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2. Shearer AE. Comprehensive genetic testing for hereditary hearing loss using massively parallel sequencing. PNAS 2011
3. Neveau J. Detection of TRPM2 deletion in UMD-1 patients analyzed by a combined CGH array and massively parallel sequencing. Sci Transl Med 2014
4. Koppaiah R. A Facile A2L mutation in glycogen storage disease type II in Brit identified through whole exome sequencing CMAJ 2015

**Complex Disorders:**
2. Subramanian A et al. Exome Sequencing in sporadic cases of schizophrenia identifies 19 putative candidate genes. PLOS One 2014

**Mitochondrial Disorders:**
2. Calvo SE et altt. Late-onset Mitochondrial Disease with Severe NGS, Six Trait Mild 2012
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**Sequencing and Data Analysis**

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**Unrivaled Content Tailored for Clinical Research**

Efforts to provide actionable content for clinical research have led to the introduction of three highly optimized designs that enable compatibility with both high output and desktop sequencers that pave the way to identification of disease-associated variants.

- SureSelect Clinical Research Panel
- OneSeq Constitutional Panel
- ClearSeq Constitutional Panel

These expert-optimized designs enable highly sensitive and accurate variant calling performance in a streamlined workflow with minimal sequencing enabling trio analysis.

**Fastest Workflows, Enriched Libraries in 1 Day**

Complement the best content with the accelerated enrichment workflow of SureSelectQXT, which enables the revolutionary 90-minute hybridization, for faster sample to answers.

**OneSeq Constitutional Panel**

- **Disease Research panels** are catalog designs developed in collaboration with leading medical genetics experts. These constitutional panels provide deep target coverage for confident variant identification.

**ClearSeq Constitutional Panel**

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**Key Features**

- **SureSelect Human All Exon V6**
- **SureSelect Focused Exon**
- **ClearSeq Focused Exon**
- **OneSeq Focused Exon**

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**Best uniformity, best depth of coverage**

- **All Exon V6**
- **SureSelect Human All Exon V6**
- **ClearSeq Constitutional Panel**
- **OneSeq Constitutional Panel**

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**SureSelect**

- **Recommended for**
- **High-output Desktop**

**OneSeq**

- **Recommended for**
- **High-output**

**ClearSeq**

- **Recommended for**
- **High-output**

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**OneSeq Constitutional Panel**

- **Constitutional Research Panel**, **Clinical Research**

- **SureSelect Clinical Research Panel**

- **SureSelect Focused Exon**

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**Hybridization and Capture**

- **Hybridization**

- **Capture**

- **Amplification of Enriched Libraries**

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**Preparation of DNA Libraries**

- **Ordering a Catalog Bait Library or Designing a Custom Panel**

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**Amplification of Enriched Libraries**

- **SureDesign web application**

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**QC Final Library**

- **SureCall data analysis software**

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**Sequencing and Data Analysis**

- **Obtain report of mutations using SureCall data analysis software**

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**For more information:**
www.agilent.com/genomics/NGSConstitutional
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SureSelect is a proven hybridization-based technology that has been instrumental in advancing NGS. With workflows that provide the fastest workflows, enriched libraries in 1 day.

### Key Features
- **SureSelect Constitutional Research**
  - 100% whole exome
  - 20X genome-wide coverage with focused exons
  - 100%–500X genes
  - ~15,000–20,000 genes

- **OneSeq Constitutional Research Panel**
  - 100% whole exome
  - ~5700 genes

- **ClearSeq Inherited Disease Panel**
  - 10.5Mb

These expert-optimized designs enable highly sensitive and accurate variant calling performance in well characterized or highly targeted panels.

### Applications

- **SureSelect Constitutional Research**
  - High-output Desktop
  - High-output (recommended)
  - Or High-output

- **OneSeq Constitutional Research Panel**
  - Desktop exome ONLY

- **ClearSeq Inherited Disease Panel**
  - Only targets genes

HybridPlex is a library prep-free next generation PCR technology that enables a streamlined workflow for target enrichment. This technology leverages the high coverage and specificity of PCR while providing ample redundancy to ensure target coverage. This PCR-compatible technology has been used in the discovery of variants involved in the biological mechanisms of cancer.

ClearSeq Disease Research panels are catalog designs developed in collaboration with leading medical genetics experts. These constitutional panels provide deep target coverage for confident variant identification.

SureSelect is an industry leader for copy number and mutation detection in one streamlined process. Powered by SureSelect, combined genome-wide linkage analysis along with whole-genome sequencing, massively parallel sequencing (WGS) and CNVs, in addition to identification of SNPs and indels.

OneSeq provides a new workflow for constitutional research, in addition to identification of SNPs and Indels, backbones probes along with probes targeting genes enables CNV and LOH detection, in addition to identification of SNPs and indels.

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HaloPlex is a library prep-free next generation PCR technology that enables streamlined workflows for targeted enrichment. This technology leverages the high coverage and specificity of PCR while providing amplification redundancy to ensure target coverage. This PCR compatible technology has been used in the discovery of variants involved in the biological mechanisms of cancer.

ClearSeq is an industry first for copy number and mutation detection in one streamlined assay. Powered by SureSelect, combined genome-wide hybridization times and different pooling strategies, highly sensitive and accurate variant calling performance is achieved with either large exome capture or highly targeted panels.

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Efforts to provide meaningful content for constitutional research have led to the introduction of three highly optimized designs that enable compatibility with both high output and custom strategies that pave the easy to hand-holding of disease-oriented variants. SureSelect Clinical Research Exome, OneSeq Constitutional Research Panel, SureSelect Focused Exome.

These expert-optimized designs combine highly sensitive and accurate variant calling technology providing deep target coverage with minimal sequencing enabling trio analysis.

SureSelect Human All Exon V6

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