NGS Disease Research Panels

# ClearSeq Inherited Disease

The answer at your fingertips

## Benefits

### Expert-defined content

- Developed in collaboration with researchers from Medical Genetics, Charite Berlin
- Highly targeted content enabling analysis of regions implicated in rare disease pathogenesis

### Deep coverage

- Optimized design enables trio analysis or deep sequencing of one sample
- Compatible with high throughput or benchtop sequencers

### Fast track to answers

- 3.5x faster sample to sequencing with SureSelect$^{QXT}$
- Accelerated sample to categorized variants with SureCall

## Overview

There are roughly over 7,000 known rare inherited disorders. Though the frequency of these disorders in the population is extremely low, collectively they affect about 8% of the general population. To date, despite research efforts to better understand the genetic basis of these diseases, only about 4,000 of these disorders have been linked to a genetic defect. The challenge in this effort is the syndromic nature of these diseases and highly variable penetrance and expressivity of these disorders which confound conventional disease classification and candidate gene identification. As a result, out of the 20,000 or so genes in the human genome, only about 2,700 genes have been directly implicated in the pathogenesis of inherited diseases.

The ClearSeq Inherited Disease, developed in collaboration with researchers from Medical Genetics, Charite Berlin, is a highly targeted panel that enables analysis of these genes by providing deep coverage or trio analysis even on a benchtop sequencer, allowing for the focused yet comprehensive variant profiling of only those regions known to cause rare inherited disorders (Fig 1).

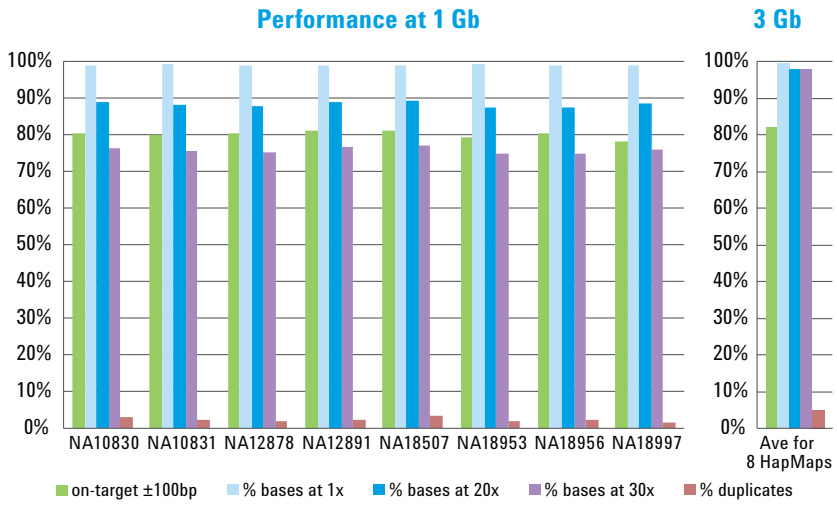


**Figure 1**. The ClearSeq Inherited Disease design provides excellent on-target that ensures deep coverage of regions of interest with only 1 Gb of sequencing, enabling compatibility with benchtop sequencers *(SureSelect$^{XT}$, MiSeq)*

# ClearSeq Inherited Disease

## Highly targeted design for deeper coverage

Current methods used in identifying the underlying genetic basis of rare diseases are challenged by the lack of sufficient sensitivity and resolution. Whole genome sequencing, though offering the single-molecule resolution of NGS has the drawback of having an extremely high number of variants identified from the many coding regions in the genome. The ClearSeq Inherited Disease Panel enables targeted analysis of only those 2,742 regions that have been shown to underlie rare disorders. When compared with other similar targeted solutions, the highly optimized and curated design of this panel coupled with the proven SureSelect technology, provides a single assay that enables deep coverage of more targets compared to similar solutions with equivalent sequencing, greatly facilitating variant calling (Fig 2). Addition of custom content using SureDesign lets you tailor this panel to meet your specific design requirements.

## Faster answers with confidence

Reduced turn-around time to answers is important in clinical research sequencing. Highly targeted analysis using this curated design, paired with efficient workflows that implement greatly reduced hybridization times, as little as 90 minutes for sample to sequencing in a day, enable the quick turn-around time that facilitates greater sample throughput and less overall costs for sample processing. Data analysis, another bottleneck in clinical research, is greatly facilitated by SureCall which provides guided workflows for single sample, paired or trio analysis from raw data to variants enabling expeditious and accurate identification of disease variants (Fig 3).

### Deep Coverage of More Targets

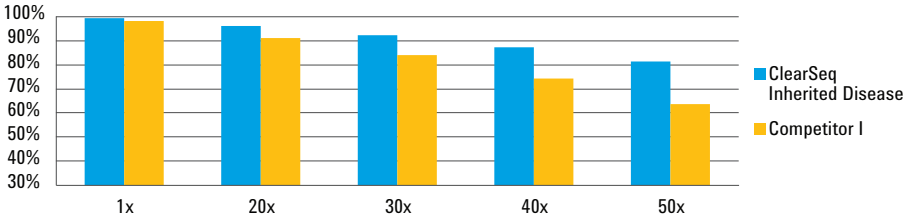


**Figure 2**. The ClearSeq Inherited Disease, designed in collaboration with researchers from Medical Genetics, Charite Berlin, provides a single assay to analyze only those regions known to cause inherited diseases. Coupled with the highly efficient SureSelect workflows, this design enables deep coverage of more targets compared to similar capture solutions within overlapping targeted regions with equivalent sequencing (ClearSeq Inherited Disease, XT: 2 Gb, 2x76 bp; Competitor I: 2.5 Gb, 2x150 bp)

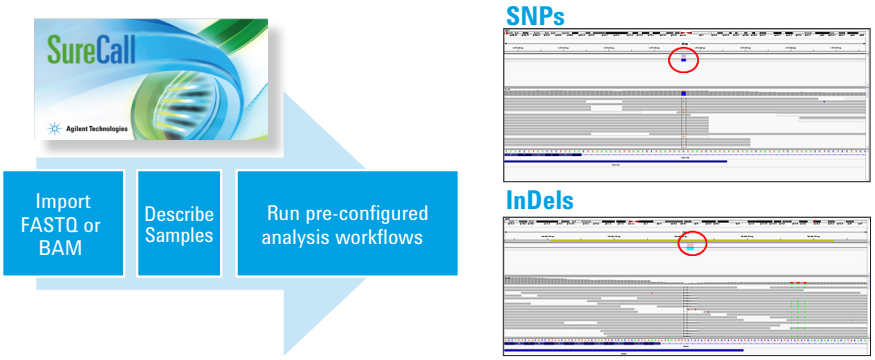


**Figure 3**. Faster time from sample to results are enabled not only by the highly efficient SureSelect workflows but also by SureCall, a software for facilitated raw data to variant calling. SureCall enables the import and analysis of FASTQs or BAMs using guided single-sample, paired or trio analysis workflows that allow for review and categorization of all or just the known variants within the sample.

| Part Number | Description |
|---|---|
| 5190-7518 | ClearSeq Inherited Disease, XT, 16 |
| 5190-7519 | ClearSeq Inherited Disease, XT, 96 |
| 5190-7520 | ClearSeq Inherited Disease, auto, XT, 96 auto |
| 5190-7521 | ClearSeq Inherited Disease Plus, XT, 16 |
| 5190-7522 | ClearSeq Inherited Disease Plus, XT, 96 |
| 5190-7523 | ClearSeq Inherited Disease Plus, auto, XT, 96 auto |
| 5190-7524 | ClearSeq Inherited Disease, XT2, 16 |
| 5190-7525 | ClearSeq Inherited Disease, XT2, 96 |
| 5190-7526 | ClearSeq Inherited Disease, auto, XT2, 96 auto |
| 5190-7527 | ClearSeq Inherited Disease Plus, XT2, 16 |
| 5190-7528 | ClearSeq Inherited Disease Plus, XT2, 96 |
| 5190-7529 | ClearSeq Inherited Disease Plus, auto, XT2, 96 auto |

ClearSeq
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