PRODUCT DATASHEET

Custom Targeted Sequencing

Highly scalable solution for the discovery of novel variants across genomic target and sample numbers

Unique Molecular Indexing (UMI) and duplex consensus calling allows for detection of rare novel variants

OVERVIEW

Broad Genomic Services provides a custom target enrichment product to support a wide range of somatic studies that require high sensitivity and specificity for low allele fraction variants. This product incorporates extremely stringent error correction using duplex Unique Molecular Indexing (UMI) and ultra deep targeted sequencing, making it particularly well suited for blood biopsy cfDNA as well as bone marrow aspirate clonal hematopoiesis studies. By combining array-synthesized custom panels, and our workflow, we can now rapidly and in a cost effective manner target disease and study-specific genes to high depth. Panel sizes can range from 10's of kilobases to multiple megabases in size and routinely achieve greater than 90% of sequenced bases on or near target, maximally utilizing gigabases of sequencing purchased. Enabling you to sequence only the targets you need, to extreme depth (20,000-100,000X raw coverage), maximizing the likelihood of detecting variants of importance at very low allele fractions.

UNIQUE MOLECULAR INDEXING AND DUPLEX CONSENSUS CALLING

Adapters with duplex UMIs allow individual DNA molecules to be "tagged" before amplification. The duplex nature of the UMI adapters enable the reconstruction of original molecules after sequencing, a second level of marking duplicate reads to develop duplex consensus reads, and highly accurate error correction for the duplex consensus reads when needed. Duplex consensus calling greatly reduces the false positive rate seen and has allowed for greater than 90% sensitivity for SNV's at a 1% MAF.

WHAT'S INCLUDED

- Project Design Consultation
- Custom Panel Design and QC
- Sample Receipt and Incoming QC
- Sample Preparation (LC) and QC
- Pilot Sequencing and Production Sequencing
- Data Delivery

INPUT REQUIREMENTS

- Due to the nature of custom projects, input requirements will be project specific, we typically require:
  - 5-50ng of cfDNA (@ 0.5ng/uL minimum concentration)
  - 100-200ng gDNA (@ 2ng/uL minimum concentration)
- Minimum sample data including Collaborator Participant ID and Collaborator Sample ID

EXAMPLE PERFORMANCE METRICS

	Panel 1	Panel 2	Panel 3	Panel 4
Number of Targets	763	8,874	8,874	7,899
Target Bases	136,085	1,659,149	1,659,149	2,077,067
Coverage Deliverable	100,000X Raw Coverage	25,000X Raw Coverage	250X Mean Target Coverage	10,000X Raw Coverage
Duplex Consensus Calling	Yes	Yes	No	Yes
Unique Molecular Indexing	Yes	Yes	Yes	Yes
% Selected Bases	91.0	88.7	89.2	84.6
% Target Bases at 2X	100.0	99.7	99.1	99.4
% Target Bases at 50X	100.0	99.7	98.4	99.2
% Target Bases at 100X	99.8	99.6	97.5	99.1
Raw Coverage	115,939X	28,301X	1,242X	10,621X
Mean Target Coverage	7,547X	2,579X	411X	1,910X

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GENOMIC SERVICES

MORE INFORMATION

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