

Transcript of Media-Decoding DNA to prevent disease

Dr. Mark Meeker, DO, former Vice President of Community Medicine, now VP and CMO of OSF HealthCare St. Mary and Holy Family Medical Centers

Polygenic testing is not diagnostic, and it is not genetic testing that typically focuses on single gene mutations.

"(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?'" (:24)

Dr. Meeker sees how 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."

It expands on risk factors beyond family history.

"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 that these preliminary screening tools just don't catch." (:14)

John Rinker, MD, Family Medicine Physician, Chief Medical Officer of OSF HealthCare Saint James -John Albrecht Medical Center

Dr. Rinker received polygenic testing because he wanted to know if he was at high risk for any of 10 major diseases.

"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." (:23)

Dr. Rinker learned he's at a higher risk of 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.” (:26)

The target group of individuals 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.” (:12)