Rob Currie
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Genetic Data: 1 in 25 Americans
BRCA 1 & 2

Familial Breast Cancer
No Familial Breast Cancer
General Population

Cumulative Risk vs. Age
Color's BRCA Test

$149  $99

What's included in your Color test

- Saliva collection kit & prepaid return label
- BRCA1 and BRCA2 test report
- Expert genetic counseling
- Latest genetics news that matters to you

Buy Color

*You must be at least 18 to use Color. If you have a personal or family history of cancer, you may want to consider Color's Hereditary Cancer Test. Learn more ›
A pathogenic mutation was identified in the BRCA1 gene.

Testing positive for a pathogenic mutation in the BRCA1 gene means your risks of developing breast and ovarian cancer are significantly greater than that of the average US woman. Your risk of pancreatic cancer is also increased by this mutation. This result does not mean that you have a diagnosis of cancer or that you will definitely develop cancer in your lifetime. Your actual risk may be different based on other genetic and non-genetic factors.

**DETAILS**

<table>
<thead>
<tr>
<th>GENE</th>
<th>VARIANT</th>
<th>CLASSIFICATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>BRCA1</td>
<td>c.181T&gt;G (p.Cys61Gly)</td>
<td>Pathogenic</td>
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*Alternate name(s):* C010, chr17:GRCA17.g.112200041A>T
*Transcript: ENST00000357854
*Genotype: Heterozygous

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No mutations were identified.

This means no pathogenic or likely pathogenic genetic variants associated with an increased risk of breast, colorectal, melanoma, ovarian, pancreatic, stomach, or uterine cancers were identified in any of the 30 genes analyzed.

This result does not eliminate your risk of developing cancer. Inherited mutations explain some cases of cancer, but most are not inherited and cannot be explained by a single cause. Some non-genetic factors that can influence cancer risk include environment and lifestyle, as well as family history without a known genetic link. Your healthcare provider can help determine how your screening plan might be influenced by your health history and other factors.

**DETAILS**

A “Variant of Uncertain Significance” (VUS) was identified. This is a genetic change whose impact on hereditary cancer risk is not yet known. This is a common finding and does not change screening guidelines. To date, most VUS’s have been found to be harmless (benign).

If it is further classified, we will try to contact you at jane@gmail.com and (415) 555-1212.

<table>
<thead>
<tr>
<th>GENE</th>
<th>VARIANT</th>
<th>CLASSIFICATION</th>
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<tbody>
<tr>
<td>MSH6</td>
<td>c.3947G&gt;A (p.Gly1316Glu)</td>
<td>Variant of Uncertain Significance</td>
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*Alternate name(s):* C023, chr17:GRCA17.g.80337365G>A
*Genotype: Heterozygous
From 2 to All Genes

BRCA1
BRCA2

5% VUS

50%+ VUS

ATM
BARD1
BRIP1
CDH1
CHEK2
MLH1
MSH2
MSH6
PMS2
EPCAM
NBN
NF1
PALB2
PTEN
RAD51C
RAD51D
STK11
TP53
Most Biomedical Data Held in Silos
We need a network for sharing
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Patient Name:  
Patient, Test:  
Report Date: 09 December 2016  
Tumor Type: Breast carcinoma (NOS)

Date of Birth: 12 December 1942  
Sex: Female  
FMI Case #: SMP60884  
Medical Record #: 12345678  
Specimen ID: Not Given  
Medical Facility: ABC Oncology  
Ordering Physician: Smith, John  
Specimen Site: Breast  
Date of Collection: 08 December 2016  
Pathologist: Public, John Q  
Specimen Type: Slide  
Specimen Received: 01 December 2016

ABOUT THE TEST:  
FoundationOne™ is a next-generation sequencing (NGS) based assay that identifies genomic alterations within hundreds of cancer-related genes.

PATIENT RESULTS
4 genomic findings
2 therapies associated with potential clinical benefit
0 therapies associated with lack of response
10 clinical trials

TUMOR TYPE: BREAST CARCINOMA (NOS)
Genomic Alterations Identified:
- PIK3CA E545K
- ATM T2333fs*40
- BCL2L1 amplification – equivocal*
- MYST3 amplification – equivocal*

Additional Disease-relevant Genes with No Reportable Alterations Identified:
- ERBB2

THERAPEUTIC IMPLICATIONS

<table>
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<tr>
<th>Genomic Findings Detected</th>
<th>FDA-Approved Therapies (in patient’s tumor type)</th>
<th>FDA-Approved Therapies (in another tumor type)</th>
<th>Potential Clinical Trials</th>
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<tr>
<td>PIK3CA E545K</td>
<td>Everolimus</td>
<td>Temsirolimus</td>
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<tr>
<td>ATM T2333fs*40</td>
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<td>None</td>
<td>Yes, see clinical trials section</td>
</tr>
<tr>
<td>BCL2L1</td>
<td>None</td>
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<tr>
<td>MYST3 amplification</td>
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* For a complete list of the genes assayed and performance specifications, please refer to the Appendix
* See Appendix for details

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Comorbid/Complication 8: NONE

Comorbid/Complication 9: NONE

Diagnosis Days from First Contact: 17

Earliest 1st Course RX Days from First Contact: 38

Grade/Differentiation: NOT DETERMINED OR NA(9)

Hist/Behav: CHOLANGIOCARCINOMA(81603)

Histology/Behavior (ICD-O-3): CHOLANGIOCARCINOMA(81603)

Hormone Started Days from First Contact:

Hormone, At UCSF: NONE, NOT PLANNED

Immunotherapy Days Started from First Contact:

Immunotherapy, At UCSF: NONE, NOT PLANNED

Last 1st Course RX Days from First Contact: 86

Last Contact or Death Days from First Contact: 186

Last Tumor Status Days from First Contact: 186

No Treatment Decision Days from First Contact:
Users
Researchers, Clinicians, Pharma, Data Analysts, Citizen Scientists

Cancer Gene Trust

Public Data

Steward Lab
Patient Participants
Private Data

Steward UCSF
Patient Participants
Private Data

Steward BRCA
Patient Participants
Private Data
Moving Compute to the Data
Unlocking life’s code.