FusionSync detection technology

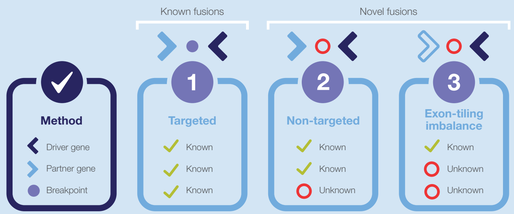
Detect novel but rare fusions, without missing the known and common

NEW

FusionSync™ detection technology, utilized in the next-generation sequencing (NGS)–based Oncomine™ Precision Assay, represents a synchronous solution optimized for clinical biomarker research. The process enables simultaneously comprehensive and sensitive gene fusion detection based on RNA analysis.

A lot has been said recently about the importance of novel fusion detection capabilities, but let's not forget that most fusions found in cancer samples involve the same driver genes and partners that are known. Yet, those can be present at a very low level, or in samples with very little material to test.

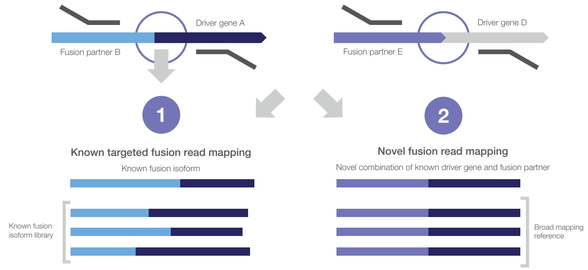
FusionSync detection technology



Targeted detection of known fusions and novel combinations of known fusion partners

Employing hundreds of fusion isoforms across varying gene partners and breakpoints between driver and partner genes assures sensitive and specific detection of known fusions, which represent the majority detected. This highly sensitive and robust methodology has the ability to detect tiny levels of a tumor-specific fusion transcript in background normal RNA and with minimal input down to 10 ng.

Differential read mapping strategies for known and novel fusion detection



Panel primers are designed to target specific exon–exon junctions of fusions where the driver gene, the partner gene, and the breakpoint between these two genes are known. The sequencing reads are mapped to a reference file that contains only the known gene fusions. Gene-specific primers situated on either side of the fusion breakpoint generate a fusion amplicon containing sequences from both partners.

Panel primers are used to detect fusions between novel combinations of known driver and partner genes. The sequencing reads are mapped to a broader reference, such as the whole exome. Mapping the reads to a broader reference allows for the detection of multiple configurations of driver and partner genes as well as detection of novel breakpoints between the known partner and driver genes.

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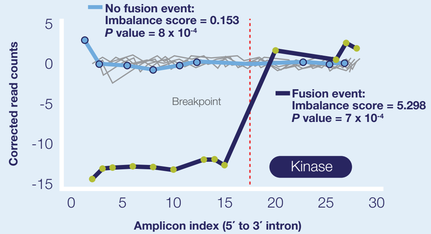
Exon-tiling imbalance approach to detect novel fusions

A partner-agnostic method that enables the detection of novel fusions and breakpoint prediction.



The software measures the intragenic 3′ to 5′ expression ratio for each gene and compares the ratio to the baseline (normal sample). Genes that do not undergo a fusion event are expected to have a 3′ to 5′ expression ratio similar to the baseline. Genes that undergo a fusion event typically have a 3′ to 5′ expression ratio greater than the baseline. This will be indicated with the statistical confidence of the imbalance score and *p* value. For each driver gene in which fusion was detected, the software also predicts the most likely position of the fusion breakpoint. The relative position of the breakpoint (red line) to the kinase domain of the driver gene is very important. If the breakpoint is found to be in the kinase domain, the fusion will be "non-activating."

Benefits of FusionSync detection technology—let's put everything in perspective

FusionSync detection technology, available with the Oncomine Precision Assay on Genexus NGS System, provides sensitive detection of fusions, both known and novel

One-day turnaround enabling you to combine your lab's immunohistochemistry (IHC) results with timely NGS insights to deliver a comprehensive report in one day*

Two touchpoints and 10 minutes of hands-on time, making the workflow easy and convenient even if your lab is new to NGS technology, and helping improve productivity for any lab

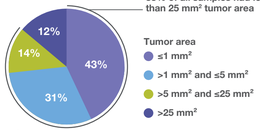
Less sample required compared to other technologies, meaning many more samples can be successfully tested

Tissue and plasma samples
- One test, one workflow, multiple sample types
- Maximizes samples that can be tested

Tissue is an issue

- Routine clinical research samples consist of small biopsies or fine needle aspirates
- Many NGS methods require much higher sample input of 50–1,000 ng

88% of all samples received in one laboratory during one year had less than 25 mm² tumor area and therefore could not be tested by methods requiring larger sample input (>50 ng of nucleic acid).**



* Specimen-to-report workflow will be available after the Ion Torrent™ Genexus™ Purification System and integrated reporting capabilities are added in 2020.
** NGS to take top spot as cancer biomarker testing broadens. *CAP TODAY* (June 2018).

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