Clairiti Diagnostics Laboratories LLC
20 High Street, Suite-207
Hamilton OH 45011
Phone: (513) 973 0062
Fax: (513) 973 0066
Email: info@claritidiagnostics.com
www.claritidiagnostics.com

Lab Director: Neil B Quigley, PhD, HCLD(ABB)
CLIA License ID: 36D2162956

Germline Variant Report

Patient

First name: John
Last name: Doe
Patient ID: library01
DOB: 1 Jan 1970
Gender: Male
Father ethnicity: UNKNOWN
Mother ethnicity: UNKNOWN

Specimen

Accession Number: 20190801003
Specimen Type: Saliva
Collection Date: 08/01/2019
Accession Date: 08/03/2019
Completion Date: 08/03/2019

INTERPRETATION

Positive

This patient is heterozygous for a Pathogenic variant in *BRCA2* gene which has been associated with increased risk of cancer. Consultation with an Oncologist is recommended

RESULTS

SNVs/INDELs (selected in report)

Gene Transcript	Exon	c.DNA Protein alteration	Variant Fraction Coverage (ref / alt)	Coding consequence	Pathogenicity	ClinVar
BRCA2 NM_000059.3	4	c.4631delA	32% (68 / 32)	p.N1544fs*24	Flagged Pathogenicity 5 | Definitely Pathogenic	Pathogenic rs80359460
Comment: The *BRCA2* is a tumor suppressor protein involved in DNA repair mechanism. The absence of this protein results in increased DNA damage and risk for cancer. The patient has a single adenine nucleotide deleted at position 4631 in *BRCA2* gene. This mutation causes a frame-shift resulting in synthesis of non-functional protein.						
ATM NM_000051.3	6	c.4709T>C	51.46 % (50 / 53)	p.V1570A	3 | Variant of Unknown Significance	VUS rs9534262

Reviewed by:
Date:
Signature:

Approved by: Operator User
Date: 16-03-2018 | 17:34:03
Signature:

Clairiti Diagnostics Laboratories LLC
20 High Street, Suite-207
Hamilton OH 45011
Phone: (513) 973 0062
Fax: (513) 973 0066
Email: info@claritidiagnostics.com
www.claritidiagnostics.com

Lab Director: Neil B Quigley, PhD, HCLD(ABB)
CLIA License ID: 36D2162956

METHODOLOGY AND DISCLAMER

Clariti NGS Methodology

Clariti Diagnostic Laboratories LLC uses Next Generation Sequencing (NGS) technology to analyze coding regions and exon / intron boundaries of selected genes. Nextera Flex For Enrichment chemistry from Illumina is utilized to prepare genomic DNA for sequencing. Genomic DNA is fragmented using Transposomes. Each patient sample is labelled with a unique DNA index to allow libraries to be developed and sequenced simultaneously. Hybrid capture chemistry is utilized to target and then amplify specific regions of interest. The target regions are sequenced with the Illumina Miniseq instrument using either, a mid-output or high-output kit. At least 99.0 % of target regions are covered at 20X or higher.

FASTQ files are analyzed by Sophia Genetics for secondary and tertiary analysis. DNA sequences are assembled and aligned against human genome build GRCh37. The genomic variants are identified and evaluated for clinical relevance using ACMG guidelines. Sanger sequencing is used to confirm all pathogenic variants.

This test was developed and tested by Clariti Diagnostics Laboratories, LLC. Performance parameters were characterized by running extensive set of control samples, patient samples and samples from normal volunteers. The test is not currently FDA cleared. The FDA has determined such clearance for this test is not necessary for laboratories with good standing with CLIA (Clinical Laboratory Improvement Amendments of 1988).

Bioinformatics analysis is provided by Sophia Genetics, Inc (Boston, MA).

Test Limitations and Clariti Disclaimer

Clariti Diagnostic Laboratories LLC uses Next Generation Sequencing (NGS) technology to analyze coding regions and exon / intron boundaries of selected genes. Nextera Flex For Enrichment chemistry from Illumina is utilized to prepare genomic DNA for sequencing. Genomic DNA is fragmented using Transposomes. Each patient sample is labelled with a unique DNA index to allow libraries to be developed and sequenced simultaneously. Hybrid capture chemistry is utilized to target and then amplify specific regions of interest. The target regions are sequenced with the Illumina Miniseq instrument using either, a mid-output or high-output kit. At least 99.0 % of target regions are covered at 20X or higher.

FASTQ files are analyzed by Sophia Genetics for secondary and tertiary analysis. DNA sequences are assembled and aligned against human genome build GRCh37. The genomic variants are identified and evaluated for clinical relevance using ACMG guidelines. Sanger sequencing is used to confirm all pathogenic variants.

This test was developed and tested by Clariti Diagnostics Laboratories, LLC. Performance parameters were characterized by running extensive set of control samples, patient samples and samples from normal volunteers. The test is not currently FDA cleared. The FDA has determined such clearance for this test is not necessary for laboratories with good standing with CLIA (Clinical Laboratory Improvement Amendments of 1988).

TEST PERFORMED

Gene Panel (2)

The patient gave his consent to analyse the following genes:
BRCA1 , *BRCA2*