

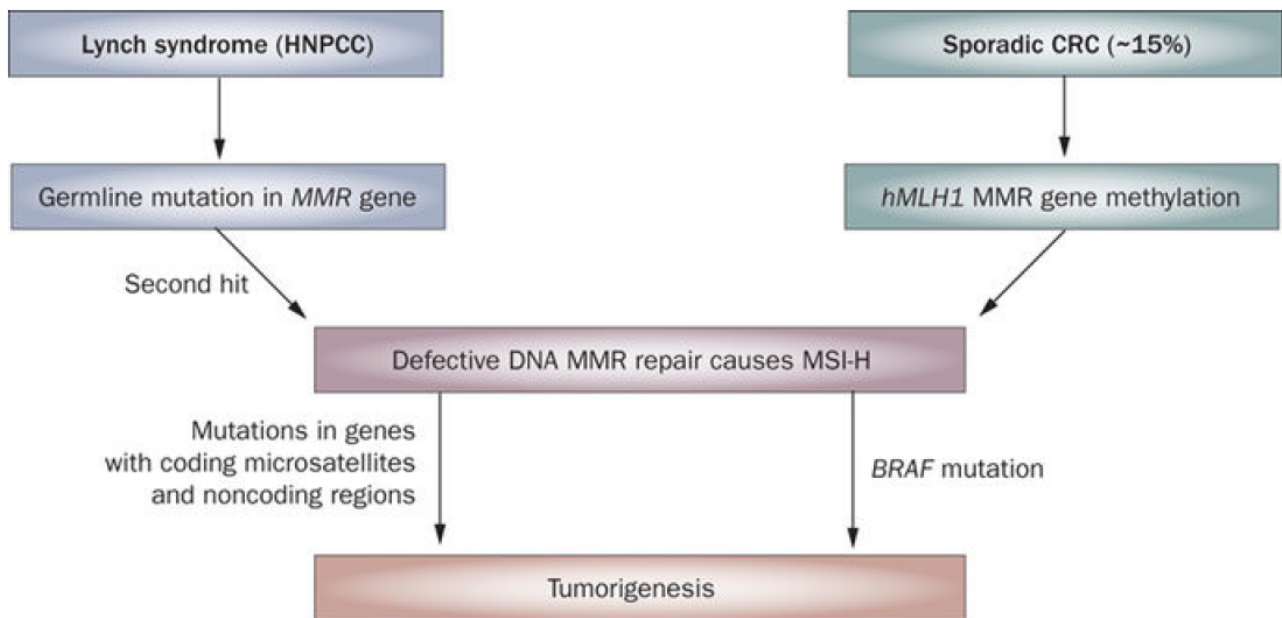
Hybrid Capture DNA Sequencing Panel for Diagnosis and Detection of Lynch Syndrome and Other Cancer Types Associated with Mismatch Repair Deficiency

Published date: May 29, 2018

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 is 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.



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)

Institution

[University Health Network](#)

Inventors

[Trevor Pugh](#)

联系我们



叶先生

电话 : 021-65679356

手机 : 13414935137

邮箱 : yeyingsheng@zf-ym.com