Editorial

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Editorial on Genomics Innovations and Advanced Technologies

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Genome technologies enable us to detect and characterize genetic inheritance, disease susceptibility, and responses to environmental influences and potential treatment regimes. These technologies, when applied effectively to examine variations in genetic makeup, RNA expression and epigenetic modifications at population scale and in high resolution, provide the basis for discovering marker genes, pathways and mechanisms of human diseases and the knowledge base for precision medicine.

A complete human genome contains three billion base pairs of DNA, uniquely arranged to give us our fundamental anatomy and individual characteristics such as height and hair color. DNA forms genes and understanding their function gives crucial insights into how our bodies work and what happens when we get sick. This was the reasoning behind the 13 years and $2.7 billion spent on the Human Genome Project. The world has quickly built on its achievements and now we can map a human genome in just a few hours and for less than a thousand dollars. Fast, large-scale, low-cost DNA sequencing has propelled genomics into mainstream medicine, driving a revolutionary shift toward precision medicine.

Early diagnosis of a disease can significantly increase the chances of successful treatment, and genomics can detect a disease long before symptoms present themselves. Many diseases, including cancers, are caused by alterations in our genes. Genomics can identify these alterations and search for them using an ever-growing number of genetic tests, many available online. If your results suggest susceptibility to a condition, you may be able to take preemptive action to delay or even stop the disease developing. “Health care will move more toward prevention rather than cure,” Malik says, and genomics will likely prove an important enabler in understanding the particular healthcare steps an individual should or should not take.

When symptoms do develop, genomics can be instrumental in diagnosing the problem. The Human Genome Project has fueled the discovery of nearly 2,000 disease genes, and these are proving highly effective at providing fast and accurate analysis. They have been especially valuable for identifying rare genetic diseases that had previously taken years to diagnose, ending the uncertainty and suffering of “diagnostic odysseys.” Ongoing research is committed to building databases of genetic biomarkers, especially for cancers. “To date, genomics has had the most impact on cancer,” Malik explains, “because we can get tissue, sequence it, and identify the alterations.” In the United States, the Cancer Genome Atlas has mapped the key genomic changes in more than 30 types of cancer. Such databases could deliver a definitive diagnosis in seconds, and even recommend targeted treatments based on the DNA of both the patient and the disease. Indeed, genetic sequencing of cancer tumors is helping not only to identify particular cancers but also to understand what causes them and what could kill them.

When it comes to treatment, genomics is driving another important element of precision medicine—pharmacogenomics. “We’ve long known that the same medication in the same dose affects people differently,” Malik says. Now we know why: Our genes influence the production of crucial enzymes in the liver that metabolize medicines. If a genetic variation stops the enzymes from working properly, the drug can build up in the body with serious side effects. Other drugs only work when broken down in the liver, so if the enzymes don't work, neither does the drug. Such gene variations, known as polymorphisms, are common, but genomics means we can test for them and compensate for them. Gene variations mean that around 30 percent of people cannot fully convert a commonly used anti-clotting drug, but gene testing means alternative drugs can be taken to the same effect.