

The Value of Molecular Diagnostics:

Advancing Cancer Treatment and Care

Cancer is a major public health challenge: in 2016, more than 1.7 million people in the U.S. are expected to receive a cancer diagnosis.¹ Diagnostic tests are at the forefront of medical innovation, providing vital insights into patient health at every stage of cancer care—prevention, detection, diagnosis, treatment, and management. With the potential to fundamentally change clinical practice, these technologies are intended to match the right patient with the right type of treatment.²

Molecular Diagnostics are a critical and transformative component of cancer care, because they help health care professionals learn the specific biology of a patient's cancer and make personalized treatment decisions accordingly.

New **gene sequencing technologies** and the discovery of **new biomarkers** have spurred further investment in research that lead to new and improved diagnostic tests. Because of better diagnostic tests, patients are receiving more effective cancer care and living fuller, longer lives. Diagnostic testing can also reduce health care costs by catching cancer in its early stages, enabling earlier interventions that can be more effective, reducing unnecessary or ineffective treatments, and guiding treatments that are more likely to prevent recurrences.

Molecular Diagnostics give clinicians the tools to **personalize** testing and treatment:

- **Risk assessment:** Diagnostic tests can be used to search for biomarkers that indicate an elevated risk of developing the corresponding cancer.
Example: Women with certain variations in the BRCA1 and BRCA2 genes are six times more likely to develop breast cancer.
- **Screening:** Screening tests are often applied to patients at high risk of a certain cancer in order to identify the disease as early as possible.
Example: Pap smears to detect human papilloma virus (HPV), which is strongly linked to cervical cancer.
- **Diagnosis:** Tests for diagnosis are used to obtain a definitive diagnosis and for general cancer typing.
Example: Diagnostic tests can identify a particular leukemia subtype and best treatment options.
- **Staging and Prognosis:** Staging or prognostic tests are used to assess the severity of the cancer and/or the risk of recurrence.
Example: Diagnostic tests can determine the risk of colon cancer returning after surgery, providing guidance for whether more aggressive treatment is needed.
- **Therapy Selection:** Many molecular diagnostic tests can indicate which treatments and therapies may work most effectively, or rule out those that are unlikely to work, for each patient.
Example: One drug only works in melanoma patients who test positive for a specific mutation.
- **Monitoring:** Monitoring tests can tell a patient and their doctor whether or not a treatment is working, or give them information about the likelihood of recurrence.
Example: Physicians can look for new mutations in a gene over the course of treatment for blood cancer to monitor and assess the effectiveness of treatment.

Cancer can develop as the result of changes in the genes that control the way cells function. These changes can be inherited or developed later in life.

Mutations are errors in the genetic code that affect cellular processes

Epigenetic changes affect gene expression without altering the genetic sequence

1. Cancer Statistics. National Cancer Institute, accessed November 16, 2015 at <http://www.cancer.gov/about-cancer/what-is/cancer/statistics>.

2. Diagnostics: Advancing patient care, AdvaMedDx, accessed November 16, 2015 at <http://advameddx.org/download/files/ADxIndustryPatientCare08192012.pdf>.