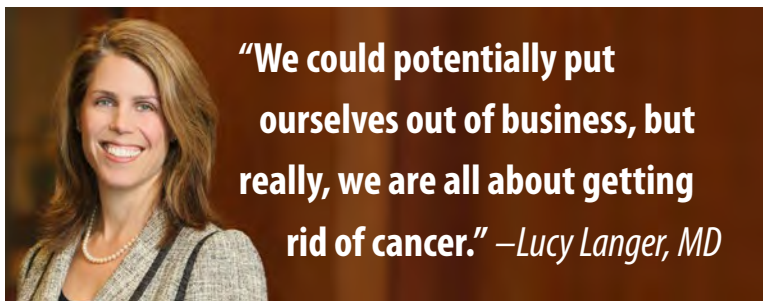
Panel-based genetic testing casts bigger net to catch signs of cancer risk

By Jon Bell
For The Scribe

When it comes to a cancer diagnosis or even sizing up someone's risk, knowledge is power. And the more knowledge there is, the better.

During the past five years or so, oncologists and patients alike have added to their knowledge base through multigene panel testing or panel-based genetic testing, a process of looking for genetic mutations that might signal a higher risk for certain types of cancer. Whereas once physicians and researchers were only able to look for one such mutation at a time, now panel testing has opened a door that allows for examination of scores of mutations in one fell swoop.

"It's now almost like a one and done," said **Lucy Langer, MD**, practice director for **Compass Oncology** and medical director of the practice's Genetic Risk Evaluation and Testing program. "The common panel is 25 genes and we can test for mutations (related to) different kinds of cancer, including breast cancer, colon, ovarian, thyroid, uterine, skin and brain. However, the more you test, the more likely you are to find something."



If the tests do detect an anomaly, Langer said patients can learn of ways to prevent cancer or detect cancer early. They don't necessarily need to panic, in large part because they've had those mutations inherited from their parents, and they have been present in their cells all their lives.

"So it's not an emergency," she said.

Additionally, because the science of genetics and mutations is still relatively young, there are many genetic alterations that aren't yet well-known or understood. As a result, it's often not clear if these changes signal an increased risk of cancer.

"It could be that the change has no impact on the gene's function at all," Langer said. "We just don't have enough information yet about many of them."

Because of the increase in uncertain test results that comes with testing more genes in one panel, companies that do genetic testing keep large databases of test results. If enough patients have the same kind of a particular alteration, and that particular alteration doesn't seem to be statistically related to any types of cancer, it can likely be ruled to be benign. Langer said despite the lack of information about some genetic mutations – and what to do about them in terms of treatment – most patients still prefer knowing about them.

"I find patients are much more comfortable knowing even if there are not clear guidelines yet," she said. "If you know you have an uncertain finding, that's where genetic counselors can come in and talk to them, and really play a role in understanding how to proceed and reduce cancer risk."

Depending on the mutation that a test identifies, there are several steps a patient could take to prevent cancer. A well-known example is for individuals with inherited mutations in the genes BRCA 1 and BRCA 2. According to the National Cancer Institute, between 55 and 65 percent of women who inherit a harmful BRCA 1 mutation will develop breast cancer by age 70. About 39 percent of women who inherit a harmful BRCA 1 mutation will develop ovarian cancer by age 70.

Women identified as having the harmful mutations may opt, after talking with physicians and a genetics counselor, to step up regular screenings, including a mammogram and a breast MRI each year. They might also begin that enhanced screening at an earlier age in the hope that, if the cancer does develop, it can be detected and treated earlier. Insurance will often cover the cost of such increased surveillance if a genetic mutation is identified.

Those with an increased risk of breast cancer also have the option of taking a medication for five years that can reduce the risk by as much as 40 percent.

Another option, depending upon the mutation and the patient's specific circumstances, is to remove the organ in question. So a patient with a high risk of ovarian cancer might have her ovaries removed, or someone with a high risk of colon cancer could have a total colectomy.

"It's really a personal choice," Langer said, "but that's when it's really important for patients to work with genetics counselors. They can engage with patients and evaluate their values and help them decide what's the best choice."

Panel-based genetic testing can also be useful in testing children who may have inherited genetic mutations from their parents. The response to a mutation might not necessarily be anything dramatic, but it would be something that parents, patients and physicians would have on their radar to monitor.

Though panel testing has been around for several years now, Langer said it continues to improve and gain widespread use. Some panels that have been developed can test for very rare mutations – those linked to, say, kidney cancer – while other large ones can test up to 99 different genes. Langer said Compass uses panel testing widely and will continue to do so in the bigger-picture aim of helping people treat, survive or even avoid cancer altogether.

"We could potentially put ourselves out of business, but really, we are all about getting rid of cancer," she said. "I think that's a long way off, but that's what we're trying to do. Cancer's not fun." ●