

**DR. CHARLES METZGER'S SATURDAY EVENING NEWSLETTER**

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**Hundreds of Genetic Variants That May Lead to Higher Cancer Risk Can Be Identified In Patients with the Invitae Common Hereditary Cancers Panel**

(Various sources)

I hope you are having a relaxing Saturday evening.

Genetic screening panels can alert patients to their potentially higher cancer risks. While they are not yet 100% reliable they can communicate the need for increased watchfulness going forward.

The United States Food and Drug Administration recently released the following:

“... the U.S. Food and Drug Administration granted de novo marketing authorization for the Invitae Common Hereditary Cancers Panel, an in vitro diagnostic test that can help detect hundreds of genetic variants associated with an elevated risk of developing certain cancers. The test can also help identify potentially cancer-associated hereditary variants in individuals with already-diagnosed cancer. The test, which is the first of its kind to be granted FDA marketing authorization, evaluates DNA extracted from a blood sample to identify variants in 47 genes known to be associated with an elevated risk of developing certain types of cancer”.

Here's the link to the press release:

[https://www.fda.gov/news-events/press-announcements/fda-grants-first-marketing-authorization-dna-test-assess-predisposition-dozens-cancer-types?utm\\_medium=email&utm\\_source=govdelivery](https://www.fda.gov/news-events/press-announcements/fda-grants-first-marketing-authorization-dna-test-assess-predisposition-dozens-cancer-types?utm_medium=email&utm_source=govdelivery).

What patients can benefit from Invitae? Here's what the University of Chicago Medical Center says:

**Who should get genetic testing?**

Traditionally, anyone should seek genetic testing for mutations if they or a close family member have one of the following:

- Diagnosis of a rare cancer (i.e., pancreatic, ovarian, male breast cancer)
- **Any metastatic prostate cancer diagnosis**
- Cancer diagnosis before age 50 (especially in a person's 20s or 30s)
- More than one cancer diagnosis in one family member
- Diagnosis of bilateral cancer in paired organs (like bilateral breast cancer)
- Three generations of cancer in a family

- Ashkenazi Jewish descent (regardless of family history)

Check out what the University of Chicago has to say about Invitae and genetic screening for cancer, here:

<https://www.uchicagomedicine.org/forefront/cancer-articles/genetic-testing-faq>

Invitae's website can be found here:

<https://www.invitae.com/>

Insurance may pay for testing. Simply ask your doctor for the test. Invitae will send a kit with swabs and instructions. Send the kit back to Invitae and you can review results on their website. And, they provide limited genetic counseling so you get answers to your questions.

Check out their site. They provide tests for much more than cancer.

### **Testing Cautions from the Harvard Medical School and University of California San Francisco.**

The University of California, San Francisco's Health Center for Clinical Genetics and Genomics has their own Common Hereditary Cancer Panel. Learn more here:

Scroll down the web page to find a list of test limitations. At this early stage of technology, a genetic test result can neither guarantee that a patient will or won't get a particular kind of cancer.

<https://genomics.ucsf.edu/content/ucsf-common-hereditary-cancer-panel>

Harvard Medical School pointed to a 2021 study finding the following:

A 2021 [study](#) published in the medical journal *JAMA Cardiology* demonstrates how direct-to-consumer testing may be misleading.

The researchers looked at genetic testing for familial hypercholesteremia. They compared the results from a comprehensive panel of genetic testing ordered by doctors (which included more than 2,000 gene variants) with results from the more limited genetic testing (24 variants) provided by 23andMe.

Among more than 4,500 people tested for a medical reason, such as evaluating an unexpectedly high cholesterol level, the more limited testing would have missed important genetic variants for

- nearly 70% of study participants
- nearly 94% of Black and 85% of Hispanic individuals
- about a third of Ashkenazi Jewish individuals.

This suggests that a large number of people would be falsely reassured by the results of their genetic tests for FH if they relied on the type of screening offered by a popular over-the-counter product. And results may be particularly unreliable among persons of color.

Here's the link to the Harvard Medical School article discussing who might get genetic testing and the need to discuss testing with our doctor:

<https://www.health.harvard.edu/blog/tempted-to-have-genetic-testing-first-ask-why-202108172571>

See you soon,

CKM