Interpretations of disease risk from genetic tests inconsistent, study shows

The first report from a big public-private project to improve genetic testing reveals it is not as rock solid as many people believe, with flaws that result in some people wrongly advised to worry about a disease risk and others wrongly told they can relax.

Researchers say the study shows the need for consumers to be careful about choosing where to have a gene test done and acting on the results, such as having or forgoing a preventive surgery.

“We have very clear documentation that there are differences in what patients are getting” in terms of how tests on the same gene variations are interpreted, said the study leader, Heidi Rehm, genetics lab chief at Brigham and Women’s Hospital in Boston.

Dozens of companies now offer gene tests to gauge a person’s risk of developing various disorders. One of the newest tests on the market costs $250 and checks about 20 genes that can affect breast cancer risk.

But not all gene mutations, or variants, are equal. Some raise risk a lot, others just a little, and some not at all. Most are of unknown significance — a quandary for doctors and patients alike. And most variants are uncommon, making it even tougher to figure out which ones matter and how much.