

# Suretox Genetic Report Overview



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<b>Client:</b> SURETOX 495 Boulevard Suite 1A ELMWOOD PARK, NJ 07407 Phys.: Dr Smith	<b>Patient:</b> TEST, PATIENT1 <b>DOB:</b> Gender: F <b>Chart#</b> <b>Specimen Type:</b> Oral Swab	<b>Lab Acc#:</b> 1606200054 <b>Collected:</b> 06/20/16 12:00 <b>Accessioned:</b> 06/20/16 16:21 <b>Reported:</b> 07/27/16 22:29
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**Discover™ Hereditary Cancer Risk Assessment Report:**  
**TEST ADMINISTERED:**

BreastDiscover Hereditary Cancer Risk Assessment. Purification of genomic DNA, Sample preparation, enrichment for 22 Inherited Cancer Genes, Illumina Next Generation Sequencing, Variant filtration described in Appendix A, Clinical Interpretation.

**CLINICAL INDICATION AND NOTES:**  
 Information provided indicates that this individual has a personal and/or family history of cancer.

**TEST RESULT SUMMARY:**

Gene/Chromosome/Exon	Genomic Location	Zygosity/MOI	Pathogenicity	Associated Disease
BRCA2/13/10	32906980	Homozygous	Highly Pathogenic	[VARIANT] CNV deletion. Associated with hereditary breast cancer
MUTYH/1/11	45797760	Heterozygous	Highly Pathogenic	This variant is a Nonsynonymous SNV mutation in the MUTYH gene

**COMMENTS:**  
 A potentially relevant CNV (BRCA2 one copy deletion) is present.

NM\_001128425.1(MUTYH):c.934-2A>G  
 This variant is a Nonsynonymous SNV mutation in the MUTYH gene. Zygosity state is a Heterozygous. It is a variant with a 0.0391 maximal allele frequency in the population databases available for review. In peer reviewed clinical literature this variant has been reported to be associated with phenotype(s) listed in the Associated disease field in the table above. Calculated severity score is 0.39000 on a scale of 0.0 – 1.0. According to HGMD/ClinVar, the ELAC2 gene is associated with Prostate cancer, association with: Prostatecancer; Cancer; Hypertrophic cardiomyopathy and complex I deficiency; Muscle and heart defects with lactic acidosis, Combined oxidative phosphorylation deficiency 17; Prostate cancer, hereditary, 2. Additional gene description (Uniprot): Zinc phosphodiesterase, which displays mitochondrial tRNA 3'-processing endonuclease activity. Involved in tRNA maturation, by removing a 3'-trailer from precursor tRNA. The gene has not been tested for essentiality and its tolerance score (RVIS) is in the top 10.12%.

- 1 Client information
- 2 Patient information
- 3 Test administered
- 4 Result Summary:  
- Positive:  
Cancerous mutation detected  
  
- Negative:  
No mutation detected  
  
- Unknown  
Clinical significance:  
New mutation detected,  
requires further research
- 5 Description of the Result

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Discover™  
 Next Generation  
 Sequencing



## WHY Get Tested for Hereditary Cancer

- Cancer screening helps to identify hereditary mutations to assess patients' risk for specific cancers.
- The National Cancer Institute (NCI) estimates that approximately 240,000 new cases of breast cancer in females, and 2,350 in males, will be diagnosed in the U.S.
- Breast cancer is the most commonly diagnosed cancer in developed countries, affecting approximately 1 in 8 (12.5%) women in their lifetime.
- Ovarian cancer is the fifth most commonly diagnosed cancer among women in developed countries, affecting approximately 1 in 71 (1.4%) women in their lifetime. It is the leading cause of death from gynecologic malignancy.
- Colorectal cancer (CRC) affects about 1 in 20 (5%) individuals in their lifetime. While a majority of CRC cases are sporadic, approximately 30% of the cases tend to be hereditary such as Lynch Syndrome.
- Uterine cancer affects about 1 in 37 (2.6%) women in their lifetime. The NCI estimated that approximately 54,870 new cases were diagnosed in the U.S. in 2015 and accounted for approximately 10,170 deaths.
- Pancreatic cancer affects about 1 in 65 (1.5%) individuals. Multiple genes are associated with increased pancreatic cancer susceptibility with approximately 5-10% of cases being hereditary.
- Kidney cancer affects about 1 in 60 (1.6%) individuals in the U.S. Renal Cell Carcinoma (RCC) is a complex disease with a diverse spectrum of tumor subtypes. Approximately 3-5% of RCC cases are hereditary.

## Cancer Targets

### Examples of Common Hereditary Cancers

Supplemental Table 1: Known associations between genes in Discover™'s Hereditary Cancer Genetic Test and cancer type. This is only an example. Discover™, tests for 105 genes. For a complete list contact Suretox Laboratory.

GENE \ TYPE OF CANCER	Breast Discover	Ovarian Discover	Uterine Discover	Colorectal Discover	Melanoma Discover	Pancreatic Discover	Gastric Discover	Prostate Discover	Lung Discover	CNS Discover	Kidney Discover	Bladder Discover
AIP							•					
ALK									•	•		
APC						•	•					
ATM	•					•						
BAP1					•							
BARD1	•	•										
BLM					•					•		
BMPR1A				•		•	•					
BRCA1	•	•				•		•				
BRCA2	•	•			•	•		•				
BRIP1	•	•										
BUB1B	•									•		
CASR											•	
CDC73		•				•		•				

How Discover™ Hereditary Cancer test works:

## Next Generation Sequencing Technology

- NGS uses a very low volume of patient genomic DNA, just 20 ng of sample extracted from a buccal swab. This volume is a thousand times less than other applications on the market.
- 80% of the sequencing is done on an Illumina platform, a highly reliable genetic testing technique.
- TruSight Rapid Capture library methodology ensures precise targeted gene capture and region enrichment.
  - Invasive sample collection is not necessary.
- Oral swabs are an effective and reliable source used to obtain genetic material.

## HOW IT WORKS

Your genetic testing will indicate 1 of 3 results: Positive, Negative, and Unknown clinical significance. Make sure to follow up with your physician for the appropriate steps following your results.

### POSITIVE RESULTS

A positive result indicates an increased possibility of acquiring certain types of cancers due to the presence of an inherited gene mutation. Please note that a positive result does not indicate that the patient will definitely acquire cancer. Positive results affect the entire family. There is a chance this mutation could be genetically passed on to the patient's children - a possibility that is increased if the spouse tests positive as well.

### NEGATIVE RESULTS

A negative result indicates that the genetic mutations the test was designed to detect have not been found. It is still important to receive routine health monitoring and screening as this result does not eliminate the risk of developing cancers. It does indicate the patient does not have a genetic risk for the cancers associated with the genes tested.

### UNKNOWN CLINICAL SIGNIFICANCE

An unknown result indicates a mutation has been detected, however that mutation has not yet been linked to an increased risk of specific cancers. Though this information may not be currently available, genetic research is making major strides towards categorizing and classifying these variants. SureGx will always be at the forefront of genetic research.

## NEXT STEP

After results of the Discover test are available, it is recommended that the patient consults with a genetic counselor for further information and interpretation of any positive results.

Please contact Suretox Laboratory for further information.