‘Healthy’ study volunteers found to harbor surprising genetic mutations

A few years ago, a team at the National Institutes of Health set out to study healthy people’s DNA. They recruited 1,000 volunteers who appeared to be free of disease based on blood tests, echocardiograms, self-reported symptoms and other things your personal doctor would normally check.

Then the scientists looked at their genomes.

What they found surprised them. In studying the A’s, T’s, C’s and G’s of the DNA they discovered that more than 100 of the volunteers had mutations that made them more susceptible to a condition like cancer, kidney disease, or even things like Birt-Hogg Dube, which is associated with skin tumors, that are very rare. And when the researchers investigated further, they verified that 34 of those individuals or their family members had been living with the disease the mutation pinpointed but had no indication this was the case. All of the participants in the study were adults 45 to 65 years of age, a time when the signs of many genetic diseases would likely already be obvious.

As DNA testing becomes more common, such findings are likely to redefine the concept of healthy. Previous studies have estimated the number of people with a genetic condition at less than 0.02 percent. The new study, published in the American Journal of Human Genetics on Thursday, indicates that the number may actually be 3 percent or more. Translated to the population of the United States that means that some 9 million people, a group the size of the population of New Jersey, may be living with a genetic condition they don’t know about.

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion and analysis. Read full, original post: NIH researcher sequence healthy volunteers’ DNA and find they aren’t so healthy afterall