

### **Report Contents**

- 1. Coriell Personalized Medicine Collaborative Research Study Report. This report includes all data included in the clinical report as well as supplemental interpretations and educational material. This research report is based on Questionnaires Finalized on 08/01/2010**
- 2. Clinical Report. This report was generated and approved by Coriell's CLIA certified genotyping laboratory.**



## Sample Results

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### CPMC Research Study Report

<b>Name:</b>	NATALIE DEMO	<b>Gender:</b>	Female
<b>Date of Birth:</b>		<b>Date Collected:</b>	11-30-2016
<b>Coriell ID:</b>	DEMONAT	<b>Date Received:</b>	11-30-2016
<b>Lab Accessioning Number:</b>	DEMONAT	<b>Date of Report:</b>	08-08-2013
<b>Ordering Physician:</b>	Dr. Edward Viner		

#### Risk of Developing Testicular Cancer Based on:

- **CPMC Testicular Cancer Variant 1 (rs995030)**

The CPMC is a research study investigating the utility of personalized genomic information on health and health behavior. At this time, the CPMC is reporting one genetic variant per health condition. **Women cannot develop testicular cancer**; therefore women will not receive risk estimates for testicular cancer. Your genetic variant result may be informative for male relatives (father, brothers, sons). These results were generated as part of this research study in a CLIA-approved laboratory.

More information about the study, how to interpret CPMC results, and how we calculate risk is available on our website <http://cpmc.coriell.org> or by contacting our genetic counselors. Participants may schedule an appointment with one of our board-certified genetic counselors through the web portal by clicking on "request an appointment". Our genetic counselors also can be reached by email at [cpmcgc@coriell.org](mailto:cpmcgc@coriell.org) or by phone at 888-580-8028.

This research report includes all data included in the clinical report as well as supplemental interpretations and educational material. Please see the report that follows for the official clinical report.

## Genetic Variant Result

### Testicular Cancer

Women cannot develop testicular cancer; therefore women will not receive risk estimates for testicular cancer. Your genetic variant result may be informative for male relatives (father, brothers, sons).

## Testicular Cancer

### Risk Due To Genetic Variant #1 (rs995030)

Your Result: 2 copies of the protective variant were detected (AA)

Non-Protective Variant = G    Protective Variant = A

#### **Women cannot develop testicular cancer.**

Your genetic variant result may be informative for your male relatives (father, brothers, and sons). Men with 1 or 2 copies of this protective variant are less likely to develop testicular cancer than men with 2 copies of the non-protective variant.

## Testicular Cancer - Variant #1 (rs995030)

We all have 2 copies of every gene, one from each of our parents.  
Each copy may have small changes called genetic variants.  
Some genetic variants are associated with an increased risk of disease.  
Some genetic variants are associated with a decreased risk of disease.

This genetic variant is **protective**. Men with 1 or 2 copies of this protective variant are **less** likely to develop testicular cancer than men with 2 copies of the non-protective variant.

### How Common Is This Variant?

Non-Protective Variant = G    Protective Variant = A

**GG - 13 in 100 people have 2 copies of the non-protective variant**

**GA - 36 in 100 people have 1 copy of the non-protective variant and 1 copy of the protective variant**

**AA - 51 in 100 people have 2 copies of the protective variant**

**This data is based on studies in African American populations.**



Gene: KITLG

Chromosome: 12q21.32

## Causes

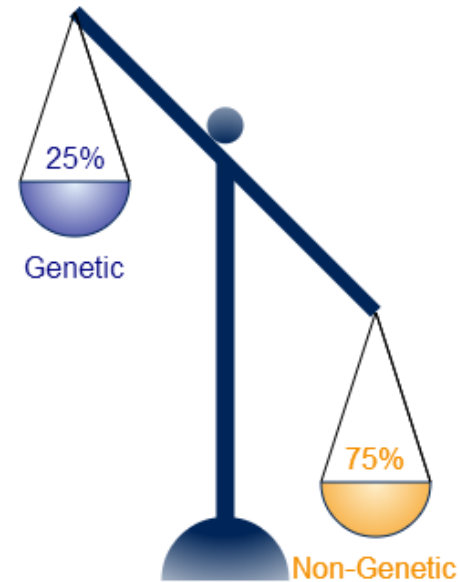
### Genetic vs. Non-Genetic Risk Factors

Testicular cancer can be caused by both genetic factors and non-genetic (or environmental) risk factors.

It is estimated that **non-genetic** factors (like ancestry and height) account for about **75%** of the risk of testicular cancer.

It is estimated that **25%** of the risk for testicular cancer is based on **genetic** risk factors. This estimate accounts for both known and unknown gene variants.

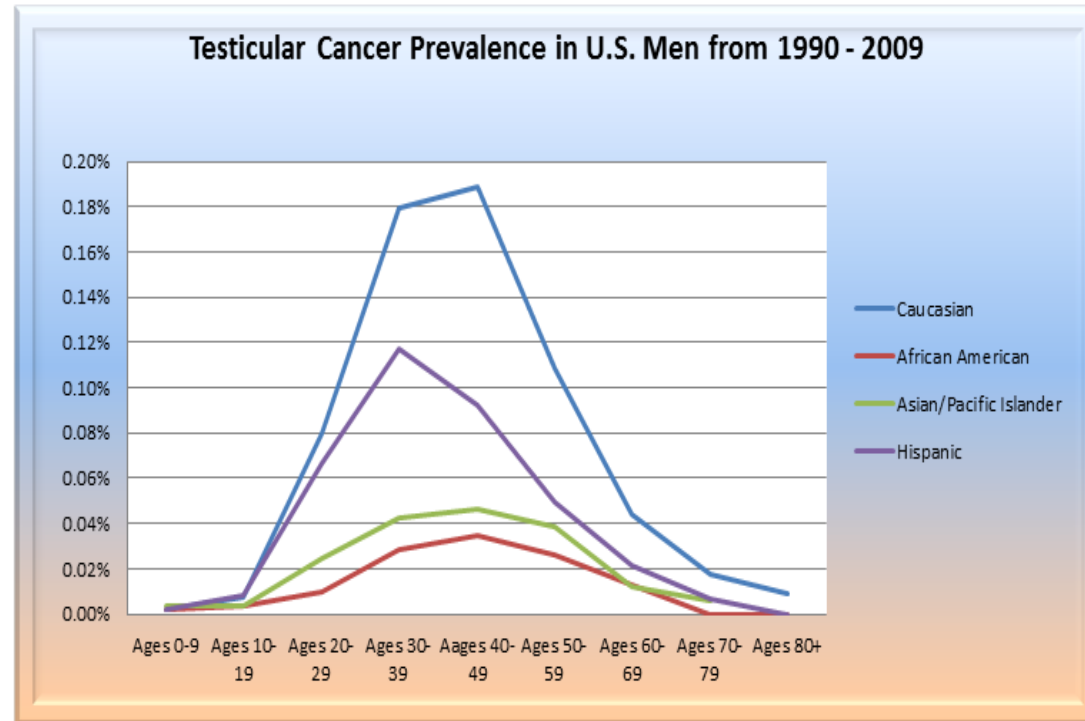
**There are many different genetic and non-genetic risk factors that contribute to the risk of testicular cancer. Women cannot develop testicular cancer; however, your genetic variant result may be informative for your male relatives (father, brothers, and sons).**



## How Common

Testicular cancer is more common among Caucasians.

Age and race contribute to the risk of testicular cancer.



## Limitations

### Testicular Cancer

- This result alone does NOT diagnose testicular cancer in men. Testicular cancer must be diagnosed by a health care provider.
- This result does NOT mean that a man will have or will absolutely develop testicular cancer.
- This result does NOT mean that a man will not develop testicular cancer in the future.
- This result ONLY assesses the risk for developing testicular cancer due to the factors presented in this report and does not mean that other genetic variants or risk factors for testicular cancer are present or absent.
- The relative risk information presented in this report represents the risk of developing testicular cancer for men who do not have a history of testicular cancer.
- Personal risk factors, such as age, family history or lifestyle, may have a greater impact on the risk to develop testicular cancer than any individual genetic variant.
- Risk estimates are based on current available literature.
- Although rare, it is possible that you may receive an incorrect result; 100% accuracy of reported results cannot be guaranteed.
- Occasionally there may be a specific variant on a gene chip that is not able to be read or interpreted. In this case you will not receive a result for that variant. It is expected that you will receive results for about 95% of variants approved by the ICOB.
- Relative risks used to estimate risk of disease for CPMC participants are based on groups of people with the same risk or protective factor as the individual CPMC participant. In some cases, the relative risk is estimated based upon an odds ratio and known or assumed disease prevalence.
- Separate risk estimates for each risk or protective factor have been given. Risk or protective factors may be related to each other and risk estimates cannot be combined.
- Risk information for non-genetic factors is based on information provided in the medical, family, and lifestyle questionnaire. If answers were not provided or if a “do not know” response was given, risk estimates for some factors may not be available.
- Risk information for non-genetic factors is based on information provided in the medical, family, and lifestyle questionnaire and may not be reflective of current risk if any of these factors have changed. Participants will be given the opportunity to update medical, family and lifestyle questionnaire responses periodically.
- Every effort will be made to provide participants with risk information based on their reported race/ethnicity. However, data may not be available for all races/ethnicities for all risk factors. Results will indicate which race/ethnicity the data given is based upon.



## Methods

### Testicular Cancer

**This condition and genetic variant(s) were approved by the Informed Cohort Oversight Board (ICOB)**

#### Test Methodology

Saliva samples were collected using Oragene DNA Collection Kits (DNA Genotek) and DNA was extracted manually according to the manufacturer's instructions. Purified DNA was quantified using UV absorbance at 260 nm. Five hundred nanograms of the resulting DNA from each sample were used as template in the Affymetrix Genome-Wide Human SNP Nsp/Sty 6.0 GeneChip assay. Data analysis was performed using Affymetrix Genotyping Console software.

See [CPMC Technical Paper](#) for genetic variant selection and reporting methodology.

[Risk interpretation based on Coriell's Testicular Cancer Risk Algorithm Version 1 (June 11, 2013)]

1. Stack, C. et al (2011). Genetic risk estimation in the Coriell Personalized Medicine Collaborative. *Genet Med.* 13(2):131-139.
2. Rapley, EA. et al (2009). A genome-wide association study of testicular germ cell tumor. *Nat Genet.* 41(7):807-810.
3. Nordsborg, RB. et al (2011). Cancer in first-degree relatives and risk of testicular cancer in Denmark. *Int J Cancer.* 129(10):2485-2491.
4. Jack, RH. et al (2007). Testis and prostate cancer incidence in ethnic groups in South East England. *Int J Androl.* 30:215-221.
5. Dieckmann, KP. et al (2008). Tallness is associated with risk of testicular cancer: evidence for the nutrition hypothesis. *Br J Cancer.* 99:1517-1521.
6. Cook, MB. et al (2010). A systematic review and meta-analysis of perinatal variables in relation to the risk of testicular cancer – experiences of the son. *Int J Epidemiol.* 39:1605-1618.
7. Czene, K. et al. (2002). Environmental and heritable causes of cancer among 9.6 million individuals in the Swedish family-cancer database. *Int J Cancer.* 99:260-266.
8. Howlader, N. et al. SEER Cancer Statistics Review, 1975-2009 (Vintage 2009 Populations), National Cancer Institute. Bethesda, MD, [http://seer.cancer.gov/csr/1975\\_2009\\_pops09/](http://seer.cancer.gov/csr/1975_2009_pops09/), based on November 2011 SEER data submission, posted to the SEER web site, April 2012.

## Sample Results



### Coriell Institute for Medical Research

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#### Clinical Report for Testicular Cancer Genetic Variant 1 (rs995030)

<b>Name:</b>	NATALIE DEMO	<b>Sample Type:</b>	Saliva
<b>Race/Ethnicity:</b>	Black or African-American	<b>Gender:</b>	Female
<b>Date of Birth:</b>		<b>Date Collected:</b>	11-30-2016
<b>Coriell ID:</b>	DEMONAT	<b>Date Received:</b>	11-30-2016
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<b>Ordering Physician:</b>	Dr. Edward Viner		

Name of Gene/Region: KITLG		Chromosomal Location: 12q21.32
Variants tested	Result	Reference Genotype
rs995030	AA	GG
<b>Interpretation</b>	<b>Women cannot develop testicular cancer.</b> This result may be informative for male relatives (father, brothers, sons). Men with this result are 85% less likely (or 0.15 times as likely) to develop testicular cancer as men with no copies of this protective variant.	
<b>Other Risks</b>	Other genetic variants and other risk factors including co-morbidities, lifestyle and family history may contribute to the risk of testicular cancer. For additional information on other risk factors please see the accompanying CPMC research report.	

Risk interpretation based on Coriell's Testicular Cancer Risk Algorithm Version 1 (June 11, 2013)

#### Test Limitations

DNA-based testing is highly accurate, however there are many sources of potential error including: mis-identification of samples, rare technical errors, trace contamination of PCR reactions, and rare genetic variants that interfere with analysis. There may be other variants, not included in this test, that influence the risk to develop testicular cancer. This test is not diagnostic for testicular cancer and cannot rule out the risk of developing testicular cancer in the future. Risk estimates are based on current available literature (see reference). This test or one or more of its components was developed and its performance characteristics determined by the Coriell Institute for Medical Research. It has not been approved by the Food and Drug Administration (FDA). The FDA has determined that such approval is not necessary. The Coriell Institute is regulated under the Clinical Laboratory Improvement Amendments (CLIA) of 1988 as qualified to perform high-complexity testing.

#### Test Methodology

Saliva samples were collected using Oragene DNA Collection Kits (DNA Genotek) and DNA was extracted manually according to the manufacturer's instructions or automatically using a DNAdvance Kit (Agencourt). Purified DNA was quantified using UV absorbance at 260 nm. Five hundred nanograms of the resulting DNA from each sample were used as template in the Affymetrix Genome-Wide Human SNP Nsp/Sty 6.0 GeneChip assay. Data analysis was performed using Affymetrix Genotyping Console software.

electronically signed by

Marie Hoover, PhD, Laboratory Director

#### References

1. Rapley, EA et al (2009). A genome-wide association study of testicular germ cell tumor. Nat Genet. 41(7):807-810.

This clinical report only includes data generated in the CLIA approved genotyping laboratory, for additional information please see the CPMC research report.