

NimbleGen 454 Optimized Sequence Capture 385K Arrays

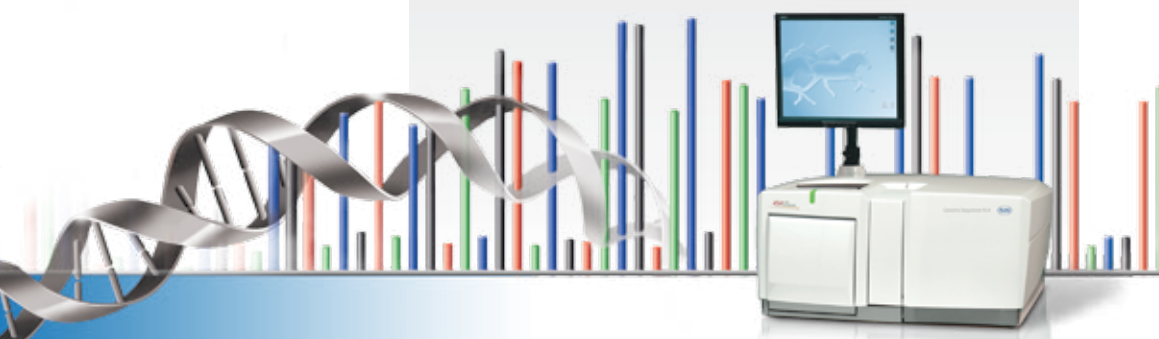
Targeted resequencing simplified

Quickly identify causative mutations in disease-associated regions

A major challenge for many researchers in human disease studies is to sequence disease-associated regions to understand the causative mutations. Such regions are often identified by genome-wide associated studies (GWAS) with sizes ranging from a few hundred kilobases to a few megabases. Targeted resequencing of such regions with traditional technologies, such as PCR and capillary sequencing is extremely labor intensive and costly.

The NimbleGen 454 Optimized Sequence Capture 385K Array capitalizes on the highly efficient enrichment of NimbleGen Sequence Capture technology and has been optimized to work seamlessly with the high-throughput sequencing of the Genome Sequencer FLX System from 454 Life Sciences. Together these Roche technologies combine to offer a powerful sequencing solution for large-scale targeted resequencing studies to significantly reduce time, labor and cost while improving data quality.

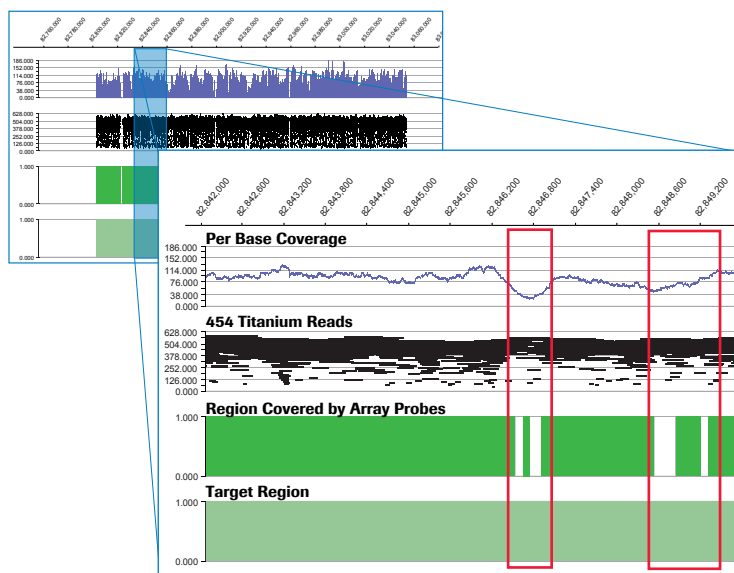
- **Target any region of interest:** Capture up to 5Mb of contiguous or non-contiguous genomic regions on a single array.
- **Generate data with confidence:** Ensure a high level of specificity and uniformity with an empirically optimized and validated design algorithm. Utilize built-in control probes to ensure system performance.
- **Utilize seamless workflow:** Highly optimized workflow reduces hands-on time, lowers sample requirements from 20µg to 5µg, and increases the usable sequence by 10%.
- **Detect important variants:** The unique 400bp long reads from the GS FLX Titanium Series enable easy detection of small indels, improve coverage in repetitive regions, and provide haplotype information.
- **Easily analyze data:** Dedicated GS Reference Mapper software reports variant locations, amino acid changes in coding regions, and known SNP information. In addition, the GS Mapper software generates capture performance metrics, such as percentage reads in target regions.



For life science research only.
Not for use in diagnostic procedures.

Optimized workflow – enrichment, sequencing, and data analysis

NimbleGen Sequence Capture technology is a revolutionary single step process for the enrichment of large, selected genomic regions from full-complexity human genomic DNA. This technology has now been fully optimized for the GS FLX Titanium Series workflow, adding 454 adaptors to fragmented genomic DNA before array hybridization, eliminating a step in the workflow and streamlining downstream sequencing processing. Roche offers a complete solution of kits, arrays and instruments specifically designed to optimize the workflow, reduce processing time, minimize costs, and enhance data quality. GS Reference Mapper software provides integrated analysis of the high-quality data and allows the researcher to more quickly and efficiently discover variants.



▲ **Figure 1: High-Performance Targeted Resequencing in a 250kb Target Region.** The system offers high specificity (most GS FLX Titanium Series reads fall into the target region) and uniform capture (high per base coverage achieved across the target region). The long reads of the GS FLX Titanium Series provide a significant coverage advantage as illustrated by continuous coverage in small regions where no probes were designed due to repetitiveness (red boxes). Refer to Table 1 for experimental details.

Resequencing of HapMap Research Sample

Experiment	Sequencing Run	Total Reads	Total Bases	On-Target Reads	Median Coverage	Target Bases with 10+ Coverage	Target Bases with 1+ Coverage	No. of Known SNPs in Target Region	No. of Known SNPs Called Correctly	SNP Detection Rate
250kb - 1	1/8 PTP	70190	27646394	75.2%	85	97.3%	98.6%	273	266	97.4%
1Mb - 1	1/4 PTP	140374	55453593	87.3%	49	92.8%	96.9%	832	803	96.5%

▲ **Table 1:** NimbleGen 454 Optimized Sequence Capture 385K arrays were used to capture both a 250kb contiguous region, and a 1Mb contiguous region in the human genome from a HapMap DNA sample. Data shown is from one of the four independent experiments for each region. (Additional data is available at www.nimblegen.com) Experiments were performed by external evaluators for each design. Sequencing data were generated using Genome Sequencer FLX System and GS XLR70 Sequencing Kit on PicoTiterPlate (PTP) device. The PTP is divided into 8 regions by a gasket, and each sample from a 250kb capture was sequenced by one of the 8 regions (1/8 PTP), while each sample from 1Mb capture is sequenced by two of the 8 regions (two 1/8 PTP, or roughly equal to one region from a 4 region gasket {1/4 PTP}). Data were analyzed using the GS Reference Mapper software, and SNP calls were compared to known HapMap SNPs.

Ordering Information

Catalog No.	Pack Size	Description
05 478 731 001	1	NimbleGen 454 Optimized Sequence Capture 385K Array
05 967 848 001	50	NimbleGen 454 Optimized Sequence Capture 385K Arrays, 50 pack
05 967 856 001	100	NimbleGen 454 Optimized Sequence Capture 385K Arrays, 100 pack

For a list of accessories required to process these arrays, download the *NimbleGen Arrays User's Guide: 454 Optimized Sequence Capture Array Delivery* from www.nimblegen.com/seqcap

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