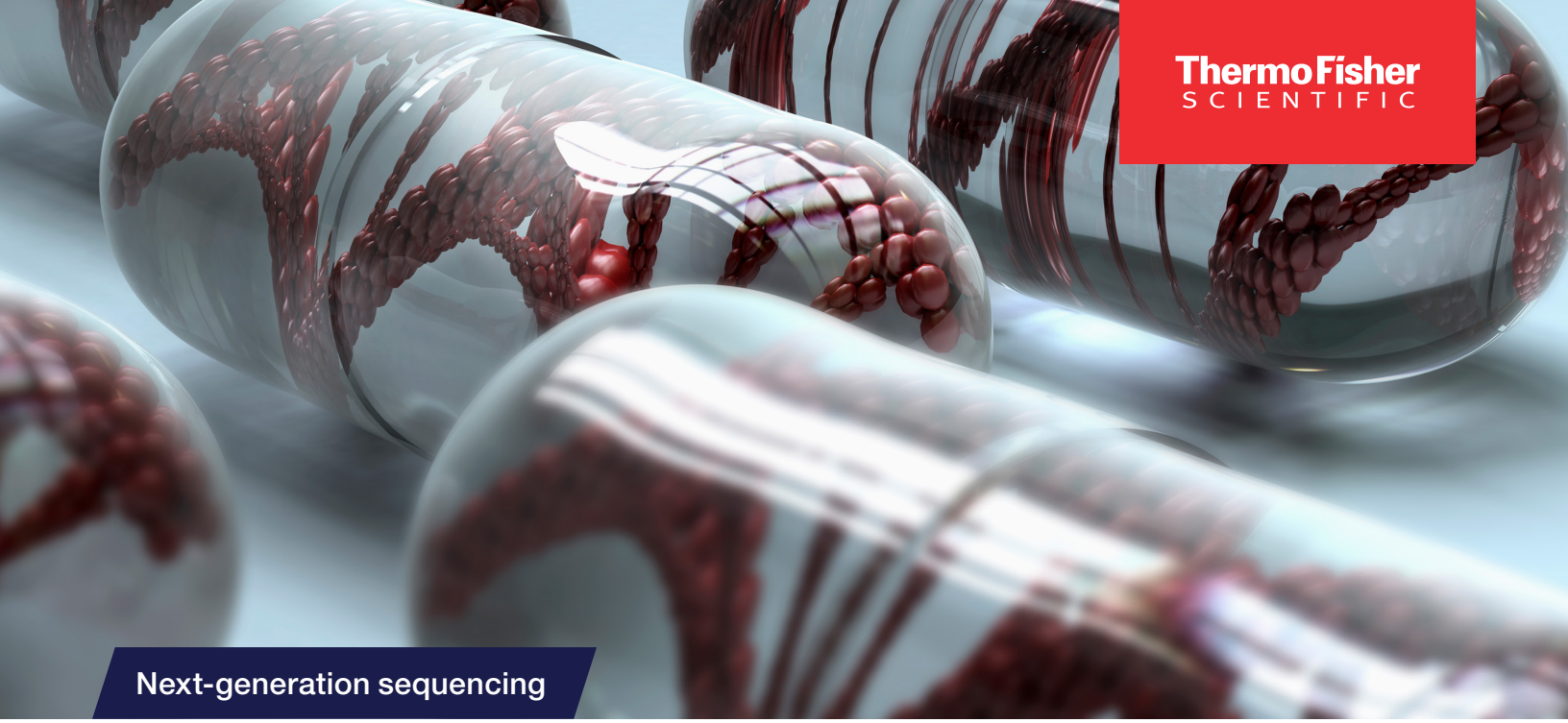

Next-generation sequencing

# Complete next-generation sequencing solution for pharmacogenomics research

## Ion AmpliSeq Pharmacogenomics Research Panel

The Ion AmpliSeq™ Pharmacogenomics (PGx) Research Panel is ideally suited to clinical research studies investigating how genomic variations in drug metabolizing enzymes (DMEs) impact drug efficacy and safety. The panel is part of a complete end-to-end next-generation sequencing (NGS) research solution for genotyping single-nucleotide polymorphisms (SNPs), as well as insertion/deletion (indel) and copy number variation (CNV) analysis in 40 known DMEs (Figure 1). The panel focuses on 136 well-documented SNPs and indel variants, and captures *CYP2D6* copy number variation at both the gene and exon 9 levels, enabling the screening of broad selection of haplotypes including *CYP2D6*36* alleles.

The Ion AmpliSeq Pharmacogenomics Research Panel utilizes Ion AmpliSeq™ chemistry to offer:

- Customizable content, allowing addition or removal of gene targets using Ion AmpliSeq Designer
- Outstanding performance on buccal swab samples from only 10 ng of input
- High coverage and unbiased amplification, resulting in robust performance and minimizing sample reruns
- Genotyping and copy number analysis in a single workflow
- Sample multiplexing flexibility enabling processing of 8–96 samples in a single batch
- Integrated downstream analysis solution

| Ion AmpliSeq Pharmacogenomics Research Panel | |
|---|---|
| **Total amplicons** | 119 |
| **Genes covered** | 40 |
| **SNV/indels detected** | 136 |
| ***CYP2D6* CNV events*** | 2 |

* CNV detection at gene level and exon 9(*36) level.

| | | | | |
|---|---|---|---|---|
| *ABCB1* | *CYP2C8* | *F2* | *MTHFR* | *VKORC1* |
| *ABCG2* | *CYP2C9* | *F5* | *OPRD1* | *HLA-A*3101* |
| *ADRA2A* | *CYP2D6* | *GABRA6* | *OPRK1* | *HLA-B*5701* |
| *ANKK1* | *CYP3A4* | *GABRP* | *OPRM1* | *HLA-B*1502* |
| *APOE* | *CYP3A5* | *GRIK4* | *SLCO1B1* | |
| *COMT* | *DBH* | *HTR2A* | *TPMT* | |
| *CYP1A2* | *DPYD* | *HTR2C* | *UGT1A1* | |
| *CYP2B6* | *DRD1* | *ITGB3* | *UGT2B15* | |
| *CYP2C19* | *DRD4* | *KIF6* | *UGT2B7* | |

**Figure 1. The Ion AmpliSeq Pharmacogenomics Research Panel analyzes genetic variants including SNPs, indels, and CNVs associated with 40 DMEs.** Panel design is customizable, enabling flexible editing to an existing gene target list.

ion torrent

# Complete sample-to-results workflow solution

The Ion AmpliSeq Pharmacogenomics Research Panel is part of a complete NGS workflow solution, enabling rapid and flexible analysis of buccal swab samples in 2 days, from start to finish (Figure 2). Each step in the workflow can be automated with minimal hands-on time. A flexible system together with a broad spectrum of throughput needs enables parallel processing of 48–384 samples in a single workflow—reducing data turnaround time significantly when project size is variable. An integrated data analysis solution completes the process, providing report-ready haplotype answers.

Our support team is ready to help new and existing customers set up and maintain all components of the system, enabling you to fully focus on your projects.

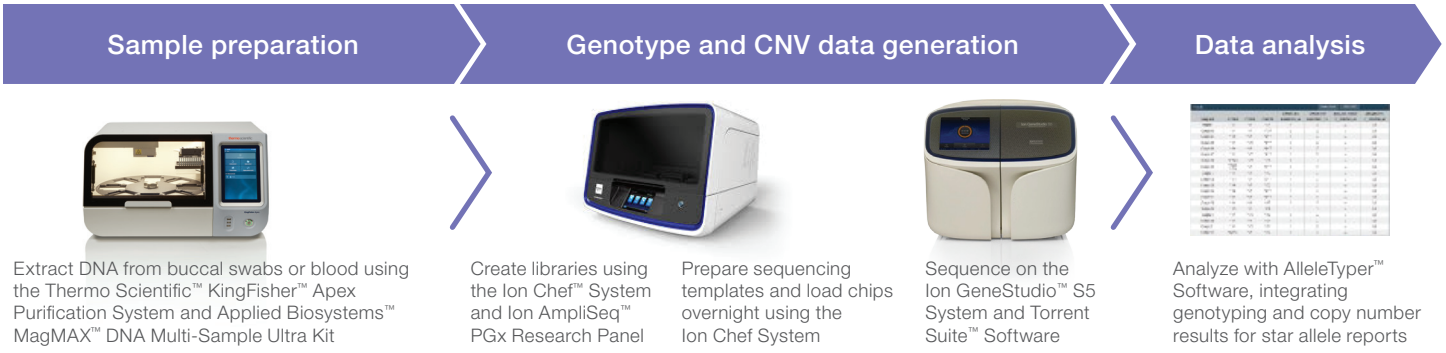


**Figure 2. Complete pharmacogenomics workflow encompassing sample preparation, genotype and CNV data generation, and data analysis.**

# Exceptional sample success

The low per-sample input requirement of only 10 ng, robust compatibility of Ion AmpliSeq chemistry with buccal swab–derived genomic DNA, and broad gene coverage enable lower assay failure rate and sample reruns (Figure 3). This can mean the difference between missing deadlines and delivering research results on time. There are 9 additional panels included in the panel that can generate unique sample-specific genetic barcodes useful for sample identification purposes. Table 1 shows the recommended numbers of samples for Ion 510, 520, and 530 Chips to achieve >500x average coverage per amplicon using estimated throughputs of 3 million, 6 million, and 20 million reads, respectively.

**Table 1. Sample multiplexing recommendations for each type of Ion chip.**

| Chip | Multiplexed samples |
|---|---|
| **Ion 510** | 48 samples |
| **Ion 520** | 96 samples |
| **Ion 530** | 384 samples |

**A**

| Genotyping across 136 genetic variants | |
|---|---|
| Cell line samples | 400 |
| Buccal swab samples | 432 |
| **Genotype accuracy** | **>99.9%** |
| **Genotype call rate** | **>99.9%** |

**B**

| *CYP2D6* gene and exon 9 CNVs | |
|---|---|
| Buccal swab samples | 432 |
| *CYP2D6* gene-level calls | 429 |
| *CYP2D6* gene-level no calls | 3 |
| *CYP2D6* gene-level false calls | 0 |
| ***CYP2D6* gene-level call rate** | **99.3%** |
| *CYP2D6* exon 9–level calls | 415 |
| *CYP2D6* exon 9–level no calls | 17 |
| *CYP2D6* exon 9–level false calls | 0 |
| ***CYP2D6* exon 9–level call rate** | **96.1%** |

**Figure 3. Genotyping and CNV data. (A)** Genotyping results from over 800 cell line and buccal swab samples demonstrate >99.9% accuracy with known genotypes. **(B)** Focusing on the 432 buccal swab samples, *CYP2D6* CNVs were called in 99.3% and 96.1% of samples at the gene and exon 9 levels, respectively, with no false positives.

Primer pools for the Ion AmpliSeq Pharmacogenomics Research Panel can be ordered through the Ion AmpliSeq Designer at ampliseq.com.

## Ordering information

| Product | Quantity | Cat. No. |
|---|---|---|
| **Sample DNA extraction using the KingFisher Apex system** | | |
| MagMAX DNA Multi-Sample Ultra Kit | 500 preps | A25597 |
| | 2,500 preps | A25598 |
| **Sequencing library construction** | | |
| Ion AmpliSeq Library Kit 2.0 | 8 reactions | 4475345 |
| | 96 reactions | 4480441 |
| | 384 reactions | 4480442 |
| Dual Barcode Kit 1-96 | 1 kit | A39360 |
| Ion Library Equalizer Kit | 96 reactions | 4482298 |
| **Templating and sequencing using the Ion Chef and Ion GeneStudio S5 systems** | | |
| Ion 510 & Ion 520 & Ion 530 Kit – Chef | 8 reactions | A34461 |
| Ion 510 Chip Kit | 8 chips | A34292 |
| Ion 520 Chip Kit | 8 chips | A27762 |
| Ion 530 Chip Kit | 8 chips | A27764 |

Learn more at **thermofisher.com/pharmacogenomics**