

In the Spotlight
**PRECISION MEDICINE &
WHAT IT MEANS FOR YOU**

BY NICOLE CHAMBERS

Precision medicine — we hear this term being thrown around but what exactly does it mean for today’s cancer patient? Simply put, it means the right test and the right treatment at the right time. Really what it boils down to is that the way patients have been receiving treatment for many years is changing quickly and dramatically due to new technologies and advancements in diagnostic testing.

Until recently, patients were treated with a “one-size-fits-all” concept. When patients are diagnosed with cancer, they usually receive the same treatment as others who have the same type and stage of cancer. Doctors use radiation, chemotherapy, immunotherapy, and surgery to treat patients. Even with patients being treated in the same or similar ways, different people may respond differently, and, until recently, doctors didn’t know why. After decades of research, scientists now understand that patients’ tumors have genetic changes that cause cancer to grow and spread. These genetic changes — that occur after you are born and throughout your life — are known as somatic mutations. Somatic testing looks at a sample of a patient’s tumor to find specific genetic and protein changes that are unique to the cancer cells.¹

With precision medicine, doctors now have the ability to look at a patient’s genes to uncover clues on how to treat the disease. They are now in the position to make treatment decisions not simply based on the specific cancer but on the genes that make up that cancer. By examining the DNA of a patient’s tumor, oncologists can identify the cancer-causing genes that make cancers grow. Speaking to *Health Matters*, Gary Schwartz, Chief of the Hematology and Oncology Division at New York-Presbyterian/Columbia University Irving Medical Center, sums this up by saying, “With the capability to now sequence DNA, we can tell the patient which genes turn cancer on and which genes turn cancer off.”² This allows doctors to understand at a genomic level what is fueling a patient’s cancer to grow.

Comprehensive Genomic Profiling (CGP) can be performed using either liquid fluid samples (blood, urine, and saliva) or by a tissue biopsy. Tissue biopsies require solid matter from the body. This specimen is removed either directly from a tumor or from bone marrow. Your doctor will advise you as to which method is better to be used specifically for you. Once the DNA in your tumor sample has been analyzed using CGP to detect mutations

and other biomarkers, your doctor may indicate if a targeted therapy, immunotherapy, or a clinical trial could be an option for you.

Genetic testing (or germline or dereditary testing) is another way oncologists are employing precision medicine. Previously, we discussed sequencing DNA and examining a patient’s genomic makeup using Comprehensive Genomic Profiling (somatic testing). Genetic testing looks at inherited traits in a person’s genes and certain cancer-relevant mutations a person could get from their parents.³ This type of testing is used to find germline mutations — inherited mutations in genes such as BRCA, PALB2, ATM, CHEK2, and others that are present at birth and significantly increase the risk of breast, ovarian, and other cancers. This type of testing is performed on blood or saliva. Certain genetic variants can make your risk of developing cancer significantly higher than that of the average person. If you have a personal or family history of cancer, genetic testing can help guide you and your healthcare provider’s decisions about your treatment or prevention plan. Genetic testing can also identify family members who may be at risk too.⁴

You may recall hearing about Angelina Jolie and her decision to have a double mastectomy since she had a genetic test performed revealing she had inherited the BRCA1 gene from her mother. Jolie’s mother died when she was just 56 and with this inherited gene, the likelihood that Angelina would develop breast cancer was 87%, and 50% likelihood of developing ovarian cancer (note this risk is different in everyone). In general, a woman who has inherited a harmful BRCA1 or BRCA2 mutation is about five times more likely to develop breast cancer than a woman who doesn’t have such a mutation, according to the U.S. National Cancer Institute (NCI). Knowing if you have a genetic variant can help you be more proactive about your health.

A new evolution in testing that is NOT considered to be precision medicine-worthy are at-home ancestry or DNA tests. Doctors are routinely asked about these tests as they have gained popularity, but they are not used in the medical community. They claim to be able to tell people more about their ancestry and other traits. But experts warn consumers to only use these tests “recreationally,” and not to rely on these tests for critical medical

information. These tests are different than what your healthcare provider would order as direct-to-consumer genetic testing provides only partial information about your health. Other genetic and environmental factors, lifestyle choices, and family medical history also affect the likelihood of developing many disorders. These factors would be discussed during a consultation with a doctor or genetic counselor, but in many cases they are not addressed when using at-home genetic tests.⁵

With the advancements made in precision medicine for oncology and the ability to find the best treatment or targeted therapy possible, all patients should be tested using a genetic and/or genomic test. These tests inform the physician on the right therapy for the patient, but they also inform the patient and their family on what has been passed down. “Even though researchers are making progress every day, the precision medicine approach to cancer treatment is not yet part of routine care for most patients.”⁶ Patients should ask their doctor if they have been tested, and if not, which testing might be best for them. Advancements are being made every day in this area, leading to a change in the way patients are treated for cancer. ●

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¹Guidelines Advise Germline and Somatic Testing, Tiffany Medina

²Dr. Gary Schwartz, <https://healthmatters.nyp.org/precision-medicine/>

³https://www.startwithstepone.com/?c3api=7641,75303615774,kwd-295923450667&gclid=EAlaIqobChMlyY39ktap5gIVENvACh1R_Q8WEAAYAAEgJzG_D_BwE#LH7KiZURIWYXq1i.99

⁴<https://www.invitae.com/en/individuals/diagnostic-genetic-testing/cancer/>

⁵<https://ghr.nlm.nih.gov/primer/dtcgenetic/dtcrisksbenefits>

⁶Precision Medicine in Cancer Treatment, National Cancer Institute