

PRINT-Decoding your DNA to prevent disease

John Rinker, DO, of OSF HealthCare, is in his mid-40s and in good health but he recently embarked on a journey that began with a quick swab inside his mouth to find out if he was at high risk of any of 10 major diseases.

The test that includes a simple cheek swab is polygenic testing. Polygenic testing focuses on the cumulative effect of many common genetic variations to determine a person's risk for developing complex diseases, while genetic testing typically focuses on a single gene or a small number of genes to identify specific genetic conditions or mutation. Why did he get the test? Dr. Rinker believes knowledge is power.

"And potentially, how is it likely if I'm affected, I want to make sure that I'm aware for my children, right? And so personally, I chose to get the test done, because if there's anything that I found, I would want to do everything in my power to make a change, if at all possible, so I can live a healthier life and a longer life."

Dr. Mark Meeker, DO, is former vice president of Community Medicine at OSF HealthCare, now vice president and chief medical officer of OSF HealthCare St. Mary Medical Center and OSF HealthCare Holy Family Medical Center. Meeker explains polygenic testing creates Polygenic Risk Scores (PRS) that aggregate the effects of thousands of genetic variants to assess a predisposition to common diseases such as cardiovascular disease, diabetes and various cancers.

"(It's not) looking for a specific change in a gene that puts you at risk of a specific disease. It's risk categorization. So, do you have an elevated risk for something like Alzheimer's or diabetes or colon cancer or breast cancer? Not 'Do you have the BRCA (Breast Cancer Susceptibility) gene, the specific gene that puts you at risk for breast cancer?' But 'Do you have a compilation of genes that puts you in a higher risk category?'"

OSF HealthCare has been offering polygenic testing at five OSF Medical Group locations including Peoria, Galesburg, Kewanee, and Pontiac. The test carries an out-of-pocket cost of \$275 because it is currently not covered by insurance. But, for some people it's worth the cost to have the knowledge or peace of mind that comes with knowing if they're at high risk for common cancers and diseases such as coronary artery, atrial fibrillation, and stroke.

Dr. Rinker has a family history of prostate cancer and fully expected he would turn up at high risk for that. However, his results showed Dr. Rinker at high risk for Alzheimer's disease. There's no cure but he says good diet, exercise and – importantly – getting enough sleep can help with prevention.

"That's definitely something I've been trying to maximize or improve, not only from an Alzheimer's perspective. But, you know, you'll find a lot of these things in the human body all interact with something else, and you're not just getting one improvement and one small outcome that you're looking at. Well, that might be what the impetus for change is (but) it's going to have a whole trickle-down effect on a whole host of other health issues or prevent a whole number of other health issues."

The target group of individuals to get polygenic testing is between 20 and 65 because that's when added screening and any lifestyle changes can have an impact. Dr. Rinker is excited about the potential to prevent disease among those who are at high risk.

"The more that we can find or identify a disease early the more we can either prevent the disease or get upstream of the illness occurring; always leads to better outcomes."

Dr. Meeker explains that polygenic tests expand the analysis beyond family history and genetic testing to broader examination of the human genome.

"If you use a questionnaire, for example, a family history and stuff to screen for certain cancers, if you add polygenic risk scores to that, you can increase the pool of high-risk people by two- or three-fold, because it catches more people than these preliminary screening tools just don't catch."

The eight-month pilot continues. Leaders will consider whether to expand the testing across the OSF Ministry by considering the number of people taking advantage of it, along with feedback from providers and patients.

While PRSs can identify individuals at higher risk, they have limitations for complex conditions influenced by both genetic and environmental factors. But, Dr. Meeker believes in the future, polygenic risk scoring will influence individual screening recommendations.

"The hope is, as we learn more in this area and get this more highly refined, that we can really target our screening so we can screen fewer people to find the ones that are really at risk, without having to screen everybody and spending all that money and having all that exposure, you know, the potential false positives."

Polygenic risk scores in the future could also help identify individuals who are most likely to benefit from specific treatments, improving the efficiency and effectiveness of clinical trials. And Dr. Meeker asserts, as research continues, more targeted, personalized approaches to care and prevention can improve overall health and wellness.

Currently polygenic testing is still in an early phase of the pilot, so it is available through five select OSF Medical Group providers in Peoria, Galesburg, Kewanee, Sheffield, and Pontiac. If you have questions, talk with your primary care provider or learn more at the [National Human Genome Research Institute](#).

Top 10 health risks included in polygenic testing:

1. Breast cancer
2. Colon cancer
3. Prostate cancer
4. Ovarian cancer
5. Coronary artery disease
6. Atrial fibrillation
7. Stroke
8. Type 1 diabetes
9. Type 2 diabetes
10. Alzheimer's disease