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Marshfield Clinic
Research Institute

Personalized Medicine Research Project

eMERGE Develops a Survey Asking About Patients' Views on Sharing Genomic Data

The Electronic Medical Records and Genomics (eMERGE) network has carried out a survey that asked patients their views on sharing genomic and clinical data for research. These views were sought after because the National Institutes of Health's policy states that genomic studies will only be funded if the study subjects agree to share their genomic data. A national policy also recommends that patients who participate in studies that use biological samples (e.g., DNA) or clinical data can provide a one-time consent for their samples and data to be used for approved research. "The views of patients on how widely their genomic and clinical data should be shared with researchers around the world are important for scientists to consider when developing bio-banks and designing genomic research studies," explains Murray Brilliant, Ph.D., Director of the Center for Human Genetics at Marshfield Clinic.

The varied types of patients and healthcare settings in the eMERGE network was ideal for testing patients' views on sharing data from genomic studies. The 11 clinical centers within eMERGE serve a large group of patients from a variety of backgrounds and are located in both urban and rural areas. Also, eMERGE already had a long-standing workgroup whose members had the skills and experience to perform studies of patients' views about being part of genomic research. An advantage was that these members had worked as a team in the past. Within the workgroup, a smaller subgroup called the "Survey Workgroup" was created to conduct

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Inside

Murray Brilliant, Ph.D.

Think Big

PMRP has been instrumental in our efforts to understand the genetic basis of many disorders. Your participation has led to new insights into the causes of these disorders and also to the identification of new drugs to treat and even prevent these disorders.

Recently, PMRP data were combined with data from all over the world in the largest study to date on the genetics of height ("Rare and low-frequency coding variants alter human adult height" Nature 542:186-190, 2016). The group that we are part of is called the "GIANT Consortium". DNA variations at hundreds of thousands of places in DNA were analyzed from over 750,000 people. These variations were checked for their effects on the height of all of these people. Nutrition and other environmental factors can affect how tall we can get, but genetics sets the height range we fall into. Around 700 variants have been found that impact on how tall or short we get, so although we inherit the range of height we can achieve from our parents, the story of why we get to be our height is rather complicated. Surprisingly, we found that

variants in genes associated with cholesterol levels and type 2 diabetes contribute to our height.

Speaking of BIG things, we are in the planning stages to expand our biobank. PMRP was designed for research, but not for clinical applications. One goal for our new biobank is to allow us to return individual results to improve clinical care, while expanding our research capabilities.

eMERGE develops a survey asking about patients' views on sharing genomic data

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the survey. The Survey Workgroup had the job of creating the survey and selecting patients to whom the survey would be mailed.

The Survey Workgroup decided on key tasks that were needed to conduct the survey and a small committee was created for each separate task. The tasks were (1) performing a review of past surveys on patients' views on sharing genomic data, (2) developing study protocols for review by Institutional Review Boards (IRBs), (3) developing the survey, (4) pre-testing the survey, (5) methods to define a sample of patients that would be mailed the survey, (6) managing study data, and (7) analysis of survey results. By using smaller committees focused on a single task, the Survey Workgroup avoided becoming too large and disorganized. Two co-leaders of the Survey Workgroup monitored the progress of each committee to ensure that each task was completed successfully.

Past surveys were reviewed to identify the key questions that needed to be asked about patients' views on sharing genomic data. Questions from past surveys were changed, as needed, to create the questions in the eMERGE survey. Questions in the eMERGE survey asked about patients' views on taking part in a bio-bank, their trust in the healthcare system and in persons who do medical research, their concerns about privacy, their health, and their age, race, education level, and residence in an urban or rural location. Persons who had at least one child under age 18 years were also asked their views about their youngest child taking part in a bio-bank. The survey considered one scenario for consent to share clinical data and three scenarios for consent to share genomic data with researchers (See chart on page 3 – Clinical and Genomic Data Sharing). Patients were randomly assigned to have one of the three bio-bank scenarios on their survey, and were asked whether they would want to participate in that scenario.

In past surveys, there were few participants who were not white, had a lower income, or had a lower education level. Therefore, the Survey workgroup decided to send surveys to an increased proportion of persons who were racial or ethnic minorities, younger adults, persons with a lower education level, and those living in rural areas. Surveys were mailed to 90,000 adult patients or parents of pediatric patients at the 11 eMERGE clinical centers. The patients could be eMERGE bio-bank participants or non-participants. About 13,000 surveys or 16% were returned. The eMERGE network will describe the results of its analysis of the survey responses in a future report.

We thank you for being part of the Personalized Medicine Research Project and eMERGE network. We also want you to know that your opinions on genomic research matter!

"Conducting a large, multi-site survey about patients' views on broad consent: challenges and solutions," Maureen Smith, et al. BMC Medical Research Methodology 2016;16:162.

Biobank Expansion

As described in the last PMRP Newsletter, Precision medicine is a new way to treat and help prevent disease. It looks at each person's genes (DNA), their surroundings and the way they live to find the best way to treat them and keep them healthy. We believe that one day everyone will have their DNA sequence known. Your medical care team will use data from your DNA to personalize your healthcare. They will use this information in a number of ways. One is to figure out what drugs work best for you so as to avoid prescribing drugs that may cause harmful side effects or may not work for you. Another is to determine your specific health risks. This would help doctors make better choices about your healthcare which can lead to you having improved health outcomes. By knowing your risk for certain disorders, we can treat you to prevent or delay the onset of those disorders. It can also help us diagnose a health problem earlier, when it can be treated more easily. This can lead to a better quality of life and reduced healthcare costs.

We plan to expand our biobank and have named the new program "One in a Million" to reflect the total number of people we envision becoming part of this initiative. We will start recruitment with the first goal of 15,000 to 20,000 people for this biobank. We believe that its success will translate into standard of care for everyone. More information on this project will be forthcoming in the next few months. We thank you for your contribution to the research that made this "GIANT" and bold new future possible!

Clinical and Genomic Data Sharing

You sign up to participate in a bio-bank at your clinic/hospital

Your clinical and genomic health information is stored in the bio-bank

Data Sharing Scenario
When you sign up, you agree to share your de-identified health information with researchers at your clinic/hospital who want to use the data for research

Bio-bank Scenarios
When you sign up, you also agree to:

Option 1	Option 2	Option 3
1. Place your de-identified health information is in a large, national database 2. Make decisions about the types of research you will allow your health information to be used for	1. Place your de-identified health information is in a large, national database 2. Allow your health information to be used for all kinds of medical research	1. Place your de-identified health information is in an online database that anyone can access 2. Allow your health information to be used for all kinds of medical research
Level of Database Access: **Restricted**	Level of Database Access: **Restricted**	Level of Database Access: **Available to Anyone**
Type of Consent to Share Health Information: **Based on Patient's Decision**	Type of Consent to Share Health Information: **Broad Consent**	Type of Consent to Share Health Information: **Broad Consent**

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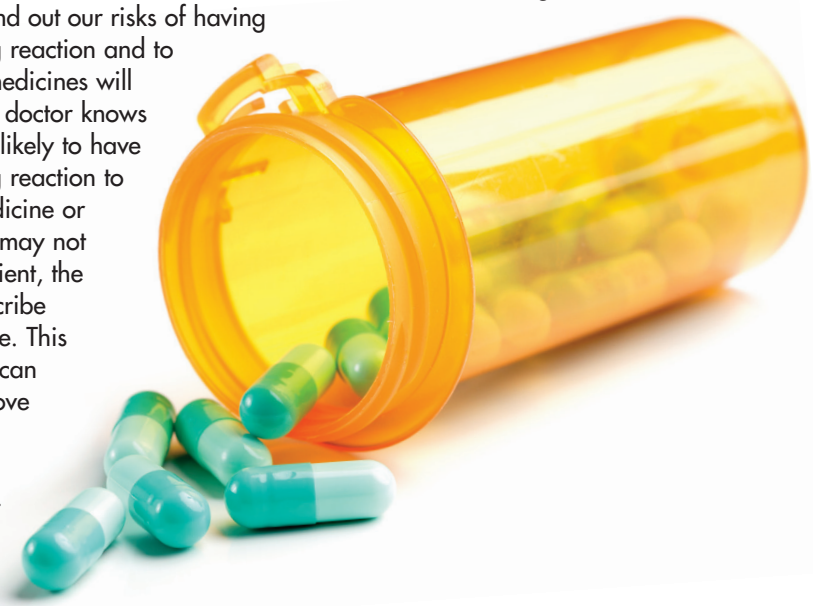
Pharmacogenetics in Action – eMERGE PGx Research at Marshfield Clinic

Personalized medicine, also known as precision medicine, is a growing field in healthcare. One area of personalized or precision medicine is called pharmacogenetics (PGx). It is the study of how our genes control how our bodies break down medicines. Sometimes, due to our genes, our bodies do not break down a medicine properly. This can cause us to have a bad reaction to the medicine or it can prevent the medicine from working as it should. A bad reaction is described as an "adverse drug reaction" and it can sometimes lead to severe illness. If a medicine does not work right, we do not get any benefit from taking the medicine. Therefore, studying how genes affect the way our bodies break down medicines can help us to find out our risks of having an adverse drug reaction and to know whether medicines will work for us. If a doctor knows that a patient is likely to have an adverse drug reaction to a particular medicine or that a medicine may not work for the patient, the doctor can prescribe another medicine. This shows that PGx can potentially improve patient health and reduce healthcare costs.

Marshfield Clinic is participating in the eMERGE PGx study. This study examines the genes that our bodies use to break down three medicines: simvastatin, clopidogrel, and warfarin. Simvastatin is used to lower the level of cholesterol in blood. Clopidogrel and warfarin are used to reduce the chance of blood clots forming in our bodies as these clots can cause heart attacks or stroke. The genetic results are entered into patients' electronic medical records so it can be seen by doctors treating the patients. A doctor who prescribes any of the three medicines to a patient for the first time is warned by an alert in the electronic medical records whether the patient is likely to have an adverse drug reaction after taking the medicine.



The doctor can then decide whether to prescribe other medicines to avoid an adverse drug reaction. Thus, PGx helps a doctor to give the right dose of the right drug to the right patient.

There are two main goals of current eMERGE PGx research at Marshfield Clinic. The first goal is to examine whether doctors prescribed other medicines to patients who had a high chance of having an adverse drug reaction and whether patients who used simvastatin, clopidogrel, or warfarin developed an adverse drug reaction. The second goal is to ask Marshfield Clinic doctors if they found the alerts in the electronic medical records to be useful for treating patients and whether the alerts were easy to use. We will use these results to improve the alerts and help doctors to provide better healthcare to patients.

Contact Us

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