

# SeqCap EZ Exome Library and 454 GS FLX Titanium Series

## *Genetic Discovery Made Easy*

Key breakthroughs of human genetic disease research rely on understanding the molecular basis of the diseases. The majority of known disease-causing mutations reside in coding regions in the genome, which comprise the most functionally relevant and best understood 1% of the human genome, the exome.

Targeted sequencing of exomes is an innovative technique to identify causative mutations for genetic disorders (both Mendelian and complex diseases) quickly and cost-effectively. By using SeqCap EZ Exome Library followed by next-generation sequencing with the 454 GS FLX System, a complete exome can be sequenced for a fraction of the cost of whole-genome sequencing.

This cutting-edge technology utilizes a single tube of stable DNA oligos to capture all exons in solution, comes with a simple, scalable workflow, and includes built-in controls to evaluate capture performance. With 2.1 million empirically optimized DNA probes, SeqCap EZ Exome Library delivers a high level of performance and substantial savings on sequencing. Together, SeqCap EZ Exome Library and the fast and accurate 454 GS FLX Titanium series chemistry, now allow researchers to obtain high-quality data to help identify important variants in the exome.

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

### Seamless, Easy, and Scalable Workflow

- Target the exome using a single tube of stable DNA oligos
- High performance capture and sequencing all from one reliable provider

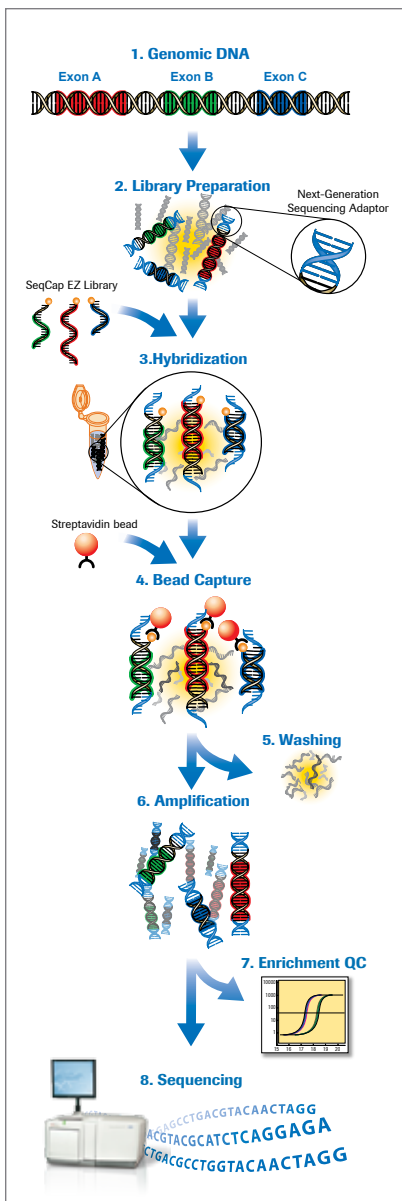
### Catch all important variants

- The unique 400bp long reads enable easy detection of small indels, give better coverage in repetitive regions and intronic regions including splicing sites, and provide haplotype information

### Analyze data easily

- Dedicated GS Reference Mapper Software reports all classes of variants and generates capture performance metrics





▲ Figure 1: SeqCap EZ Exome Protocol.

HapMap Rates		All Differences	
Overall False Positive Rate		0.05%	
Overall False Negative Rate		13.86%	
Insertions and Deletions Called			
Size	Insertions	Deletions	Total
1+	4,243	5,213	9,456
3+	1,105	1,479	2,584
9+	159	254	413
15+	66	113	179
Larger Structural Variations Called		538	

```
>chr4 170837604 170837643 GCGGGAGTCGGAGACGAGGGAGAGGGGGAGACCTGGAA - 13 62% + CLCN3 rs66819520 ,rs55701258,rs62333177,rs62333178
Reads with Difference:
chr4 170837572+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAAAGCGGGAGTCGGAGACGAGGGAGAGGGGGAGACCTGGAAAGCGGGAGTGGAGACGAGGG-AG 170837672
.....
F30EVM02FUHD6 244+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAA-----A--GCGGGAGTGGAGACGAGGG-AGAGGGGAG 305
F30EVM01EIFR0 217+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAA-----A--GCGGGAGTGGAGACGAGGG-AGAGGG-AG 277
F30IE302ID9F8 214+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAA-----A--GCGGGAGTGGAGACGAGGG-AGAGGG-AG 274
F30EVM02GRE2M 154+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAA-----A--GCGGGAGTGGAGACGAGGGAGAGGGGGAG 216
F30IE301BRXVA 139+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAA-----A--GCGGGAGTGGAGACGAGGG-AGAGGG-AG 199
F30EVM02GALU9 292- TCCAGCCTTGGCAACAGAGGGAGACCTGGAA-----A--GCGGGAGTGGAGACGAGGG-AGAGGG-AG 232
F2YHYH02G5AFQ 344- TCCAGCCTTGGCAACAGAGGGAGACCTGGAA-----A--GCGGGAGTGGAGACGAGGG-AGAGGG-AG 284
F30EVM01CH1NU (2) 332- TCCAGCCTTGGCAACAGAGGGAGACCTGGAA-----A--GCGGGAGTGGAGACGAGGG-AGAGGG-AG 272
.....
Other Reads:
.....
F30IE302F293Y (2) 171+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAAAGCGGGAGTCGGAGACGAGGGAGAGGGGGAGACCTGGAAAGCGGGAGTGGAGACGAGGG-AGAGGG-AG 271
F30IE301BUC2F 292- TC-AGCCTTGGCAACAGAGGGAGACCTGGAAAGCGGGAGTCGGAGACGAGGGAGAGGGGGAGACCTGGAAAGCGGGAGTGGAGACGAGGG-AGAGGG-AG 193
F30IE302FXB8S 54+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAAAGCGGGAGTCGGAGACGAGGGAGAGGGGGAGACCTGGAAAGCGGGAGTGGAGACGAGGG-AGAGGG-AG 154
F2YHYH01A0U9S (2) 15+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAAAGCGGGAGTCGGAGACGAGGGAGAGGGGGAGACCTGGAAAGCGGGAGTGGAGACGAGGG-AGAGGG-AG 115
F30IE301EY7B7 2+ TCCAGCCTTGGCAACAGAGGGAGACCTGGAAAGCGGGAGTCGGAGACGAGGGAGAGGGGGAGACCTGGAAAGCGGGAGTGGAGACGAGGG-AGAGGG-AG 102
.....
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▲ Figure 2 : Example of a deletion heterozygote

Ordering Information:	Catalog No.	Pack Size	Description
	05 933 374 001	4	SeqCap EZ Human Exome Library LR, 4 reactions
	05 933 382 001	48	SeqCap EZ Human Exome Library LR, 48 reactions
The design targets 180,000 coding exons (from CCDS database) and ~550 miRNA exons (from miRBase). SeqCap EZ Human Exome Library LR is optimized for long read sequencing with the 454 GS FLX System.			
Annotation files and a list of accessories required to process the SeqCap EZ Exome Library are available at <a href="http://www.nimblegen.com/seqcapEZ">www.nimblegen.com/seqcapEZ</a> .			

▶ Table 1: Genetic variants are readily detected by targeted 454 GS FLX System sequencing of samples prepared by SeqCap EZ Exome Library. HapMap sample NA11881 was used in this experiment, and the sequencing data was analyzed with GS Reference Mapper software. Data obtained from two GS FLX Titanium Series sequencing runs.

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