CLARITAS GENOMICS

Claritas Clinical Exome

More Than Just An Exome

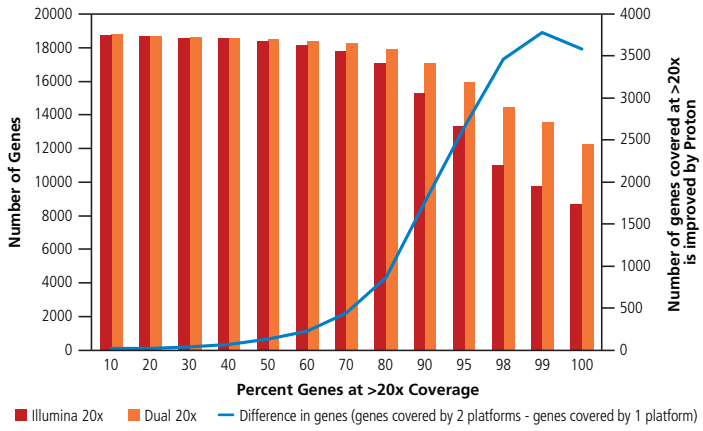
Key Features

1 Innovative Design

Two independent and complementary target capture and sequencing technologies provide:

- Increased sensitivity in detecting variants, as using two unique platforms increases the possibility of detecting variants that a single platform may miss

Improvement in Gene Coverage at >20x Using Two Platforms



- Confirmation of 95% of variants on both platforms with PPV>99.998%. The remaining variants that are potentially pathogenic are confirmed by Sanger sequencing prior to reporting

2 Increase Variant Detection

- Add the companion Deletion/Duplication analysis, a custom-designed array that uses a 400k Agilent aCGH platform to evaluate >4000 genes at high resolution
- Add analysis of the complete mitochondrial genome, sequenced to detect mutations and heteroplasmy.

3 Access to Exome Data

- Interactive variant browsing is available via the WuXi NextCODE™ platform
- Access exome data via a secure download of VCF and BAM files

Turnaround Time

- 12-14 weeks

Test Characteristics

- Orthogonal sequencing is carried out using Illumina NextSeq™ and ThermoFisher Ion Proton™ systems to generate a variant list
- **Orthogonally-confirmed variants demonstrate high specificity (PPV~99.998%)**
- >100x mean coverage on both platforms; >98% at 20x
- Sanger sequencing follow-up for unconfirmed variants
- Protein coding sequences and 10 bp of adjacent intronic sequences are analyzed
- This assay will detect SNVs, insertions, and deletions less than 10 bp.
- Trios are encouraged but not required
- Companion deletion/duplication analysis available for an additional charge
- Analysis of mitochondrial DNA available at no additional charge
- More information on methodology is available at Chennagiri et al., *Orthogonal NGS for High Throughput Clinical Diagnostics*, Sci Rep 6, 24650 (2016)

Additional Service Highlights

- Providers may request a reanalysis of a patient's case. The first reanalysis is performed at no charge when the request is made within 2 years of the date that the report was issued
- Providers may request a review of variant classification at any time
- Providers may access the Interpretive Genomics Service at Boston Children's Hospital, which provides consultation with experts in genes and/or phenotypes.
- Patients and families may opt-out of receiving secondary findings

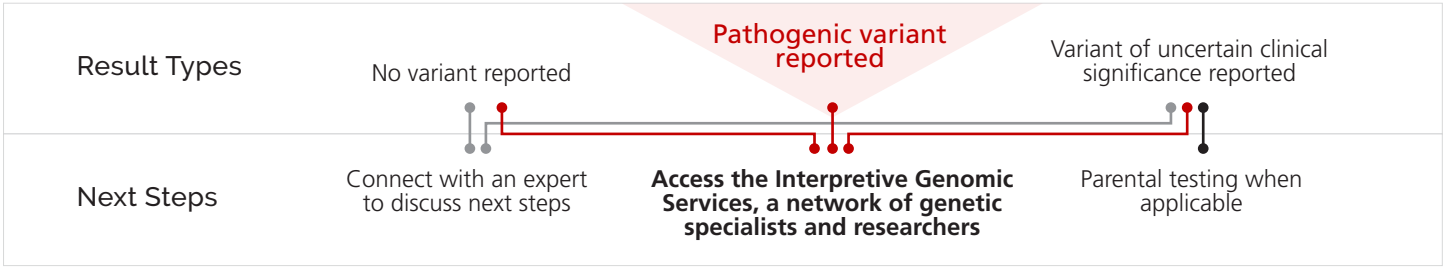
LEARN MORE AT CLARITASGENOMICS.COM

Innovative Approach to Results

Phenotypically-driven trio analysis increases confidence in results	Orthogonal technologies enable the most accurate and comprehensive results	Independent platforms provide superior sensitivity and specificity	Immediate confirmation of variants reduces turnaround time	Variant interpretation performed by credentialed Genetic Counselors and Medical Directors

Support Services

Our Genetic Counselors and Medical Directors are ready to answer your questions about genetic testing



Providers may request raw data in the form of BAMs and VCFs, or via access in the NextCODE interactive platform

How to Order

1 Download and complete the requisition form
www.claritasgenomics.com

2 Obtain the patient's sample

Whole blood
- CCE only: 0.5-3cc
- CCE with Del/Dup Analysis: 2-5cc
- EDTA (preferred), NaCitrate, or ACD

Extracted DNA
- 10µg at 50ng/µL

Saliva
- Contact Client Services for sample requirements

3 Prepare the sample for shipment
- Complimentary kits are available for samples shipped within the United States and include a prepaid airbill
- Order kits at www.claritasgenomics.com

4 Ship overnight for receipt Monday through Friday

Claritas Genomics
Attn: Specimen Processing
99 Erie Street
Cambridge, MA 02139

Connect to The Claritas Network

Access the Interpretive Genomics Services at Boston Children's Hospital, which provides consultation with experts in genes and/or phenotypes

Take advantage of partnerships and save on send-out costs

Questions or Comments? We Want to Hear From You.

Email us at clientservices@claritasgenomics.com
Call us at 617.553.5880
Toll-Free at 855.373.9003
Fax at 617.553.5842

For more information about our tests, products and services, **visit our website** at www.claritasgenomics.com

CLARITAS GENOMICS

LEARN MORE AT CLARITASGENOMICS.COM