Kailos™ Cancer Screening Test Results

Dr. Ronald McGlennen, Medical Director
CLIA#: 01D2016114

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PATIENT: Doe, John (M)
DOB: 1985-01-05
PATIENT ID:

COLLECTED: 10/04/2016
RECEIVED: 03/08/2017
REPORTED: 03/08/2017

SAMPLE TYPE: Buccal
PHYSICIAN: Dr. Example
PRACTICE: Example Practice

ACCESSION: PX941521

RESULTS

Everyone carries DNA changes, known as variants. Following sequencing of the patient's DNA, these variants were analyzed for their effect on their risk of developing cancer. Those that are known to increase risk are reported in the section below.

CLINICAL SIGNIFICANCE	GENE	VARIANT	ASSOCIATED WITH
Pathogenic	**BRCA2**	**NC_000013.10:g.32912339_32912340delGT; NM_000059.3:c.3847_3848delGT**	**Hereditary cancer-predisposing syndrome**

ANALYSIS AND INTERPRETATION

The data indicates that the patient has a gene change that increases their risk for one or more cancers. This does NOT mean that they have cancer. Knowing about an increased risk for cancer can allow you to recommend changes to their screening (i.e. mammograms, colonoscopies) and medical management plan to reduce the chance of cancer developing, or catch a cancer early at a more treatable stage.

Patient consultation with a genetic counselor who has training and experience in cancer genetics is strongly recommended. Topics of discussion should include: (1) cancer risks and other disease risks, (2) type and frequency of cancer surveillance, (3) cancer prevention options and strategies, and (4) the impact of this result on the potential cancer risks for family members. Prior to this consultation, you and your patient can view an informational video on what it means to have variants that increase cancer risks.

In addition, information about cancer surveillance and prevention options that can be recommended to an individual with a genetic risk for cancer can be found on the following websites:

National Society of Genetic Counselors: www.nsgc.org
National Cancer Institute: http://prevention.cancer.gov

METHODOLOGY AND LIMITATIONS

The Cancer Screening Panel uses state-of-the-art sequencing technology to provide high quality results. Genomic DNA is extracted from dry buccal swabs using magnetic particle processing. DNA from patient samples are amplified with primers specific for the targeted regions using Oligo Directed Patch PCR (Varley, et. al.). Positive and negative controls are used with each run. Barcoded patient samples and positive controls are paired-end sequenced using Illumina NextGen sequencing technology. Targeted sequencing is performed on the entire coding region and intronic/exonic boundaries unless otherwise noted below. Sequences are aligned to the human reference genome and variants (Small Nucleotide Variations, Insertions and Deletions) are called. Large indels (>50 bp), rearrangements, rare abnormalities and structural variations may not be detected. Rare diagnostic errors may occur if variations occur in primer site locations.

The regions sequenced include many, but not all, genes that have been shown to affect our risk of developing cancer and/or impact medical management. The following genes are sequenced: APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, EPCAM, FH, FLCN, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, RAD51D, RINT1, SDHB, SMAD4, STK11, TP53, VHL, and XRCC2. Exon 30 of ATM, exon 1 of EPCAM, exons 11 and 14 of FCLN, exon 1 of PALB and exon 1 of RINT1, as well as some highly repetitive or low complexity regions, have also been excluded from analysis. Over 97% of the coding regions of these genes are covered by this panel. For a detailed list of gene transcripts and the reportable range, please visit www.kailosgenetics.com/panel/csp-2.3.1.

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Extensive computational analysis is performed to validate the variants and reduce the likelihood of error. Qualified variants are compared to the NIH ClinVar database. Variants are reported as being Pathogenic or Likely Pathogenic if there is (a) sufficient support in ClinVar or (b) Kailos determines that a mutation would likely cause a deleterious frameshift or premature stop. Kailos utilizes up-to-date information on DNA variants that increase cancer risk found at the National Institutes of Health. This information increases continually and this report reflects the current state of knowledge at the time of reporting. The pathogenic classification of variants may change as new scientific information is learned. All data is reviewed for release by our Medical Director and/or our CLIA Lab Manager.

All pathogenic or likely pathogenic variants within the targeted regions are reported. We do not report benign and likely benign variants, as these variants most likely do not cause an increased cancer risk. Variants of uncertain significance (VUS) are also not reported as they would not be used to change medical management.

LEGAL

The information presented on this report is provided as supplementary health information. The results presented are intended for use by a physician or other healthcare professional to advise a patient on their health care. This test is not a 510k cleared test, but managed by CMS and FDA under the Clinical Laboratory Improvement Amendment (CLIA) as a LDT. The ordering physician is responsible for the diagnosis and management of disease and decisions based on the data provided. Results are dependent on adequate specimen collection and processing.

Genetic testing was performed in the Kailos Genetics CLIA facility at 601 Genome Way; Huntsville, Al. 35806. CLIA#: 01D2016114. Medical Director: Ronald McGlennen MD, FCAP, FACMG, ABMG. This report was reviewed and approved for release by CLIA Lab Manager & Supervisor: Michele R. Erickson-Johnson, PhD, MB(ASCP)^{CM}.