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CLIA# 11D2065318 | LABSOLUTIONS.COM

Report Date
Oct 6, 2017

PREDICT™ *Hereditary Cancer Risk Assessment*

CLIENT INFORMATION		PATIENT INFORMATION		SPECIMEN INFORMATION	
Client	LAB SOLUTIONS	Patient Name	TEST-34-POS	Accession ID	TEST-34-POS
		Date of Birth	Mar 12, 1919	Specimen Type	Saliva
Physician	DR. B	Sex	Female	Collection Date	Oct 5, 2017
Client ID	2	Ethnicity		Analysis Date	Oct 6, 2017

RESULT: POSITIVE - PATHOGENIC MUTATION IDENTIFIED

- **Positive - Pathogenic Mutation Identified.**

  Clinically Significant as defined in this report is a genetic change that has the potential to alter medical intervention.

- **Variant of Uncertain Significance Identified. See Additional Findings.**

GENE	PATHOGENIC MUTATION	INTERPRETATION
RAD50	c.2903G>A (p.G968E)	Increased risk for hereditary cancer-predisposing syndrome
FANCC	c.355_360delTCTCATinsA (p.S119fs*8)	Increased risk for cancer

GENES TESTED

APC ATM BARD1 BMPR1A BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EPCAM FANCC MEN1 MET MLH1 MRE11 MSH2 MSH6 NBN NF1 NTRK1 PALB2 PMS2 PTCH1 PTEN RAD50 RAD51C RAD51D RET SMAD4 STK11 TP53 VHL

QIAGEN Clinical Insight - Interpret software was used in sequence interpretation.

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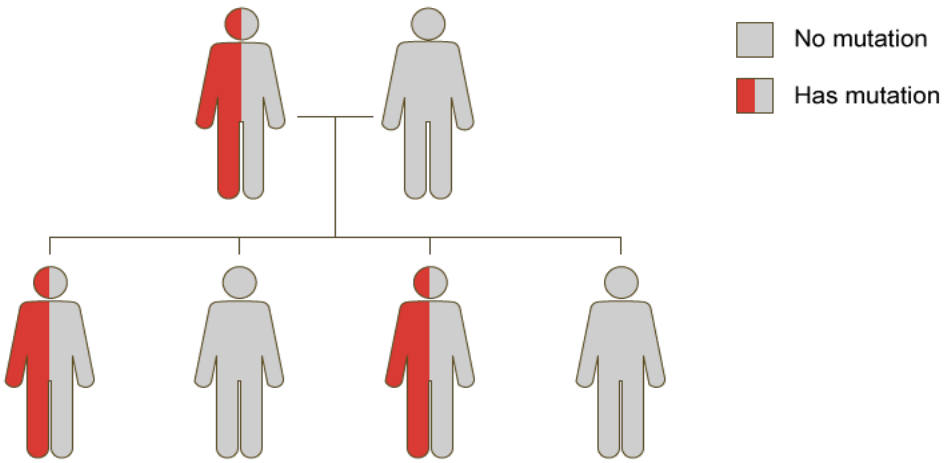
MUTATIONS IN THE FAMILY

Family members may also be at risk. There is a 50/50 random chance of passing on a mutation to your sons and daughters. They can be tested for the mutation that was found in you.



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CLINICALLY RELEVANT RESULTS: PATHOGENIC

RAD50 NM_005732.3 c.2903G>A (p.G968E) Likely Pathogenic Heterozygous Exon 18	**Interpretation:** Patients with a mutation in the RAD50 gene have an increased risk to develop female breast cancer, ovarian cancer, and possibly other types of cancer. This result should NOT be interpreted to mean that this patient HAS cancer, or WILL GET cancer of any of the types indicated above or any other cancer. This result simply says that this patient is at increased risk (when compared to the general population) for these conditions and should follow up with appropriate medical professionals as indicated. Genetic counseling is suggested.
FANCC NM_000136.2 c.355_360delTCTCATinsA (p.S119fs*8) Pathogenic Heterozygous Exon 5	**Interpretation:** Patients with a mutation in the FANCC gene have an increased risk to develop female breast cancer, and possibly other cancers such as pancreatic cancer. This result should NOT be interpreted to mean that this patient HAS cancer, or WILL GET cancer of any of the types indicated above or any other cancer. This result simply says that this patient is at increased risk (when compared to the general population) for these conditions and should follow up with appropriate medical professionals as indicated. Genetic counseling is suggested.

ADDITIONAL FINDINGS: VARIANTS OF UNCERTAIN SIGNIFICANCE

RAD51D NM_002878.3 c.695G>A (p.R232Q) Uncertain Significance Heterozygous Exon 8	**Interpretation:** This patient has been identified to carry a variant of unknown significance (VUS). Available science has not yet confirmed that the variant is harmful to this or any patient with enough confidence at present. The patient should be made aware, but explain that there is not enough conclusive information currently. No action needs to be taken by the physician or patient at this time.
CDK4 NM_000075.3 c.696G>A (p.L232L) Uncertain Significance Heterozygous Exon 7	**Interpretation:** This patient has been identified to carry a variant of unknown significance (VUS). Available science has not yet confirmed that the variant is harmful to this or any patient with enough confidence at present. The patient should be made aware, but explain that there is not enough conclusive information currently. No action needs to be taken by the physician or patient at this time.

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GENES TESTED

APC ATM BARD1 BMPR1A BRCA1 BRCA2 BRIP1 CDH1 CDK4 CDKN2A CHEK2 EPCAM FANCC MEN1 MET MLH1 MRE11 MSH2 MSH6 NBN NF1 NTRK1 PALB2 PMS2 PTCH1 PTEN RAD50 RAD51C RAD51D RET SMAD4 STK11 TP53 VHL

METHODS AND LIMITATIONS

Testing Methods:

Genomic DNA from the submitted specimen was extracted and enriched for the targeted gene regions using a proprietary targeted capture system, **PREDICT**™, developed by LabSolutions. The products were then sequenced on an Illumina NextSeq with 2x150 paired-end reads. The sequences generated were then aligned to reference sequences based on human genome (hg19) using the Biomedical Workbench workflow. All sequence alterations are described according to nomenclature guidelines set forth by the Human Genome Variation Society (HGVS). The variants are classified according to the ACMG-recommended classification.

QIAGEN Clinical Insight - Interpret software was used in sequence analysis and interpretation. The application was internally designed and developed by QIAGEN. All analyses were based on: QIAGEN Clinical Insight-Interpret (5.0.20171003), Ingenuity Knowledge Base (Narnia 170928.001), CADD (v1.3), CentoMD (-), EVS (ESP6500SI-V2), Allele Frequency Community (2017-07-03), JASPAR (2013-11), Vista Enhancer hg18 (2012-07), Vista Enhancer hg19 (2012-07), gnomAD (2.0.1), Clinical Trials (Narnia 170928.001), BSIFT (2016-02-23), TCGA (2013-09-05), PolyPhen-2 (v2.2.2), 1000 Genome Frequency (phase3v5b), Clinvar (2017-06-01), DGV (2016-05-15), COSMIC (v81), ExAC (0.3.1), HGMD (2017.2), PhyloP hg18 (2009-11), PhyloP hg19 (2009-11), DbSNP (150), TargetScan (6.2), SIFT4G (2016-02-23)

LABORATORY STATEMENT

The purpose of this sequencing report is to provide general health screening information. The content is not intended to be a substitute for professional medical advice, diagnosis, or treatment. LabSolutions is a CLIA-certified and COLA accredited laboratory for performance of high-complexity testing and has developed this test and its performance characteristics. This test has not been cleared or approved by the U.S. Food and Drug Administration, however, FDA approval or clearance is currently not required for clinical use of this laboratory-developed test. The results of this sequencing tests are not intended to be used as the sole means for clinical diagnosis or patient management decisions. This report forms one of the multiple pieces of information that clinicians/health-care providers should consider in guiding their treatment options for each patient. Counseling and informed consent are strongly recommended for genetic testing.

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TAKE HOME POINTS FROM THIS REPORT

RESULT: *RAD50*	Your test shows that you have a mutation in the ***RAD50*** gene. This result should be considered positive.
PERSONAL RISKS	You are at increased risk for female breast cancer, ovarian cancer, and possibly other types of cancer based on this genetic result.
FAMILY RISKS	Your close family members may also be at increased risk based on this genetic information. You may share this information with them, so they may make a decision to be tested for the same mutation as you have.
NEXT STEPS	It is suggested that you share this specific information with your family so they may decide to get tested if they wish.
MANAGEMENT OPTIONS	The decision as to how to act on the information provided in this report is one that is unique to each individual, their family, and their healthcare provider. Choices may include earlier or more increased screening, additional testing, preventative medication, and potentially prophylactic surgery. It is critical to discuss options with a qualified healthcare professional and a genetic counselor prior to choosing any definitive options.

RESULT: *FANCC*	Your test shows that you have a mutation in the ***FANCC*** gene. This result should be considered positive.
PERSONAL RISKS	You are at increased risk for female breast cancer, and possibly other cancers such as pancreatic cancer based on this genetic result.
FAMILY RISKS	Your close family members may also be at increased risk based on this genetic information. You may share this information with them, so they may make a decision to be tested for the same mutation as you have.
NEXT STEPS	It is suggested that you share this specific information with your family so they may decide to get tested if they wish.
MANAGEMENT OPTIONS	The decision as to how to act on the information provided in this report is one that is unique to each individual, their family, and their healthcare provider. Choices may include earlier or more increased screening, additional testing, preventative medication, and potentially prophylactic surgery. It is critical to discuss options with a qualified healthcare professional and a genetic counselor prior to choosing any definitive options.