# Cystic fibrosis carriers are at increased risk for cystic fibrosis-related conditions

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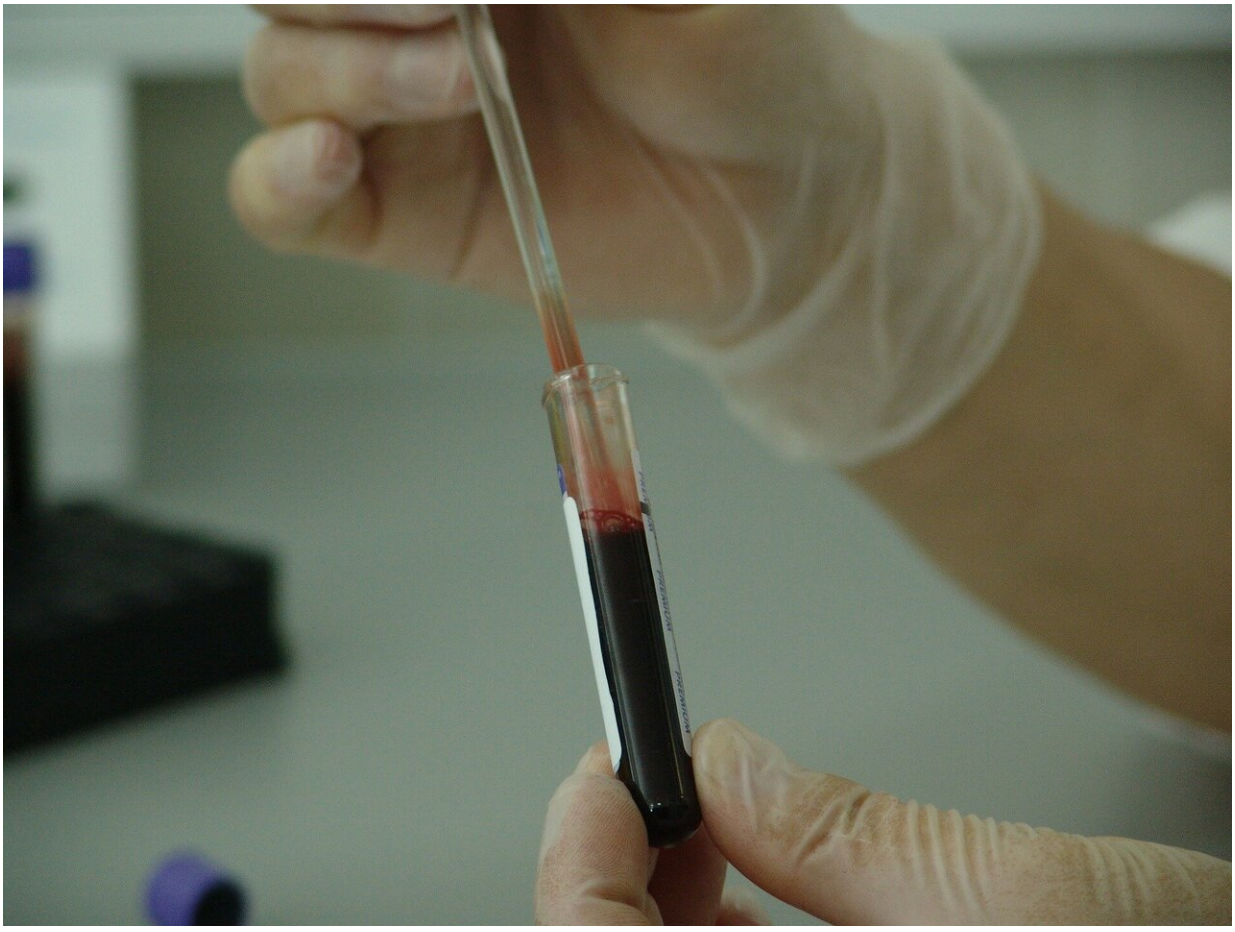

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Conventional wisdom says that having just one mutated copy of the

cystic fibrosis gene has no effects on a person's health—the disease occurs when both copies of the gene are mutated. But a new study from the University of Iowa suggests that may not be the case.

The research, published recently in the journal *PNAS*, found that people with one mutated copy of the cystic fibrosis (CF) gene, sometimes called CF carriers, are at increased risk for all of the conditions that affect people with CF.

Importantly, although the relative risk is higher, the study shows that the absolute risk—the likelihood of a CF carrier getting many of these conditions—is still very low. However, because more than 10 million Americans are CF carriers, the new findings suggest that the amount of illness caused by CF-related conditions could be substantial.

"CF carriers are nowhere near as at-risk as patients with CF," says Philip Polgreen, MD, UI professor of internal medicine and epidemiology and senior author on the study. "But compared to people with no CF mutations, they have a slightly higher risk for some diseases."

Some previous reports have linked being a CF carrier with an increased risk for some CF-related conditions, like pancreatitis, male infertility, and airway infections. The new study shows that CF carriers also have an increased risk for other CF-related conditions, including type 1 diabetes, gastrointestinal cancer, and newborn failure to thrive.

The UI team used data from the IBM Watson/Truven Health Analytics MarketScan Database, a very large database of health information, to analyze the effect of CF carrier status on increased risk for CF-related conditions.

They identified 19,802 CF carriers whose diagnosis had been confirmed by genetic testing and matched each carrier to five people without CF (a

total of 99,010 people). The analysis showed that for 59 CF-related conditions, carriers were at an increased risk, and the risk was significant for 57 of the conditions. In addition, the more prevalent a condition is in people with CF, the more prevalent it is in people who are carriers.

The researchers also constructed a "validation cohort" comprised of mothers whose children were diagnosed with CF (meaning the moms must be carriers). Health records of the women, from before their children were diagnosed, revealed similar findings to the big database study.

The findings may have implications for certain lifestyle choices. Due to the increased risk for respiratory disease or pancreatic problems, for example, Polgreen says it may be even more important for CF carriers, compared to non-carriers, to avoid excess alcohol consumption.

From a broader perspective, the researchers also think the study reveals the potential power of combining genetic testing information with health information from very large observational databases of medical records.

"More and more individuals are receiving genetic testing from their providers or from private companies, and if this information can be incorporated into health care records, there could be many new opportunities to discover both population-level health risks and individualized treatment options," says Aaron Miller, Ph.D., UI assistant professor of epidemiology and first author on the study

**More information:** Aaron C. Miller et al, Cystic fibrosis carriers are at increased risk for a wide range of cystic fibrosis-related conditions, *Proceedings of the National Academy of Sciences* (2019). DOI: 10.1073/pnas.1914912117

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