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Genetic information has been used extensively in medical practice for many years. One of the first questions a doctor will ask a new patient is for a brief snapshot of their family medical history. This query is an attempt to characterize a person's genetic tendency towards specific ailments. In fact, it is a snapshot of an individual's genetic predisposition towards disease, albeit on the basis of a rather small sample of related individuals. This information, however, is of critical importance to individuals and doctors as they make healthcare and lifestyle decisions. Personal genetics has the ability to greatly improve the accuracy in predicting an individual's predisposition to diseases, giving doctors and individuals a more complete understanding of a person's "family history" of disease.

Many personal genetic tests have already found medical and commercial success. One of the most well known examples of genetic testing, the *BRCA1* (1) and *BRCA2* test, has already helped many women better manage their risk for breast cancer. Individuals with a deleterious mutation in one of these genes often choose to undergo more frequent cancer screening, which helps to identify any tumors earlier and leads to an improved clinical outcome. This same procedure could improve clinical outcomes in other cancers and diseases. Now, more mutations are being identified that may influence an individual's risk of developing a given disease. Knowledge of carrier status for this information may have a similarly positive effect on preventative medicine and early detection of disease.

However, genetic testing is most effective when augmented with other information. One argument against personal genetic testing is that environmental and lifestyle effects may influence an individual's risk for a disease as much as, if not more than genetic variants. However, this actually makes genetic testing even more useful because it allows individuals to take lifestyle steps to reduce their environmental risk for a disease. For many chronic and metabolic diseases, the best treatment is preventative medicine before the onset of the disease. Knowing one's genetic risk may help to encourage healthy behaviors that reduce the risk or magnitude of a disease. For instance, a Stanford study where an individual's knowledge of an elevated risk for Type 2 Diabetes led to a dramatic lifestyle shift that may have reduced the effect of the disease (2). Had the individual not known about his genetic predisposition, they may have not undergone lifestyle changes with the same fervor and, instead, may have developed a worse clinical outcome.

Prediction of disease risk from genetic information is not perfect and there is much work to be done to fully understand the genetic architecture of complex diseases. However, there is a huge potential benefit from personal genetic testing that can be unlocked by researchers and companies in collaboration with the public. It is imperative that researchers, companies, doctors, and the public continue to work to understand the genetic architecture of disease in a way that allows this new information to be openly shared and understood.

References

1) Hall et. al. 1990. "Linkage of early-onset familial breast cancer to chromosome 17q21." *Science* 250(4988):p1684-1689.
2) Chen et. al. 2012. "Personal omics profiling reveals dynamic molecular and medical phenotypes." *Cell* 148(6):p1293-1307.