What is Pharmacogenetics?

Pharmacogenetics (PGx) is the study of genetic variations in the human body in relation to a person's ability to metabolize and process prescription and OTC drugs. While pharmacogenetics gained traction after a 1950s study of the effects of specific medications on different individuals based on their genetic makeup, medical science has known for over 1,500 years that many individuals react differently to certain medications.

Pharmacogenetics: The Basics

Researchers were able to conclude that individuals who have certain genetic variations respond differently to prescribed drugs than those who are without those variations. That is to say that the same dosage, timing and frequency of a drug treatment plan of care may have the ability to metabolize as anticipated in one patient, while it may metabolize differently in another, all based on that individual's genetic makeup.

The study of pharmacogenetics and the process of pharmacogenetic testing allows healthcare providers the ability to analyze gene markers that can highlight a patient's inability to process or metabolize a drug as expected. This allows the provider to prescribe a drug treatment plan personalized to a specific patient.

How Pharmacogenetic Testing Works

Pharmacogenetics in practice is built on the foundation of pharmacogenetic testing, which evaluates the basic unit of genetic material, known as a gene, and the segments of DNA that provide instruction for producing specific proteins or enzymes. Each gene within the human body consists of a unique genetic code that is made up of different nucleotides, and individual genetic variations occur throughout the population. While some genetic variations do not produce noticeable negative effects, others are known to cause specific disease or conditions. Similarly, some genetic variations are known to impact an individual's response to certain drugs.

Pharmacogenetic tests seek out genetic variations within individual patients that are associated with responses to prescription medications, found in genes that are used to produce drug-metabolizing enzymes, medication targets, or the proteins involved in the immune system process. Healthcare providers use pharmacogenetics testing to determine which genetic variations are predominant, and this information is used to understand a patient's future response to certain medications. Providers can perform pharmacogenetic testing prior to or throughout the duration of a drug treatment plan to help understand an individual's potential drug response.

The study of pharmacogenetics and the process of pharmacogenetic testing is important to the future of healthcare as more prescription drugs come to market. Individuals who are in need of a drug treatment program for the management or cure of specific diseases can be negatively impacted by the adverse drug reactions that result from less than ideal medication regimens. Pharmacogenetics helps alleviate the need for trial and error in the treatment of patients, and offers a way to strategically target care based on an individual's genetic code.

In 2003, the medical world recognized the significant possibilities that would become available by the sequencing of the complete set of genetic information for humans, the human genome.

Francis Collins, the director of NHGRI (National Human Genome Research Institute), noted that the genome could be thought of in terms of a book with multiple uses: "It's a history book – a narrative of the journey of our species through time. It's a shop manual, with an incredibly detailed blueprint for building every human cell.

And it's a transformative textbook of medicine, with insights that will give health care providers immense new powers to treat, prevent and cure disease.

Pharmacogenetics: The Key to Precision Medicine

Pharmacogenetics, an important component of precision medicine, is the application of how genes affect a person's ability to metabolize drugs. In layman's terms, precision medicine is a tool that helps your healthcare provider customize a medication treatment plan designed to provide individuals with the best possible outcomes while minimizing potential adverse drug reactions (ADRs).

Providers and pharmacists have long understood that not every drug performs the same way for every patient. A medication that works well for one person can have harmful side effects for someone else. For example, some people may not break-down (metabolize) certain medications as expected. As a result, a dosage level that would be safe for most individuals would be toxic for these patients. Other individuals might process medications too quickly for the drugs to have their intended effect.

Reduced Trial and Error

Precision medicine can help replace the trial and error process that exists today where providers prescribe drugs and dosage levels based on general population data, the one size fits all approach.

Today, the DNA ANALYTICAL scientific team uses open array technology to identify 180 alleles present on 54 genes that determine if patients can metabolize the more than 1600 medications checked. When shared with your healthcare providers, your individualized DNA drug sensitivity test report will help them to create a more personalized medication therapy program.

For more information, visit the White House precision medicine web page: wh.gov/precision-medicine

Why Is PGx Testing Important?

Employing DNA drug sensitivity testing to screen medications before therapy helps patients avoid drugs they won't process and/or metabolize well, and instead provides the prescriber insights that can help them prescribe the right medication from the beginning. This can help accelerate the benefits from medications, reduce wasted time and expense on ineffective medications, and possibly even save lives.

When prescribing or taking medications, there isn't room for error

But that's the problem patients, pharmacists and providers face every time a prescription is written: the possibility that the medication won't work, or that it will have a toxic or life-threatening side effect. Prescribers must rely on general population statistics to select effective medications for their patient and hope their patient isn't part of the cohort that experiences dangerous side effects or an adverse drug reaction. The PGX test gives providers and pharmacists specific data to help make evidence-based decisions personalized to the patient's genetic profile.

Reduced Adverse Effects

According to the Food and Drug Administration, adverse drug reactions, or ADRs, are one of the leading causes of mortality and morbidity in the healthcare system, ahead of pulmonary disease, diabetes, accidents and automobile deaths. The number of adverse drug reactions is slated to increase as more medications become available in the market, and as more healthcare providers implement drug treatment plans that consist of more than four prescription medications.

The World Health Organization (WHO) defines an adverse reaction as "[a] response to a drug which is noxious and unintended, and which occurs at doses normally used in man for the prophylaxis, diagnosis, or therapy of disease, or for the modifications of physiological function."[1]

Unfortunately, many medications are prescribed without determining how a patient's genetic profile influences the assimilation and metabolism of medications. And while some side effects are a challenge to avoid, some ADRs are the result of drug treatment plans that are not well-suited for an individual patient.

Pharmacogenetic testing provides patients and providers with a profile of genetic variances that will assist in designing a treatment plan that will reduce the occurrence of ADRs. When providers have a better understanding of how a patient's genetic profile may interact with specific medications, a more effective drug treatment plan can be set in motion.

Medication Adherence

Poor medication adherence is an increasing concern in the medical community, especially for patients who are diagnosed with chronic conditions or diseases. This is because patient behavior related to the proper use of prescribed medications and drug treatment plans varies from individual to individual based on a bevy of factors. Individual patients who have a high degree of confidence in the medications prescribed by a healthcare provider typically have a high level of medication adherence, meaning prescriptions are taken as directed in the proper dosage for the recommended amount of time. Alternatively, patients who lack confidence in the safety or efficacy of a prescribed medication generally fail to follow the regimen of care as closely as providers deem necessary for successful drug treatment.[2]

Pharmacogenetic testing, also known as PGx testing, provides healthcare providers a sound method to reduce the potential for poor medication adherence by increasing patient confidence in prescribed drug treatments. Pharmacogenetics offers additional insight into how patients' genetic profiles may affect the outcome of or reaction to specific medications. Pharmacogenetic testing lays part of the groundwork for precision care, based on the individual needs and genetic makeup of a single patient.

Armed with pharmacogenetic testing results, providers can speak with more authority on an individual patient's drug treatment plan, ultimately leading to increased patient confidence and medical adherence.

Reduced Medical Costs

High medical costs are directly correlated to incorrect or inefficient drug treatment plans in a number of individual patient cases. Patients who go through several rounds of various prescribed medications in an attempt to find the right fit have a higher propensity to spend more on care. Higher costs are the result of an increased number of healthcare provider visits, multiple costly prescriptions and the need for treatment related to adverse drug reactions or side effects of inappropriate or ill-suited drug treatment recommendations.[2]

Knowing one's genetic profile and related metabolism of certain medications starkly decreases the possibility of providing a drug treatment plan with unwanted effects. Logically, PGx testing prior to prescribing certain medications has the potential to reduce the total expenses associated with ineffective health care for patients by reducing the need for treatment plan changes.

Optimal Dosing

Prescription medications are tested thoroughly prior to being released for approved use in the treatment of diseases and conditions, but the research in pharmaceutical development has its limits. Drug manufacturers and medical researchers use a sample group of individuals to ascertain whether a drug is effective in treating specific ailments, and decisions about efficacy are based upon that population set. While a medication in a specific dose may be effective for a portion of the population based on that truncated research group, not every individual will experience similar results from what is deemed the standard dosage of a prescribed medication.[3]

Pharmacogenetic testing provides a direct path to individualized treatment based on the genetic traits present in each patient. By utilizing precision medicine backed by pharmacogenetics, providers have the ability to pinpoint the optimal dosage of certain medications that have the greatest potential to result in treatment success for patients.

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