

How genetic testing will help personalize your medicine

For much of modern medical history, treatment has centered around the average patient. Discovering treatments which work for most people, most of the time has been a necessary starting point. However, treating every patient according to an average is rarely the most effective treatment method and can potentially even cause harm in some cases.

When the [U.S air force first designed its planes](#), it based every measurement of the cockpit – from the shape of the seat, to the height of the windshield, to the distance between seat and pedals – according to the average of dimensions from hundreds of pilots. Nevertheless, unexplainable crashes kept occurring.

A young researcher tasked with studying the conundrum discovered the flaw: **no individual is average**. By replacing the average-sized designs with new versions that could be adjusted to the individual, the problem was solved. Now we are discovering that the flaw of averages – and the need for personalization – is equally important in medicine.

We now know that certain ethnic groups are more susceptible to genetic conditions and respond differently to treatment. Likewise, women can present with very different symptoms to men for the same disease. Genetic testing moves vastly beyond even these differences – opening up treatment possibilities tailored to each specific individual.

Safer prescription and administration of drugs

Individual genetic makeup can uncover the difference between an effective drug and a severe allergic reaction. The study of how genes affect drug response is known as **pharmacogenomics**.

Genetic differences can determine which drugs are selected for treatment. One drug, ivacaftor or Kalydeco, is used to treat cystic fibrosis – it's a first-line treatment, [but only for the 5% of CF sufferers](#) who have a specific genetic mutation.

In other cases, genetic testing is used to determine safe dosage levels. Thiopurine drugs are used to treat leukemia but can cause [dangerous levels of bone-marrow suppression](#). The dosage window between effective treatment and toxicity is small. Individuals with a certain TPMT gene mutation are ten times more sensitive and have a ten times smaller window – genetic testing can identify them and protect them from these toxic side-effects.

Advanced cancer treatments

There are over 100 types of cancer and over a third of people will be diagnosed with one of them at some point during their lifetime. As the second biggest killer after heart disease, few people escape its effects – either via themselves or by seeing their loved ones affected.

Cancer is caused by mutations within a cell's DNA which cause it to grow abnormally and uncontrollably. Some of these genetic mutations are caused by exterior damage – sun and smoking, for example – while some are present at birth. Genetic testing of an individual can evaluate their risk of developing certain types of cancer, but tumours can also be genetically tested to determine their makeup.

One of the first examples of personalized medicine, dating back to the 60s, involves a breast cancer hormone therapy known as tamoxifen. It targets estrogen receptors present on the cancer cells. Some breast cancers do not exhibit these receptors – [rendering tamoxifen useless in these cases](#).

Understanding not only the genetic makeup of the patient, but of the tumour itself, has led to new classifications of tumours and new treatment opportunities. Whereas historically cancers have primarily been classified by the point they originate from on the body – lung, breast, pancreas – classifying them according to certain genomic markers [opens up new avenues for effective treatment](#).

Early risk detection and intervention

Almost all disorders – whether genetic or acquired – are most effectively treated with early intervention. Genetic testing can be performed in utero, at birth and later in life.

Some disorders are easier to test for – those directly caused by a single gene or small number of genes, such as cystic fibrosis, sickle cell anaemia and muscular dystrophy. Where available, early intervention can be started as soon as the diagnosis is made, reducing the severity of the symptoms and improving quality of life.

Other more complex diseases can have dozens of gene variants associated with increased risk – [over 90 gene variants](#) have been linked to an increased risk of breast cancer. Genetic testing cannot directly say whether or not an individual will be affected in their lifetime, but high risk individuals can be better informed and prepared.

As modern genomics continues to advance, the progression towards personalized medicine will only accelerate. The potential benefits in terms of treatment efficacy, risk assessment and harm reduction cannot be understated.

The accumulation of this level of personal medical data, however, comes with its own set of challenges. Private genetic information can have significant consequences in the wrong hands – for example, when it comes to health insurance coverage. Patient security from both a technological and legal standpoint needs to be a priority, and here novel technologies such as blockchain can play an important role and create unprecedented value for the precision medicine ecosystem.