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The Illumina® Cancer Discovery Initiative

Cancer is one of the leading causes of death worldwide. Increasing health care costs and the escalating prevalence of disease among aging populations have spurred the need for innovative diagnostic approaches. As with many complex diseases, the variation of genetic associations poses a challenge for a standardized treatment approach, as each manifestation can have different, multiple underlying genetic factors. Recent technological advancements in DNA sequencing and microarray-based genetic analysis offer the potential for new discoveries, leading to a revolution in cancer therapy. These developments provide hope for earlier disease detection and future treatments tailored to an individual's genetic disposition for a brighter prognosis.

What is Illumina Doing?

As a leading provider of integrated solutions that advance the understanding of genetics and health, Illumina is working to develop better, more sensitive, cutting-edge cancer diagnostic tools. Our ground-breaking next-generation sequencing technology is transforming genetic research, enabling scientists to peer into the human genome, epigenome, and transcriptome at a depth and resolution previously not achievable. With such a wealth of genetic information, researchers are able to compare data across multiple individuals and populations to identify genetic disease associations. As the technology advances, it is becoming faster, cheaper, and more accessible, enabling discoveries that can one day translate into powerful cancer diagnostic tools.

Illumina's Cancer Discovery Initiative is focused on applying our core technologies on oncology discovery, validation, and, eventually, product development. We launched this initiative by targeting ovarian, gastric, and colorectal cancers. Why these particular ones? Currently, these cancers are either detected in later stages, resulting in low survival rates and/or pose significant therapeutic challenges. Early detection and prognostic evaluation directly impact a successful patient outcome, making these areas where Illumina technology can make a difference. By working with groups to discover biomarkers for early detection and the likelihood of therapeutic response, we can help drastically improve patient care.

Cancer Discovery Roadmap

Samples → Discovery → Validation → Dx Services → Dx Products

Ovarian cancer
Colorectal cancer
Gastric cancer

Cancer type	Diagnostic dilemma	Discovery goal
Ovarian	• Most diagnoses occur in later stage • 5-year survival decreases dramatically as disease advances	• Biomarkers for early detection • Biomarkers predicting platinum therapy resistance
Gastric	• Metastasis occurs in 80-90% of individuals with gastric cancer • Six month survival rate is 65% with early stage diagnosis and <15% with advanced stages	• Biomarkers for early detection • Biomarkers predicting platinum therapy resistance
Colorectal	• 50% of cancers treated with surgery alone relapse • Therapies selected by stage of diagnosis not well associated with likelihood of response	• Biomarkers for disease prognosis and relapse

The Cancer Initiative Has Three Goals:

1. *Identify early detection biomarkers for at-risk populations*
2. *Identify markers that predict which patients will respond to therapy*
3. *Identify markers of disease prognosis*

To achieve these goals, Illumina is partnering with leading cancer research institutions to undergo a multi-stage discovery and development process. The first step is to obtain samples for tumor and normal paired specimens. Next, the genomes, methylomes, and transcriptomes from these samples will be sequenced in an effort to identify disease-associated markers followed by validation using Illumina technologies and the ability to offer diagnostic services. Ultimately, we hope to deploy novel diagnostics on Illumina's BeadXpress® platform after prospective trials and regulatory clearance. We began with the ovarian cancer initiative and are progressing rapidly towards offering services for this disease in Illumina's Clinical Laboratory Improvement Amendments (CLIA) laboratory.

Spreading Awareness

Research efforts like these can take years to translate into a diagnostic test. In the meantime, each of us can join the cause against cancer by spreading awareness about the disease. Illumina is working with the Ovarian Cancer National Alliance and the Clearity Foundation to get the word out. Visit these groups at www.ovariancancer.org and www.clearityfoundation.org to learn more.

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