17015 - Hybrid Capture DNA Sequencing Panel for Diagnosis and Detection of Lynch Syndrome and Other Cancer Types Associated with Mismatch Repair Deficiency

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Technology description

Integrative Panel to Detect Mismatch Repair Deficiency
Proper detection of the genetic and epigenetic alterations underlying cancer is critical for correct diagnosis and selection of treatment plans. In particular, determination of whether an MMR-associated cancer arose due to inherited mutations or somatic lesions in essential for guiding treatment plans of patients and, in the case of inherited cancer syndromes such as Lynch Syndrome, monitoring of family members of patients.
Here, we introduce a novel method of using hybrid-capture DNA sequencing to detect point mutations, insertions and deletions, copy number alterations, structural rearrangements, promoter methylation, and microsatellite instability (MSI) to enable diagnosis of various MMR-associated and related cancers. This hybrid-capture approach utilizes a novel targeted probe panel enabling simultaneous readout of all of the aforementioned clinically-relevant sources of genome variation.
In contrast to existing methods, this technology eliminates the need for sequential testing by multiple independent and complex tests (e.g., IHC), which are often not available in a single laboratory. This technology, therefore, has the potential to bring significant time and cost savings while providing best-in-class data quality for the detection of diagnosis of MMR-associated cancers.

Technology Description

Investigators at the University Health Network have recently developed a novel low-cost method and device that can measure the signals originating from a fluorescent biomarker in a patient and display them in a quantitative real-time, high resolution format.
Healthy tissue preservation is of the utmost importance when performing re-sectioning procedures–especially in the brain and eye. Presently, the practice of using fluorescent biomarkers in combination with 'image-guided surgery' is based only on qualitative validations. As a result, tissues fluorescing at any intensity are surgically removed. However, it has been shown that strong and weak fluorescent signals have different implications pathologically. By providing surgeons with solid quantitative

information in addition to qualitative data, this invention has the potential to drastically improve not only clinical decision making but also diagnostic accuracy.
This quantitative technique is based on sound biophotonic principles, and is supported by both in-vivo and ex-vivo testing. Quantitative information from this technology has additional advantages because it eliminates inter-observer variations, provides a linear response to biomarker concentration, minimizes not only the effect of tissue-to-detector geometry but also optical tissue properties and reduces the influence of auto fluorescence from cells. The technology is compatible with microscopes, macroscopes, confocals, endoscopes, bronchoscopes and laparoscopes. The clinical feasibility of this technology has been successfully demonstrated in patients undergoing prostatectomy during clinical trials at the University Health Network and for brain gliomas at the Dartmouth Medical School.

Images

Image: Schematic representation of Lynch Syndrome vs. sporadic (somatic) colorectal cancer with regards to inactivation of selective MMR genes. Source: Sinicrope, Nature Reviews Clinical Oncology (2010).

Application area

Detection and diagnosis of Lynch Syndrome vs. somatic colorectal cancer
Detection and diagnostic subtyping of endometrial cancer
Detection and diagnosis of Constitutional Mismatch Repair Deficiency (CMMRD) in pediatric patients
Detection of mismatch repair (MMR) deficiency-associated microsatellite instability (MSI)

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