by the NIH. Data to be shared include average low-HDL, type of cataract, body mass index and genotype.



MARSHFIELD CLINIC®
Research Foundation

Continued on page 3

From the Director



Cathy McCarty, Ph.D.

I am writing this column in August. Along with the bountiful harvest of cucumbers and zucchini that have appeared at work to be shared with all, we have lots of PMRP news to share with you.

In collaboration with Dr. Joe Kenmitz at the University of Wisconsin and with funding from the National Institutes of Health, Dr. Joe Welter, a Marshfield Clinic obstetrician, and I will be starting a study of endometriosis, a condition in which tissue similar to the lining of the uterus is found elsewhere in the body, most often in the pelvic region. The primary symptom is pelvic pain, and an estimated 30 – 40 percent of women with endometriosis are unable to have children.

We are honored to be asked to participate in a leadership summit on the integration of personalized medicine into health care practices. The summit, sponsored by the

U.S. Department of Health and Human Services, will be held in Utah in October.

By the time you read this newsletter, the Center for Human Genetics will have moved into our lab and office space in the new Laird Center for Medical Research. Mark your calendars for the official dedication to be held on Friday, October 10, and a public open house and tours the morning of Saturday, October 11. We think the new arrangement, with close proximity of the research and clinical labs, along with our colleagues in the Biomedical Informatics Research Center, will shorten the time between scientific discoveries and clinical applications.

Please note on the list of PMRP Advisors on the last page that we have two new additions to the PMRP Community Advisory Group. I am delighted to welcome Noreen Moen and Pat Gall. We were saddened at the loss of our friend Tom Berger and will miss his contributions to the group and the community. You can read more about his life in this newsletter.

You can also read about dbGAP, a resource that will allow researchers to share data and combine data from various studies to increase our ability to detect genetic associations with disease and medication response. The PMRP was always designed to be a national resource to allow us to discover the information necessary to personalize health care, and we believe that the sharing of data will help us to do just that. With the passage of the Federal genetic non-discrimination act that you can also read about in this newsletter, we hope that concerns some people might have about the potential for discrimination in health insurance related to genetic results will be relieved.

PMRP enrollment is still open and we are continuing to collect dietary history and physical activity information for subjects already enrolled. If you know anyone who is 18 years or older and lives in one of the 19 ZIP codes around Marshfield and would like to participate, tell them to stop by the Lawton Center on the Marshfield campus or to call our toll free number, 1-888-334-2232 or 715-389-7733.

In Memoriam: Community Advisory Group member Tom Berger



Tom Berger

Tom Berger, 57, of Kronenwetter, who served on the Personalized Medicine Research Project's Community Advisory Group since its inception, died June 14 at his home after a six-year battle with cancer.

As a longtime newspaper reporter and editor, his interest in improving the health of his communities was deep and apparent.

Tom graduated from the University of Wisconsin-Eau Claire in 1972 with a journalism major, a field of endeavor he called a way to learn for a lifetime. He began working as a journalist at the Wisconsin Rapids Daily Tribune and spent most of 1977 to 2002 at the Wausau Daily Herald as a reporter, city editor and editorial page editor. In 2002, Berger became the Marshfield News-Herald's managing editor. Just prior to making that change, however, he learned he had colon cancer. What ensued was a battle that also included cancer of the liver and lung. He retired in May from the News-Herald.

Courageous, dedicated, champion, mentor and passionate are words used to describe Tom. He is remembered as a soft-spoken man

whose passion for community journalism was second only to his family. He said his lifetime of work brought him deep satisfaction. "My philosophy was to make relaying information an art form, to be a thoughtful, creative, intelligent witness," he said. "I think truth prevails in the end. It might not be fun, it might not be pretty. ... But I have belief in the good of the whole."

Besides the Community Advisory Group, he participated in a number of causes, including The Neighbors' Place in Wausau; the fight against Alzheimer's disease after his mother-in-law died from it; and, later, the fight against cancer. He is survived by Fran, his wife of 30 years; and their three children, Lindsay, 20, Christopher, 17, and Nathaniel, 13. Memorials may be directed to: Children's College Fund, 1954 Plantation Lane, Mosinee, WI 54455.

National Institutes of Health policy will facilitate data-sharing, aid PMRP in its work

(continued from page 1)

What is a genome-wide association study (GWAS)?: Any study of genetic variation across the entire human genome that is designed to identify associations with observable traits such as blood pressure and weight, or the presence or absence of a disease. This information will help lead to improvement in the prediction of disease and in an individual's care, and ultimately to personalized medicine. To learn more about genome-wide association studies, go to <http://grants.nih.gov/grants/gwas/index.htm>.

What is the GWAS policy?: The genome-wide association studies policy calls for GWAS researchers funded by the National Institutes of Health to: 1) submit de-identified genetic (genotypic and phenotypic) data to a centralized NIH repository; and, 2) submit documentation that describes how the researchers will protect privacy and confidentiality of research participants.

How does this sharing policy benefit Marshfield's Personalized Medicine Research Project?: It allows scientists outside Marshfield to study PMRP data, thus extracting more knowledge from PMRP studies. It also allows Marshfield scientists to gain access to other data for comparison.

Can non-research entities (e.g., law enforcement agencies, insurance companies, employers) request access to identifiable information?: The NIH does not possess any direct identifiers within the NIH GWAS data repository, nor will the NIH have access to the link between the data keycode and the identifiable information that may reside with the primary researchers and institutions for particular studies. The NIH explicitly encourages researchers submitting GWAS data to consider the potential appropriateness of obtaining a Certificate of Confidentiality (see <http://grants.nih.gov/grants/policy/coc/>) as an added protection

against any future compelled disclosure of identities for studies planning to collect genome-wide association data. Marshfield plans to write a data use statement to restrict access by insurance companies and only allow medical research questions. Marshfield has a Certificate of Confidentiality.

What security measures are in place to prevent unauthorized access to the NIH GWAS repository?: Datasets are stored in the NIH GWAS data repository under strict security provisions, including multiple firewalls, separate servers, and data encryption protocols.

What if I have questions about data sharing?: Contact PMRP staff at 1-888-334-2232 or 715-389-7733, or visit the PMRP on the Web at www.marshfieldclinic.org/pmrp.

Frequently asked questions about genome-wide association studies can be viewed at http://grants.nih.gov/grants/gwas/GWA_S_faqs.htm.



The Center for Human Genetics and the Personalized Medicine Research Project have moved into the new Laird Center for Medical Research. A public open house and tours will be held the morning of Saturday, Oct. 11, 2008.

Genetic research, testing likely to get boost from new genetic non-discrimination law

The Genetic Information Nondiscrimination Act (GINA), signed into law on May 21 by President George Bush, prevents health insurers and employers from discriminating based on an individual's genetic information or genetic predisposition toward a specific disease.

Less widely reported is the positive effect GINA could have on lessening the "fear factor" for an individual who is deciding whether to share genetic information by participating in a research study or by undergoing genetic testing, issues of great importance to the Personalized Medicine Research Project (PMRP) and residents of the Marshfield Epidemiologic Study Area.

It took 13 years for GINA to become law. Considered by some to be premature when first introduced, the legislation took on new urgency in 2003 with completion of the map of the human genome, which greatly accelerated the search for genes involved in susceptibility to common diseases. Genetic tests now encompass more than 1,500 conditions.

The PMRP is the largest population-based genetic research project in the United States, involving nearly 20,000 residents of the Marshfield Epidemiologic Study Area.

Said Cathy McCarty, Ph.D., Director of the PMRP, "It is important that research participants are fully informed about the new law and its implications."

Passage of GINA is welcome news, said Jeff Struewing, M.D., Personalized Medicine Research Project Program Officer from the National Human Genome Research Institute.

"These protections should allay some of the fears that participants have about participating in genetic research studies and the sharing of coded information within the scientific community," Dr. Struewing said.

In addition to research, GINA has implications for patients. With a national standard in place for protecting individual genetic information, genetic counselors expect more people to seek genetic testing that can help in the prevention or management of a broad range of diseases and conditions, said Marshfield Clinic Genetic Counselor Christina Zaleski, M.S.

But because genetic test results can be complicated and sometimes difficult to interpret, the National Society of Genetic Counselors advises patients to meet with a counselor before genetic testing, so that the patient fully understands the results and their implications, Zaleski said.

Federal agencies must still write the regulations that will guide health insurers and employers about how to comply with GINA. The health insurance regulations will take effect in May 2009, and the employment regulations six months after that.

Contact Us

This newsletter is a publication of the Personalized Medicine Research Project, Marshfield Clinic Research Foundation, 1000 N. Oak Ave., Marshfield, WI 54449-5790.

To contact the PMRP, phone 1-888-334-2232 or 715-389-7733, or visit the PMRP on the Web at <http://www.marshfieldclinic.org/pmrv>

To contact newsletter editor Scott Heiberger, phone 715-389-7541 or e-mail heiberger.scott@mcrf.mfldclin.edu.

PMRP Advisors

| | |
|---------------------------------------|---|
| Phil Boehning Loyal | Mark Krueger Marshfield |
| Sharon Bredl Stratford | Julie Levelius Stratford |
| Margaret Brubaker Stratford | Jerry Minor Pittsville |
| Margy Frey Marshfield | Noreen Moen Marshfield |
| Pat Gall Marshfield | Mike Paul Auburndale |
| Jodie Gardner Spencer | Marlin Schneider Wisconsin Rapids |
| Nancy Kaster Pulaski | Scott Schultz Osseo |
| Colleen Kelly Marshfield | Jean Schwanebeck Pittsville |
| Norm Kommer Colby | Mike Kobs Marshfield |
| Darlene Krake Marshfield | |