Agilent OneSeq Target Enrichment

Comparison Study

Benefits

- **ONE Revolutionary Assay**
  Comprehensive, all-in-one detection of genome-wide CNVs, LOH, and targeted mutations.
- **ONE Simple Solution**
  Agilent SureCall software enables you to analyze, visualize, and contextualize OneSeq data using a single application.
- **ONE Streamlined Workflow**
  Obtain more information in a single reaction.
  Gain more information cost-efficiently with only 7 Gb of sequencing, compared to deep 30x WGS.

Overview

Agilent OneSeq Target Enrichment offers a simple, streamlined, cost-effective solution for detecting single gene mutations, CNVs, and aneuploidies versus Whole Genome Sequencing (WGS). The current cost and turnaround time of deep coverage WGS prevent it from being used in high-throughput clinical research laboratories.

The OneSeq Target Enrichment kits are based on the Agilent SureSelect technology, and consist of a set of backbone baits for genome-wide copy number change detection by comparing an experimental sample to a known reference sample. The majority of the baits target genomic regions with high minor allele frequency SNPs, allowing for the detection of cnLOH and UPD. Additional baits that target specific regions of interest allow for the detection of mutations and indels.

The catalog OneSeq Constitutional Research Panel (Figure 1) is a 28 Mb design. It includes baits (12 Mb) for a functional copy number resolution of 300 kb and cnLOH resolution of 5 Mb in the genome-wide backbone, and a higher 25–50 kb resolution in disease-associated ClinGen regions. It also includes all content (16 Mb) from the Agilent SureSelect Focused Exome Panel targeting disease-associated genes. The OneSeq CNV Backbone + Custom Panel allows for the addition of the genome-wide backbone to any custom target gene panel, up to 12 Mb, using Agilent SureDesign, a free web-based design application.

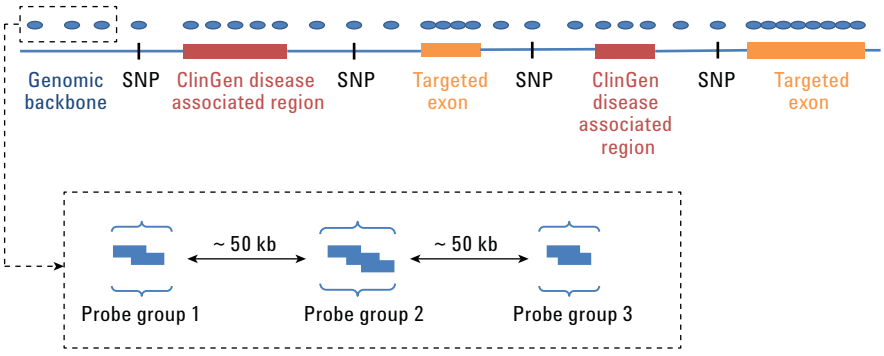

Figure 1. Bait design schema used for Agilent OneSeq Target Enrichment.

	DNA samples	Aberrations	Affymetrix CytoScan	Illumina CytoSNP	Agilent 4x180K	Illumina WGS	Agilent OneSeq
CNV (134 to 620 kb)	NA03997	134 kb CNV 3/2	✓	✓	✓	✓	✓
	NA16455	163 kb CNV 3/2	✓	✓	✓	✓	✓
	NA11419	230 kb CNV 3/2	✓	ND	✓	✓	✓
	NA08254	370 kb CNV 3/2	✓	✓	✓	✓	✓
	NA20409	503 kb CNV 1/2	✓	✓	✓	✓	✓
	NA03997	620 kb CNV 1/2	✓	✓	✓	✓	✓
CNV (>1Mb)	NA03997	20 Mb CNV 3/2	✓	✓	✓	✓	✓
	NA08254	14 Mb CNV 1/2	✓	✓	✓	✓	✓
	NA16455	-18q (6 Mb)	✓	✓	✓	✓	✓
	NA16447	-18q (13 Mb)	✓	✓	✓	✓	✓
	NA50122	-18q (16 Mb)	✓	✓	✓	✓	✓
	NA02948	Trisomy 13	✓	✓	✓	✓	✓
	NA01359	Trisomy 18	✓	✓	✓	✓	✓
	NA04592	Trisomy 21	✓	✓	✓	✓	✓
cnLOH	NA20409	UPD	✓	✓	✓	ND	✓
	NA20408	UPD	✓	✓	✓	ND	✓
INDELs	NA09551	13 bp	ND	ND	ND	ND	✓
	NA16382	26 bp	ND	ND	ND	ND	✓

Table 1. Thirteen Coriell samples with 18 known aberrations were analyzed with three different microarray platforms, Agilent OneSeq Target Enrichment, and low-pass (1-2x) WGS. All large copy number changes could be detected with all platforms. Using our analysis method, one small copy number change could not be detected using Illumina microarrays. As expected, INDELs could not be detected with any of the microarray platforms. The read-depth of the low-pass WGS was too low to be able to detect UPD. In conclusion, the different assays were not able to detect all aberrations, whereas Agilent OneSeq Target Enrichment was able to detect all expected aberrations.

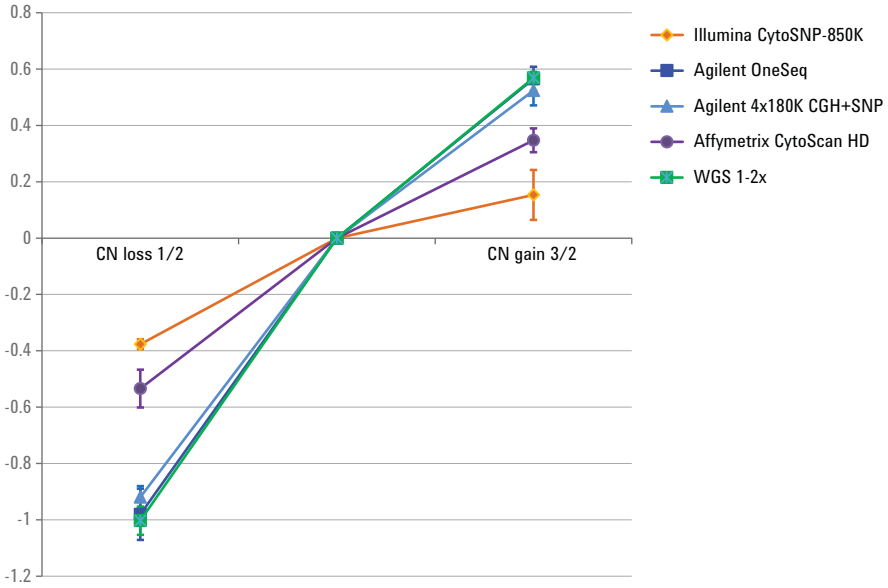

Figure 2. Plot of measured $\log_2$ ratios compared to theoretical values for the chromosomal aberrations described in Table 1 representing 1, 2, and 3 copies. Agilent OneSeq, Agilent 4x180K CGH+SNP microarrays, and WGS gave $\log_2$ ratios for all regions close to the theoretical values. In contrast, $\log_2$ ratios for the Affymetrix and Illumina microarrays were compressed compared to the theoretical values.

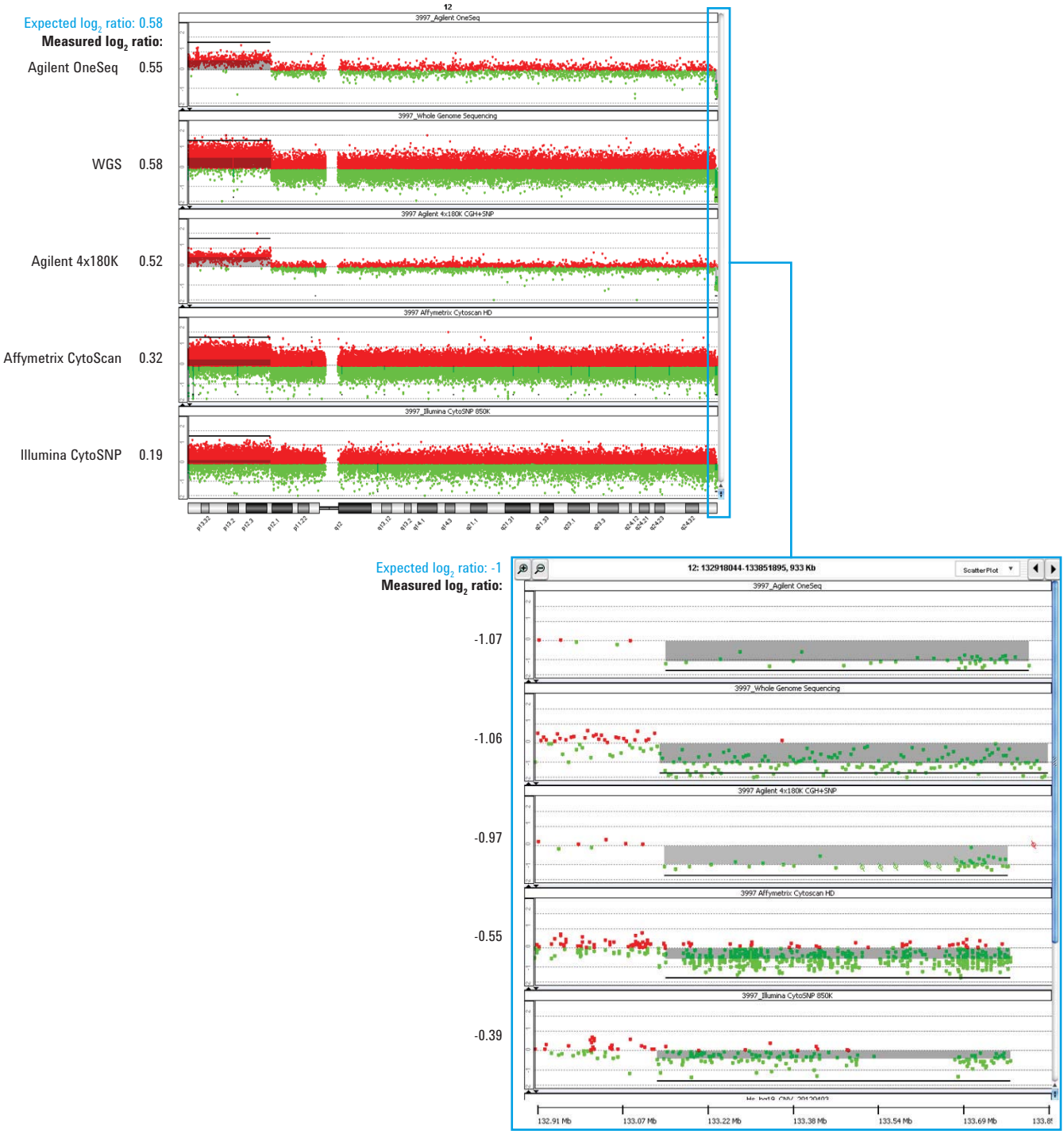

Figure 3. The known copy number gain of part of the p-arm and the known copy number loss in the telomeric region of chromosome 12 in Coriell sample NA03997 was detected on three different microarray platforms, Agilent OneSeq, and low-pass WGS. The average log_2 ratio for each aberration is indicated next to the graphs, and the values for Agilent OneSeq were very close to the theoretical log_2 ratios. The data are shown in Agilent Genomic Workbench Software. Settings used to analyze the log ratios were: ADM-1, threshold 7.0, fuzzy zero on, minimum number of probes 5, minimum absolute log ratio 0.09.

Ordering Information

	Description	Part number
Target enrichment for SNPs, LOH, and genome-wide CNV detection	Agilent OneSeq Constitutional Research Panel, 16 rxn	5190-8702
	Agilent OneSeq Constitutional Research Panel, 96 rxn	5190-8703
	Agilent OneSeq Constitutional Research Panel, 96 rxn Auto	5190-8704
	Agilent OneSeq CNV Backbone + Custom Panel (up to 12 Mb design)	Please refer to the following website for custom part number configurations: www.genomics.agilent.com
Library prep regions	Agilent SureSelect Reagent Kit, HSQ, 16 rxn	G9611A
	Agilent SureSelect Reagent Kit, HSQ, 96 rxn	G9611B
	Agilent SureSelect Reagent Kit, HSQ, 96 rxn Auto	G9641B
Reference DNA	Agilent OneSeq Reference DNA, Male, 1 vial, 50 rxn	5190-8848
	Agilent OneSeq Reference DNA, Female, 1 vial, 50 rxn	5190-8850

FOR MORE INFORMATION
www.agilent.com/genomics/oneseq

NGS resource page
www.agilent.com/genomics/NGSResource

U.S. and Canada, call 800-227-9770 or for other regions, consult www.agilent.com/genomics/contactus

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