CLARITAS GENOMICS

Claritas Clinical Exome

Fast. Confirmed. Focused.

Confirmed, clinically-relevant results within 4 weeks

Highlights

The first-line test for a patient with complex syndromic features

The next step for a patient with complex syndromic features, when no genetic test has uncovered a molecular explanation for the patient's medical issues

Ideal for patients who have conditions with genetic and phenotypic heterogeneity where a single gene test may not fully assay all causes

Uses an innovative dual-capture, dual-platform method that immediately confirms approximately 90% of variants, facilitating a rapid return of results

97% of the target region is covered at >20X by Illumina NextSeq™ and LIFE Ion Proton™ systems

Interactive variant browsing via the WuXi-NextCODE™ platform

Rapid Report

- Orthogonally-confirmed
- Clinically-relevant variants
- Phenotype-related
- Based on proprietary variant assessment and classification methodology

Complete Report

- Adds Sanger-confirmed variants
- Select gap filling
- Analysis of variants in regions of low coverage

Test Characteristics

Typical mean coverage is greater than 100X for both the Illumina NextSeq and LIFE Ion Proton systems ensuring that even DNA that is difficult to sequence can be analyzed

99.0% sensitivity and 99.5% positive predictive value

Platform	Sensitivity	PPV
Illumina NextSeq	97.6%	99.6%
LIFE Ion Proton	94.7%	99.6%
Combined	99.0%	99.5%

Protein coding sequences and 10 bp of adjacent intronic sequences are analyzed

The assay will detect SNVs, insertions and deletions less than 5 bp. Copy number variants and larger indels may not be detected

A companion deletion/duplication analysis adds an additional level of evaluation that increases the detection ability for clinically relevant findings

Trios are encouraged

Patient and families may opt–out of receiving secondary findings

Turnaround Time

- Rapid Report: 4 weeks
- Complete Report: Additional 2-8 weeks depending on patient phenotype

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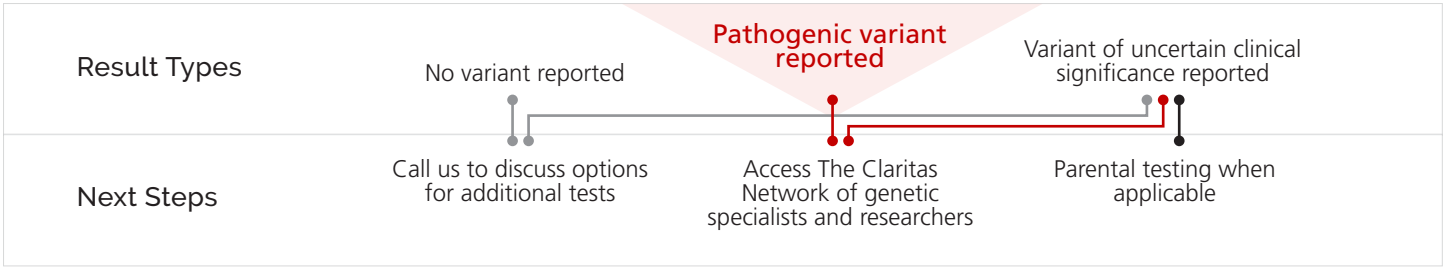
Innovative Approach to Results

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



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

How to Order

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

2 Obtain the patient's sample

Whole blood
- Children or adults: 2-5cc
- Infants under 2 years: 0.5-2cc
- EDTA (preferred), NaCitrate, or ACD

Extracted DNA
- 10µg at 50ng/µL

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 our network of researchers and specialists

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

Contact your Claritas Field Representative for more information and logistical support

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