Dartmouth researchers get personal with genetics

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Two recent studies by Dartmouth researchers use individual genetic data to reveal the powers and limits of our current understanding of how the genome influences human health and what genes can reveal about the ancestry of the people of New Hampshire.

Published in the Sept. 11 issue of the American Journal of Human Genetics, Dartmouth Professor Jason Moore and Vanderbilt Professor Scott Williams analyzed how personal genetic testing companies are using still-nascent genome data to judge the health of their customers. Their paper is titled "Epistasis and its Implications for Personal Genetics."

People can now buy inexpensive kits, submit a DNA sample (often a swab from the inside of a cheek or a little bit of saliva), and receive data about their susceptibility to a number of gene-influenced ailments, such as prostate cancer, Alzheimer's, or type II diabetes. Moore and Williams argue that our knowledge of the human genome and its relationship to human health, while growing by leaps and bounds, is still in its infancy.

"The relationship between health and genetics is very complex," says Moore, professor of genetics and of community and family medicine at Dartmouth Medical School (DMS). "It's often a combination of multiple genes and multiple environmental factors that work together to increase or decrease your risk of disease. I don't think the knowledge base is sufficient to put genetics in the hands of the public quite yet." Moore is also the Frank Lane Research Scholar in Computational Genetics and

Director of Bioinformatics at DMS

The authors admit that genetic research is progressing, and they cite the example of the discovery of the BRCA1 and BRCA2 genes and their role in breast cancer. However, the authors caution that, while there is no question these genes are involved in breast cancer, the underlying mechanisms behind the genetic risk are still being worked out.

"There is a perception that these tests can provide answers," says Moore. "I used my own genetic material for this study, and my results didn't really tell me anything I didn't know, based on family history."

Moore and Williams call for refocusing and stepping up the research on gene-to-gene and gene-to-environment interactions. They explain that for many years, researchers have focused on single genes and clinical endpoints. The time has come, they say, to embrace rather than ignore the complexity of human traits as they are expressed by the whole genome working in concert.

"Although genetic testing for common human diseases is not yet useful, using genetic testing results to reveal an individual's ancestry is increasingly reliable," says Moore. He and PhD candidate Chantel Sloan recently mined some genetic data for a study that examined the population structure of New Hampshire residents.

Published in the September issue of PLoS ONE (a journal of the Public Library of Science), they study by Sloan and Moore and their colleagues analyzed more than 1,000 genetic markers from 864 people in New Hampshire. They discovered six subgroups of people with distinct genetic backgrounds including a group of Finnish and Russian/Polish/Lithuanian ancestry.

"I knew that people would be primarily European," says Sloan. "What I

didn't expect was the strong connection between genetic structure and people of Eastern European ancestry, which I learned is consistent with New Hampshire census and immigration data from 1870 to 1930."

Sloan used data initially compiled for a cancer study, so the genetic markers were cancer susceptibility genes rather than known ancestral genes, and the population being analyzed was not racially or geographically distinct. The results challenge the assumption that large numbers of special genetic markers are needed to discover genetically distinct groups of people.

"This is an example of how personal genetic data can be used to help inform people of their ancestry," says Moore. "Informing people of their future health is still out of reach, though."

Source: Dartmouth College