What are genes?

Genes are pieces of DNA that have the instructions needed to make our bodies work. DNA stores these instructions in the form of a code. This is the code that you inherit from your parents and that you pass on to your children.

What is pharmacogenomics?

Pharmacogenomics (pharm-a-co-gen-o-mics) is the study of how genes affect the way the body processes, or metabolizes, medications. It is well-known that the same medication can work well in one person, but not in another. There can be negative side effects in one person, but not in another. Scientists have developed some genetic tests that can help predict who will not respond well to certain medications. In the future, there will be more of these tests available. The goal of pharmacogenomics is to use genetic information to help make sure patients are prescribed the best medication for them from the beginning of treatment. This will hopefully prevent or reduce side effects and re-hospitalizations from bad reactions to medications.

Why is my doctor recommending the CYP2C19 Genotyping test?

Studies have shown that some changes in the *CYP2C19* gene can make clopidogrel (Plavix®) not work as well in some people. As a result, they have a higher risk for heart attacks and strokes. Knowing if you have specific changes in the *CYP2C19* gene may help your doctor decide which medication and dose is the most appropriate for you.

What are the possible results of testing?

The *CYP2C19* Genotyping test ordered by UMMC doctors looks for six changes in the gene. The result is your genotype. The genetic changes called *2 (“star 2”), *3, *4, *6, and *8 make the gene non-functional and clopidogrel not work as well. The change called *17 causes clopidogrel to be metabolized faster than normal. A *1 result means that the above genetic changes were not found. Since we each have two copies of each gene, there are many possible combinations of genotypes one can have (see chart). The combination of genotypes is your diplotype and may predict how the body will respond to clopidogrel.

Diplotype	Predicted Effect	Type of Metabolizer
*1/*1	Normal activity	Normal
*1/*17 and *17/*17	Increased activity	Rapid and Ultra-rapid
*1/*2, *1/*3, *1/*4,*1/*6,*1/*8 *17/*2, *17/*3, *17/*4,*17/*6, or *17/*8	Decreased activity	Intermediate
*2/*2, *2/*3, *2/*4, *2/*6, *2/*8 *3/*3, *3/*4, *3/*6, *3/*8 *4/*4, *4/*6, *4/*8 *6/*6, *6/*8, or *8/*8	Significantly decreased activity	Poor

The *CYP2C19* Genotyping test result will be placed in your UMMC medical record so that doctors can use this information to help decide which medications to give you in the future.

Limitations of testing

This test only looked at the most common changes in *CYP2C19*. Other unknown changes in the gene may change the way clopidogrel is processed in the body. Therefore, a “normal” result does not guarantee that your body will respond to clopidogrel.

Also, please be aware that genes are just one of the things that can affect how someone responds to clopidogrel. Having Type 2 Diabetes, kidney failure or taking certain other drugs at the same time as clopidogrel can also change how well the drug will work for you. Your doctor will consider all of these factors when deciding on the best treatment for you.

How will these results affect my medical care?

CYP2C19 genotype results can affect choice or dosing of multiple medications. If *CYP2C19* Genotyping test results predict that you are an intermediate or poor metabolizer of clopidogrel, your doctor may recommend that you take a different medication. If you are prescribed clopidogrel, YOU SHOULD NOT STOP TAKING IT OR CHANGE YOUR DOSE unless told to do so by your doctor.

Can the results of this genetic test be used against me by my insurance company?

Right now the *CYP2C19* Genotyping test is only used to predict drug response. Research may link *CYP2C19* changes to some sort of disease in the future. If this happens, someone could try to make it harder for you to get a job or insurance. There are laws against using genetic information this way, but they may not give full protection. We believe that the chance these things will happen is very small.

Who can I contact if I have questions about the test?

We encourage you to discuss these results with your cardiologist and other doctors. You can also contact us to request a copy of your results.

Check out the following websites for more information on pharmacogenomics:

http://ghr.nlm.nih.gov/handbook/genomicresearch/pharmacogenomics

http://www.nigms.nih.gov/Research/SpecificAreas/PGRN/Background/pages/pgrn_faq.aspx

Contact Information

Translational Genomics Laboratory
655 W. Baltimore St., BRB 7-037
Baltimore, MD 21201
Phone: 410-706-3339
FAX: 410-706-0287

UNIVERSITY of MARYLAND
SCHOOL OF MEDICINE

Translational Genomics Laboratory

CYP2C19 Genotyping

Patient Information