CLARITAS GENOMICS

Bone Marrow Failure Region of Interest

Minimize serial panel analysis
and provide prompt treatment plans

Key Features

1 Expert Design
Evaluates a comprehensive gene list of 86 genes, designed in collaboration with experts in Hematology

2 Two-Part Approach to Reporting
Rapid reporting of orthogonally-confirmed variants, followed by complete reporting of additional relevant variants

3 Rapid Expansion to a Clinical Exome
If no pathogenic variant is uncovered, providers may order expansion into the Claritas Clinical Exome

4 Access to Exome Data
Interactive variant browsing via the WuXi NextCODE™ platform and access to exome data via a secure download of VCF and BAM files

Indications For Testing

- Pediatric patients with bone marrow failure, as inherited forms may account for 25-40% of children who present with symptoms
- Dyskeratosis congenita, Diamond Blackfan anemia, Fanconi anemia, Shwachman Diamond syndrome, amegakaryocytic thrombocytopenia, single lineage thrombocytopenias, hypoplastic aneimas and neutropenias
- Patients undergoing hematopoietic stem cell transplantation, as rapid TAT is crucial
- Patients with MDS and AML, as this assay also examines recurring variants in these conditions in order to provide assessments for patients at risk of clonal evolution and disease progression

Turnaround Time

- Rapid Report: 4 weeks
- Complete Report: Additional 7 weeks

Rapid Report

- Orthogonally-confirmed, phenotype-related variants
- Reported rapidly to impact patient management

Complete Report

- Adds Sanger-confirmed variants and select gap-filling
- Includes companion deletion/duplication analysis when ordered

Test Characteristics

- Assesses 86 genes associated with inherited Bone Marrow Failure. Gene list is updated based on input from Claritas scientists and partners
- Orthogonal sequencing is carried out using Illumina NextSeq™ and ThermoFisher Ion Proton™ systems to generate a variant list
- **Orthogonal-confirmed variants demonstrate high specificity (PPV~99.998%)**
- >100x mean coverage on both platforms; >98% at 20x
- Sanger sequencing follow-up for unconfirmed variants
- Trios are encouraged, but not required
- Companion deletion/duplication analysis available for an additional charge
- More information on methodology is available at Chennagiri et al., *Orthogonal NGS for High Throughput Clinical Diagnostics*, Sci Rep 6, 24650 (2016)

LEARN MORE AT CLARITASGENOMICS.COM

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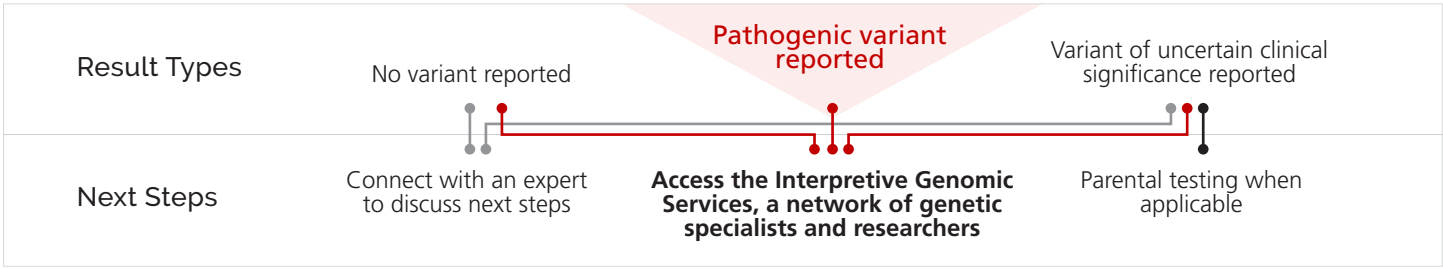
Innovative Approach to Results

Saves time and money by minimizing sequential testing	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
- ROI only: 0.5-3cc
- ROI with Del/Dup Analysis: 2-5cc
- EDTA (preferred), NaCitrate, or ACD

Extracted DNA
- 10µg at 50ng/µL

Saliva
- Contact Client Services for sample requirements

Note: Saliva samples are required for patients who have had a bone marrow transplantation, patients undergoing chemotherapy, or in individuals with leukopenia. Previous BMT may affect the accuracy of results and must be disclosed on the requisition form. Contact us for a complimentary saliva collection kit.

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

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