

## GENOMIC DNA SEQUENCING & CLINICAL DIAGNOSTIC TESTING

Retrogen has been a trusted provider of quality genomic services since 1992. Founded by scientists experienced in high throughput DNA sequencing, human genomics, and genetic analysis research, we continue to use the most advanced technologies to offer a suite of solutions that accelerate your gene function studies and genetic disease analyses.

Our mission is to become the partner of choice for researchers, physicians, and genetic counselors. We promise to quickly and securely deliver the highest quality results. We help scientists accomplish research projects faster, without additional expense. We help medical professionals accurately diagnose and the improve quality of life for people with inherited genetic diseases.

### BENEFITS

- High sequence quality – using MiSeq, Ion Torrent PGM, and ABI 3730xl
- Fast turn-around times – less than 24 hours, with free local pick up
- Affordability – from ready-to-run to FDA submission sequencing
- All work done at Retrogen is performed under GLP, CLIA, and CAP laboratories

### GENOMIC SERVICES

At Retrogen, we've equipped our sequencing facility with state-of-the-art DNA analysis instruments and advanced bioinformatics capabilities to provide you with consistent and accurate sequence data.

**Illumina MiSeq Personal Sequencer** – Capable of producing up to 7.5 Gb output, MiSeq offers paired-end sequencing, and maximizes efficiency with up to 96-sample indexing.

**Life Technologies Ion PGM** – Using semiconductor chip technology, Ion PGM is ideal for sequencing genes, small genomes, panels of genes, and gene expression profiling.

**Applied Biosystems 3730xl DNA Analyzer** - With accurate base calling, it is well suited for *de novo* sequencing, resequencing, fragment analysis, and SNP genotyping.

#### Services Offered

- DNA sequencing
- Next-generation sequencing
- Epigenetics
- Fragment analysis
- 16S rRNA sequencing
- Mutagenesis
- Gene synthesis
- SNP discovery

### CLINICAL DIAGNOSTICS

As a CLIA and CAP certified laboratory, Retrogen offers diagnostic testing for genetic diseases, from single variant detection to whole genome sequencing. Tests include complete gene analysis by Sanger DNA sequencing, the gold standard for genetic testing. All tests must be ordered through a health care provider such as a physician or genetic counselor.

#### Available Tests By Disease

- Bronchiectasis with or without elevated sweat chloride 1, modifier of - OMIM 211400
- Congenital bilateral absence of vas deferens - OMIM 277180
- Cystic fibrosis - OMIM 219700
- Juvenile Polyposis Syndrome (JPS) OMIM 174900
- Hereditary hemorrhagic telangiectasia syndrome OMIM 175050
- Pancreatitis, idiopathic - OMIM 167800

#### Tests

- BMPR1A – Whole Gene Sequencing
- CFTR – deltaF508
- CFTR – 33 Variant Sequencing
- CFTR – 95 Variant Sequencing
- CFTR – Whole Gene Sequencing
- SMAD4 – Whole Gene Sequencing