

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

Name:	STEVE CPMC	Gender:	Male
Date of Birth:		Date Collected:	11-30-2016
Coriell ID:	DEMOSTEVE	Date Received:	11-30-2016
Lab Accessioning Number:	DEMOSTEVE	Date of Report:	11-14-2012
Ordering Physician:	Dr. Edward Viner		

Risk of developing ulcerative colitis based on:

- **CPMC Ulcerative Colitis Variant 1 (rs11209026)**
- **Family History**

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. Since most common health conditions are caused by an interaction between more than one genetic factor and non-genetic factors such as lifestyle, the genetic variant risk in this report does not represent your complete genetic risk for ulcerative colitis. These results were generated as part of this research study in a CLIA-approved laboratory.

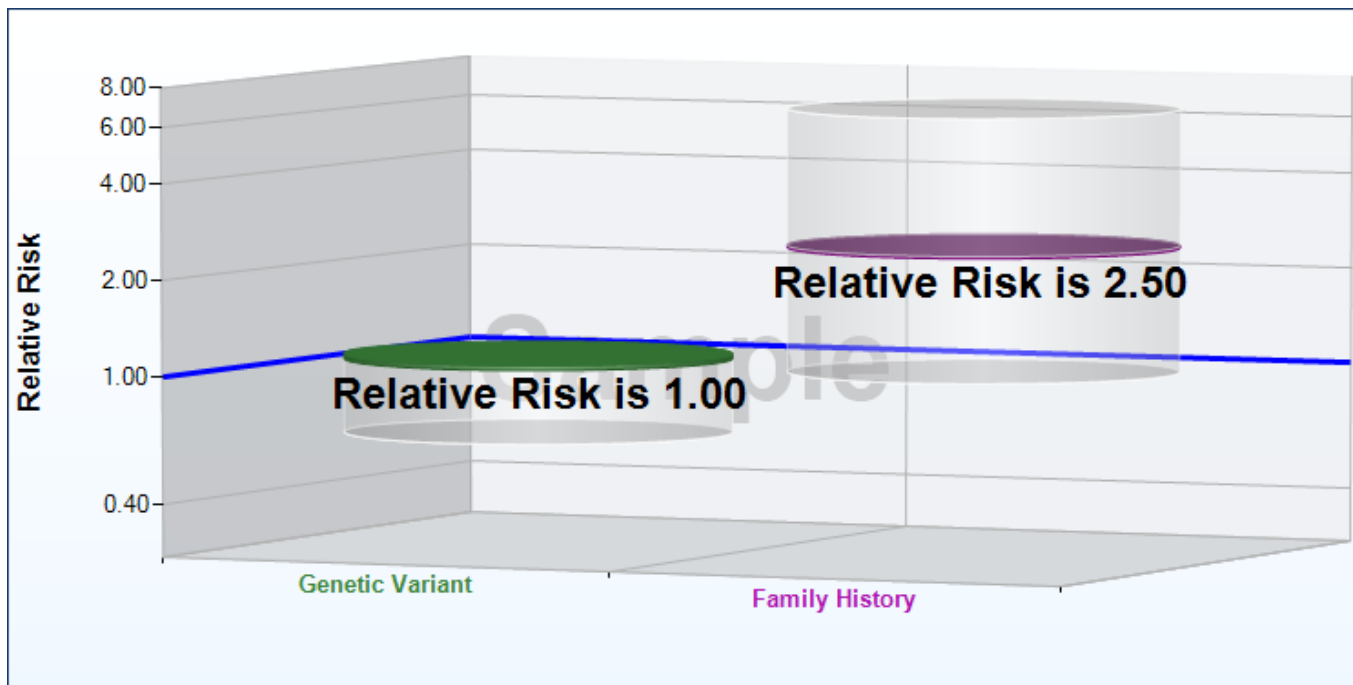
More information about the study, how to interpret CPMC results, and how we calculate risk is available on our website <http://cpmc.coriell.org> or by contacting our genetic counselors. Participants may schedule an appointment with one of our board-certified genetic counselors through the web portal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

Ulcerative Colitis

Risk factors may be related to each other and risk estimates cannot be combined.
 This graph provides a summary of the relative risks for genetic variant and family history.



You reported you are Caucasian, 60 years old or older; 3 in 1,000 Caucasians in your age group have ulcerative colitis.

Chart Color	Relative Risk Due To:	Your Risk	Minimum Risk	Maximum Risk	Interpretation
	Genetic Variant	1.00	0.57	1.00	You have 2 copies of the non-protective variant. Based on this result, you are at a higher risk to develop ulcerative colitis compared to someone with one or two copies of this protective genetic variant.
	Family History	2.50	1.00	6.80	Based on your family history, you are 2.5 times more likely to develop ulcerative colitis than someone who does not have a first degree relative (parent, sibling, or child) with either ulcerative colitis or Crohn's disease. Having one first degree relative with either ulcerative colitis or Crohn's disease contributes to your risk of ulcerative colitis.

Ulcerative Colitis

Risk Due To Genetic Variant #1 (rs11209026)

Your Result: 2 copies of the non-protective variant were detected (GG)

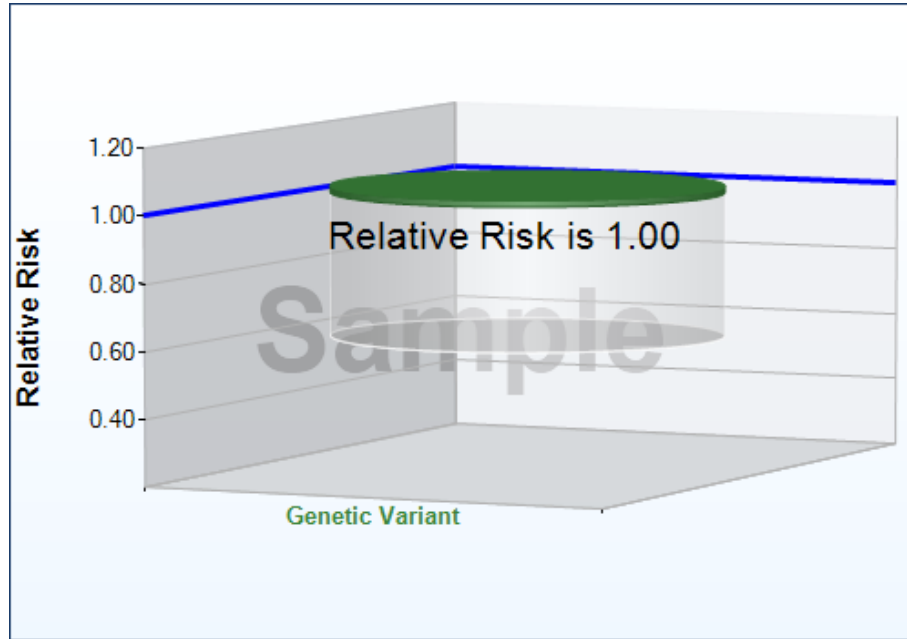
Non-Protective Variant = G Protective Variant = A

Chart Color	Your Risk	Minimum Risk	Maximum Risk	Interpretation
	1.00	0.57	1.00	You have 2 copies of the non-protective variant. Based on this result, you are at a higher risk to develop ulcerative colitis compared to someone with one or two copies of this protective genetic variant.

Genetic Variant Risk is based on the number of copies of this protective genetic variant.

People with one or two copies of the protective variant are compared to people with no copies of the protective variant to determine relative risk.

A relative risk less than 1.0 indicates a decreased risk.

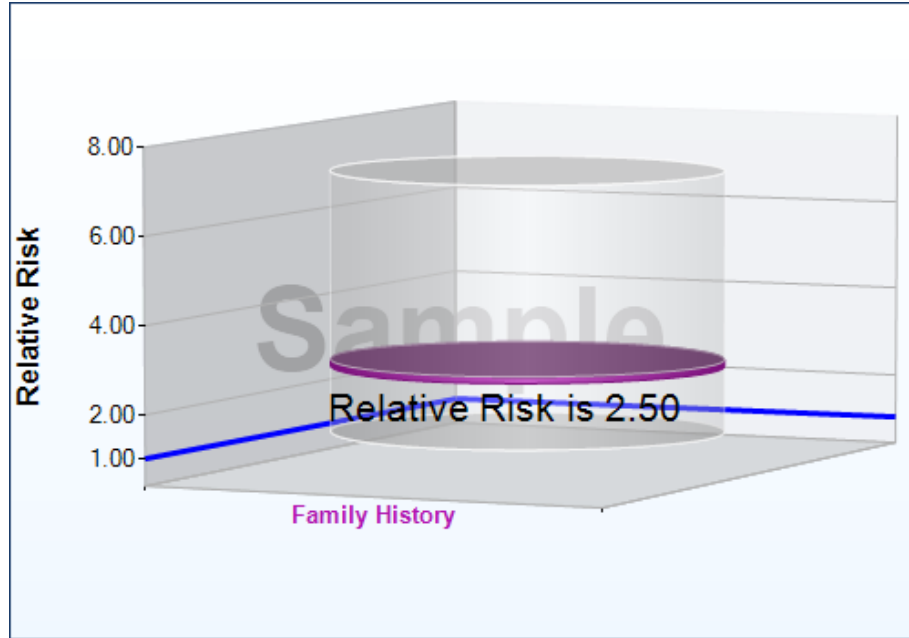


These risk estimates are based on studies in Caucasian populations.

Ulcerative Colitis Risk Due To Family History

You reported that a first degree relative (parent, sibling or child) has ulcerative colitis or Crohn's disease.

Chart Color	Your Risk	Minimum Risk	Maximum Risk	Interpretation
	2.50	1.00	6.80	<p>Based on your family history, you are 2.5 times more likely to develop ulcerative colitis than someone who does not have a first degree relative (parent, sibling, or child) with either ulcerative colitis or Crohn's disease.</p> <p>Having one first degree relative with either ulcerative colitis or Crohn's disease contributes to your risk of ulcerative colitis.</p>



Risk is compared based on family history.

People with one or more first degree relatives (parents, siblings, or children) with either ulcerative colitis or Crohn's disease are compared to people with no first degree relatives with either ulcerative colitis or Crohn's disease to determine relative risk of developing ulcerative colitis.

A relative risk greater than 1.0 indicates an increased risk.

These risk estimates are based on studies in Caucasian populations.

Ulcerative Colitis - Variant #1 (rs11209026)

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**. Having one or two copies of this variant **lowers** your risk for ulcerative colitis.

How Common Is This Variant?

Non-Protective Variant = G Protective Variant = A

GG - 89 in 100 people have 2 copies of the non-protective variant

GA - 10 in 100 people have 1 copy of the non-protective variant and 1 copy of the protective variant

AA - 1 in 100 people have 2 copies of the protective variant

This data is based on studies in Caucasian populations.



Gene: IL23R

Chromosome: 1p31.3

Causes

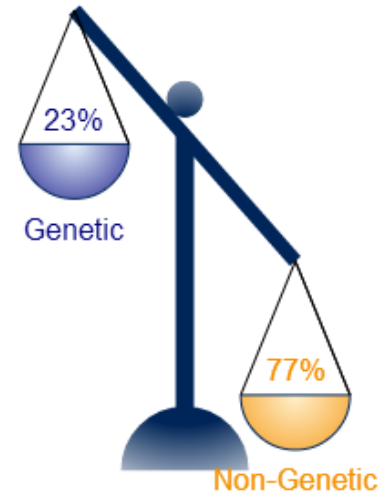
Genetic vs. Non-Genetic Risk Factors

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

It is estimated that **non-genetic** factors (like use of the prescription drug Accutane) account for about **77%** of the risk of ulcerative colitis.

It is estimated that **23%** of the risk for ulcerative colitis 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 ulcerative colitis. We are only able to tell you about 1 genetic risk factor at this time.

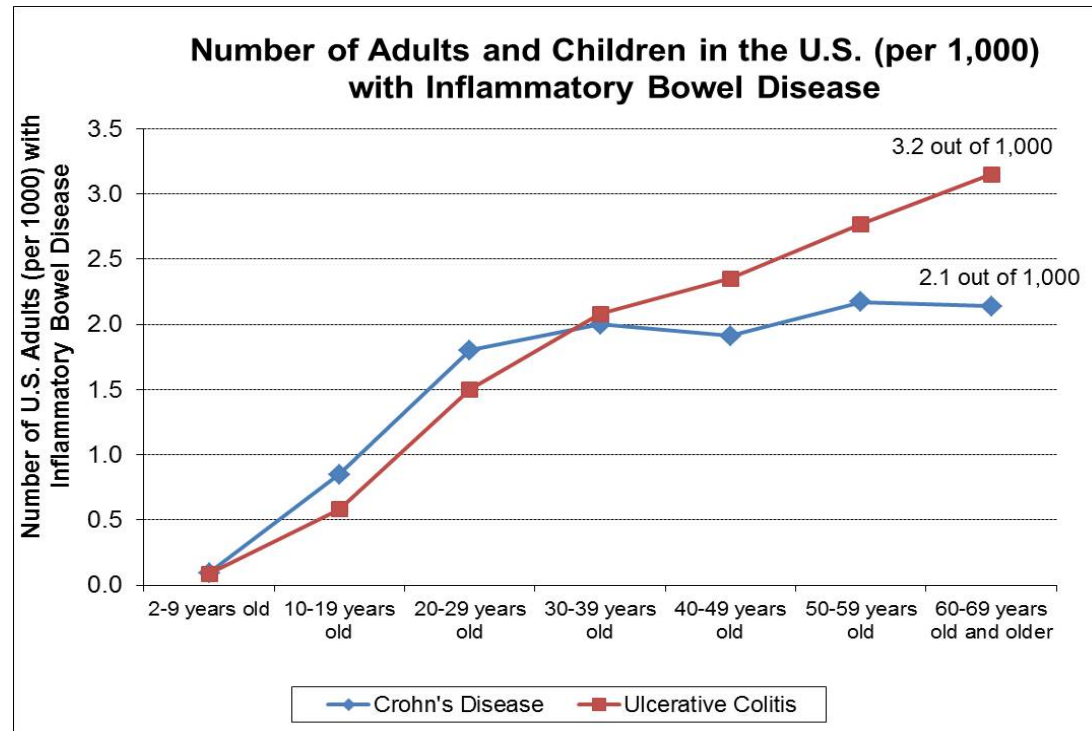


How Common

Ulcerative colitis is more common than Crohn's disease.

You reported you are Caucasian, 60 years old or older; 3 in 1,000 Caucasians in your age group have ulcerative colitis.

Your age contributes to your risk of inflammatory bowel diseases like ulcerative colitis and Crohn's disease.



Limitations

Ulcerative Colitis

- This result alone does NOT diagnose ulcerative colitis. Ulcerative colitis must be diagnosed by your health care provider.
- This result does NOT mean that you have or will absolutely develop ulcerative colitis.
- This result does NOT mean that you will not develop ulcerative colitis in the future.
- This result ONLY assesses your risk for developing ulcerative colitis due to the factors presented in this report and does not mean that other genetic variants or risk factors for ulcerative colitis are present or absent.
- Personal risk factors, such as age, family history or lifestyle, may have a greater impact on your risk to develop ulcerative colitis 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 you provided in your medical, family, lifestyle questionnaire. If you did not provide answers or if you answered "do not know", risk estimates for some factors may not be available.
- Risk information for non-genetic factors is based on information you provided in your medical, family, lifestyle questionnaire and may not be reflective of your current risk if any of these factors have changed. You will be given the opportunity to update your medical, family and lifestyle questionnaire responses annually.
- Every effort will be made to provide you with risk information based on your reported race/ethnicity. However, data may not be available for all races/ethnicities for all risk factors. Please see your individual results to determine which race/ethnicity the data given is based on.
- For some risk factors data may be provided by gender. Every effort will be made to provide you with risk information based on your reported gender. However, when risk data is not available for both genders, risk results for the available gender will be provided.

Methods

Ulcerative Colitis

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 Ulcerative Colitis Risk Algorithm Version 1 (November 12, 2012)]

1. Stack, C. et al (2011). Genetic risk estimation in the Coriell Personalized Medicine Collaborative. *Genet Med.* 13(2):131-139.
2. Kappelman, M.D. et al. (2007). The prevalence and geographic distribution of Crohn's disease and ulcerative colitis in the United States. *Clinical Gastroenterology and Hepatology.* 5:1424-1429.
3. Silverberg, M.S. et al. (2009). Ulcerative colitis-risk loci on chromosomes 1p36 and 12q15 found by genome-wide association study. *Nature Genetics.* 41:216-220.
4. Geary, R.B. et al. (2010). Population-based cases control study of inflammatory bowel disease risk factors. *Journal of Gastroenterology and Hepatology.* 25:325-333.

Sample Results



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Clinical Report for Ulcerative Colitis Genetic Variant 1 (rs11209026)

Name:	STEVE CPMC	Sample Type:	Saliva
Race/Ethnicity:	White (Caucasian)	Gender:	Male
Date of Birth:		Date Collected:	11-30-2016
Coriell ID:	DEMOSTEVE	Date Received:	11-30-2016
Lab Accessioning Number:	DEMOSTEVE	Date of Report:	11-14-2012
Ordering Physician:	Dr. Edward Viner		

Name of Gene/Region: IL23R		Chromosomal Location: 1p31.3
Variants tested	Result	Reference Genotype
rs11209026	GG	GG
Interpretation	Individuals with this result are at a higher risk to develop ulcerative colitis compared to someone with one or two copies of the protective variant. These results are based on studies in Caucasian populations. When race/ethnicity specific risk estimates are not available, risk estimates based on Caucasian populations are provided.	
Other Risks	Other genetic variants and other risk factors including co-morbidities, lifestyle and family history may contribute to the risk of ulcerative colitis. For additional information on other risk factors please see the accompanying CPMC research report.	

Risk interpretation based on Coriell's Ulcerative Colitis Risk Algorithm Version 1 (November 12, 2012)

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 ulcerative colitis. This test is not diagnostic for ulcerative colitis and cannot rule out the risk of developing ulcerative colitis 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

{0}, PhD, Laboratory Director

References

1. Silverberg, M.S. et al. (2009). Ulcerative Colitis-risk loci on chromosomes 1p36 and 12q15 found by genome-wide association study. Nature Genetics. 41:216-220.

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