Case Study Challenge 2018
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Genetic Testing: A Paradox of Big Data

Background

The human genome is composed of 23 chromosomes and the mitochondrial chromosome and contains approximately 22,000 genes. The Human Genome Project was an international project to identify these genes and their locations. It was completed in 2003 and the information is available in genetics databases called biobanks. Scientists are now working on understanding how these genes work together in health and disease. However, tracking gene interactions and billions of possible mutations requires the analytic capabilities of another development of the 21st century: big data. Big data allows storage of and access to large amounts of data over a network of computers. Thousands of computers in buildings called server farms can access data from anywhere in the world faster than an individual computer. Big data allows scientists to sort through medical records and gene data for correlations between genes, mutations, disease, and treatments. Big data analyses make it possible to use genetic testing to diagnose and treat a disease or help people plan ahead by uncovering genetic mutations.

Genetic mutations are responsible for some diseases. These mutant genes may be inherited. In the 1970s, the first disease genes identified enabled couples to use genetic tests to determine whether they carried Tay-Sachs or sickle-cell genes and newborns could be tested for an abnormal phenylketonuria gene. Today, most states mandate testing for 32 disease genes specified by the Health Resources and Services Administration.

Many medications that are available today are given to patients with a standard dose. However, people react differently to medications. Pharmacogenomics, the study of how genes affect drug action, aims to develop safer, more effective medications tailored to individual genetics. Big data analyses of genetic tests and medical records help predict how the body will react to a medication before administering the drug. Some of the diseases studied include cardiovascular disease, Alzheimer's disease, cancer, and asthma. Once a genetic test is developed, it can reduce side effects, speed treatments, and improve outcomes. For example, the VKORC1 gene is responsible for resistance to warfarin, a drug used to prevent blood clots. The FDA-approved label for warfarin states that VKORC1 information can help determine the dose of warfarin.

Big data analyses have led to a profusion of companies selling direct-to-consumer (DTC) genetic testing. These companies market to consumers rather than healthcare providers, and allow people access to their genetic information. The data are added to biobanks that can be used to discover new genes, interactions between genes, or interactions between genes and the environment.

Genetic testing and personalized treatment are improving quality of life. However, much is still unknown about gene interactions. Information is not available for all genetic conditions and the available information does not definitely tell you whether you will get a particular disease. A patient with the BRCA1 cancer gene, may not develop the disease because of other unidentified genes or lifestyle choices. DTC results are shared with researchers and can be accessed by law enforcement. How do we balance this availability of genetics data with privacy and healthcare?

The future

The Genetic Information Nondiscrimination Act (GINA) prohibits health insurance companies from denying coverage or increasing costs based on genetics. GINA does not apply to life, disability, or long-term care insurance, small businesses, or the military. Thus, these agencies can ask about genetic information and may use genetics to limit coverage or employment. Does DTC data shared for research remain anonymous. A hacker could sell your genetic information to insurance companies or ransom your DNA, threatening to post it online. Laws will need to catch up with technology.

Some disease-causing genes are expressed with environmental stimuli such as diet and some diseases result from several genes. Therefore, physicians don't necessarily understand how a mutation affects a healthy individual. Moreover, false positives occur in 40% of DTC tests. Learning that you have a disease gene can cause anxiety and unnecessary treatment. Women have chosen prophylactic mastectomies and hysterectomies based on incorrect genetic tests.

Replacing mutated genes to prevent diseases that are caused by a single gene, such as cystic fibrosis, is becoming possible. However, other conditions, including diabetes, encoded by several genes make genetic modification challenging, if even possible. This leads to the question whether individuals should know they have such genes.

Discussion Questions

Explain whether you would you agree to let a DTC company share your data?

Will you change your diet if one DTC test says you need a low-fat diet but another doesn't mention diet?

Does identifying a person's cancer genes benefit family members?

Should people know whether they have a gene for an untreatable killer disease?

A biotechnology company makes a drug from an antitumor gene in your DNA. Who owns the gene and patent?

6 Academic Resources

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