Personalized Medicine

At its core, personalized medicine uses information about a person’s genetic background to tailor strategies for the detection, treatment or prevention of disease. This may include genetic screening tests to identify susceptibility to disease or more precisely pinpoint existing conditions. It may also be used to guide pharmaceutical choices, highlighting the brand and dose of medication best suited for a patient. The goal of personalized medicine is to help physicians and their patients identify the best course of action to prevent or manage a disease based upon the patient’s genetic and environmental profile.

Drawing an analogy from the world of fashion, personalized medicine is the equivalent of a custom-made suit or outfit, designed with an individual’s unique body measurements. This type of tailored approach provides a much better fit than purchasing something off the rack.

As has already been noted in this guide, people vary from one another in many ways – what they eat, their lifestyle, the environmental factors to which they are exposed and variations in their DNA. Some portion of this genetic variation influences our risk of getting or avoiding specific diseases. Certain changes in the DNA code influence the course of disease, impacting the age of onset for symptoms or the speed of progression. Genetic variation also contributes to differences in how drugs are absorbed and used by the body [see the section on pharmacogenomics on pg 46].

This newfound knowledge is rapidly moving into the clinical setting. At the forefront are a series of drugs such as Gleevec®, Herceptin® and Iressa® known to be most effective in people with a specific genetic profile [set of genetic variants].

Straightforward genetic tests are performed to identify who will benefit from these medications. More precise diagnostic tests are in development that better classify disease subtypes or progression. The information identified in our genome will help develop a lifelong plan of health maintenance tailored to our genetic profile.

One of the holy grails in personalized medicine is the so-called $1,000 genome — the ability to sequence a human’s genetic information at an economically feasible price. Recent advances in sequencing technology have moved the field closer to this figure. In addition to issues of cost, there are other challenges to personalized medicine, including concerns about patient privacy, confidentiality and insurability after taking a genetic test. Will the knowledge that specific genetic variation increases disease risk lead to greater or reduced prejudice or discrimination? How will access to genetic testing and personalized medicine be equitable? Does our current healthcare system need to change in light of this genetic approach, and if so, which new model will be best?