



The Future of Medical Care
A genomic medicine story



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Genomic Medicine is an emerging discipline that helps physicians prevent and diagnose illnesses, and create customized treatment plans for patients based on their individual genomic information.

Some diseases such as cystic fibrosis and muscular dystrophy are controlled by a single gene. Physicians can use genomics to identify the presence of a mutation in that gene at birth, in utero, or even before conception by examining the parents' genetic profiles. In the case of cystic fibrosis, over 900 gene mutations can lead to symptoms of the disease. About 4% of cases are the result of a mutation that is known to respond to a certain type of medication. If a patient's DNA shows they have this specific mutation, physicians can immediately prescribe an effective treatment rather than relying on trial and error.

Infectious diseases are caused by microorganisms such as bacteria, viruses, and fungi. An example is hepatitis C, which is caused by the hepatitis C virus and passed when blood from an infected person gets into the system of another. Chronic Hep C, which can cause cirrhosis and liver cancer, is a major health concern affecting over 185 million individuals worldwide. Recently, researchers have discovered that variations in both the genes of the patient and the genes of the virus can influence the effectiveness of different medical treatments. Examining the patient's unique genomic information can help physicians recommend the best course of care.

Complex disorders like cancer, cardiovascular disease, and diabetes are caused by an interplay of gene variants, lifestyle choices, and environmental exposure. Oncology, the study of cancer, is on the cutting edge of using genomic markers to not only screen for cancer, but to create tailored treatment strategies for patients. Colorectal cancer is one of the most common cancers in both men and women. Between 15 and 20% of patients have a mutation in a gene that is part of a cellular signaling pathway. Regular treatment with aspirin decreases the activity of this pathway, increasing survival rates. A test for the presence of this specific mutation can determine if an aspirin regimen is the best treatment plan for the patient.

With recent advancements in genomic medicine, we now have tools to more effectively diagnose and treat disease through customized care. But these developments raise many ethical issues that need to be considered, including who has access to this kind of treatment, and what entities may be privy to an individual's genomic information.