

# PREVENTION REQUIRES KNOWLEDGE





This report was prepared for

Example Data

# **Genetic Predisposition Report**

The data presented in this report assumes\*; Gender: Male Ethnicity: European Customer ID: Example Data \* If the data assumed is incorrect, please contact the provider of your test to have it changed. English (US) CR-001 November 27, 2009, 11:38 am

# Before looking at your results

#### Scope of the genetic test

The purpose of this molecular genetic test is to ascertain if you or the person being tested is carrying mutation(s) predisposing you to or causing the specific diseases or conditions covered by the test. It is important to understand that due to the complexity of DNA based testing and the important implications of the test results, you may want to consult your physician or a genetic counselor. This report is provided to you for informational and educational purposes, and it does not replace a visit to a physician, nor does it replace the advice or services of a physician.

#### Nature of the genetic test

The molecular genetic test is based on the study of SNP (Single Nucleotide Polymorphisms) which are genetic sites that vary between individuals with an incidence equal to or higher than 1%. Since this technique looks at the sites that differ most from one individual to another, this test is very successful at identifying DNA markers associated with diseases or conditions. Techniques based on SNP provide information about the most common variation in the human genome for research - and you - to study. It is still important to keep in mind that rare variations that affect certain diseases may not be covered by a SNP test as they still have not been discovered, or because their incidence is very low.

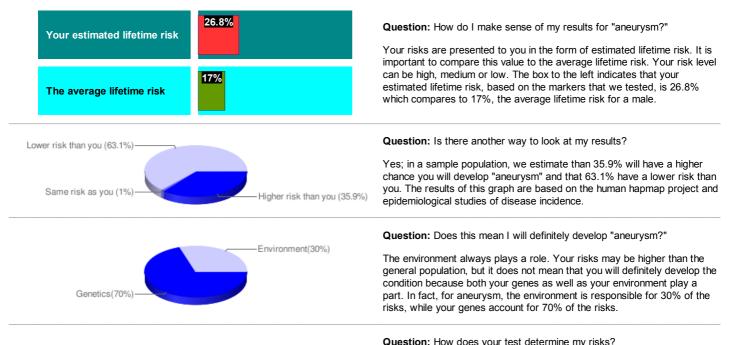
#### Limits of the genetic test

The information presented in this report is based on the latest scientific research. The diseases that are included in this report have generally been replicated in more than one important ethnic group. This yields a lot of confidence to the results that are reported. However, we cannot exclude the possibility that future research may improve upon the accuracy of the results. It is also important to note that not all disease associations have been reported for all ethnic groups and for this reason, genetic research is not comprehensive. When a disease association for your ethnicity has not yet been reported, the closest ethnicity can be used to generate the part of the report covering said disease association; in which case it will be mentioned in the report which ethnicity is assumed for each disease covered. A genetic test based on SNP cannot reveal large microsatellite genome rearrangement, insertion/deletion events or copy number variations.

#### How to interpret your results

#### Example & common questions

The information for each condition is presented in four sections. Let us take the example of Aneurysm to illustrate how the report should be read.



Question: How does y	our test determine my risks
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Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs10958409	intergenic	AG	1.22	0.2688
rs1333040	intergenic	TT	1.23	0.3136
rs700651	BOLL	GA	1.05	0.42

We look at segments of your DNA where one-allele difference modifies your odds of developing aneurysm. At the location(s) displayed to the left, your genotype indicates the DNA that you inherited from both of your parents. Each letter corresponds to a different parent. Certain letters are associated with a higher risk. Your risk will reflect the number of risk allele copies that are present in your genotype for each marker.

#### Example & common questions (cont'd)

#### Question: What should I do next?

First, we recommend that you bring this report to your doctor. Your doctor will be able to do an initial assessment of symptoms and determine if you have this condition or if it has not yet developed. We encourage you to contact us for any technical questions you may have or to instruct your doctor to contact the distributor you bought your test from.

Question: Why is the condition not reported for my ethnicity?



It requires quite a lot of data to be able to estimate a lifetime risk for a condition. First, the data about the incidence of the disease for both genders of the ethnic group must be available. Second, genetic twin studies to determine the contribution of the environment to the disease must also have been reported for the ethnic group. Finally, each SNP that is tested for must have been successfully reported for - again - the same ethnic group.

This high requirement on research data has the consequence that the lifetime risk cannot be explicitly given for all ethnic groups because the required data may not yet be available. However, the SNP that we report here have generally been replicated in multiple different ethnic groups (e.g. European, Japanese, African) and are therefore believed to be independent of ethnicity.

# Summary of your results

The condensed view of your results is presented here. More details about each condition are presented in the following pages, sorted alphabetically. The genetic risk scoring/relative risk calculation methods used to calculate your results were developed and their performance characteristics determined by the use of standard practices. Based on your DNA and the conditions covered by this test, the following is your summary of results.

Name of the condition	Your lifetime risk	The normal risk	Your genetic risk level
	High genetic risk level		
Aneurysm	26.8%	17%	high
Atrial fibrillation	30.7%	25%	high
Coronary heart disease	74.2%	49%	high
Lung cancer	34.9%	17.2%	high
Peripheral vascular disease	20.4%	14.5%	high
	Medium genetic risk level		
Migraine	23.2%	20%	medium
Obesity	25%	25%	medium
Osteoarthritis	40.9%	43%	medium
Prostate cancer	10.5%	16%	medium
Skin cancer	31.2%	35%	medium
Type 2 diabetes	21.4%	24%	medium
Venous thromboembolism	23.3%	25%	medium
	Low genetic risk level		
Age related macular degeneration	6.8%	8%	low
Alzheimer disease	3.3%	6%	low
Bladder cancer	4.6%	4.1%	low
Breast cancer	0.4%	0.4%	low
Celiac disease	0.7%	1%	low
Colorectal cancer	5.4%	6%	low
Gastric cancer	2%	2.3%	low
Graves disease	0.2%	1%	low
Lupus	0.2%	0.25%	low
Multiple sclerosis	0.4%	0.2%	low
Psoriasis	2.3%	11.4%	low
Rheumatoid arthritis	0.6%	1%	low
Type 1 diabetes	1.3%	1%	low
Source : Example Data			

# Age related macular degeneration

Macular degeneration is a medical condition that usually occurs in older adults which results in a loss of vision in the center of the visual field because of damage to the retina. It occurs in dry and wet forms. It is a major cause of blindness in the elderly.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 6.8 out of every 100 person. These results indicate your odds of developing this condition.

Individuals from the average population are estimated to develop this condition in 8 out of every 100 person. These results apply to an average sample of european

# Summary of your genetic results

ancestry.

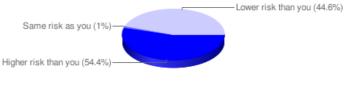
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

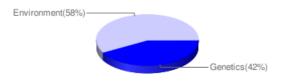
Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs800292	C2	CC	0.67	0.6084
rs1061170	CFH	СТ	1.26	0.4032
Source : Examp	le Data			

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

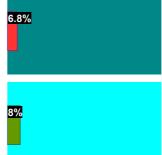


#### Factors responsible for Age related macular degeneration









### Alzheimer's disease

Alzheimer's disease is the most common form of dementia. It is incurable, degenerative, and terminal. Generally, it is diagnosed in people over 65 years of age, although the less-prevalent early-onset Alzheimer's can occur much earlier.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 3.3 out of every 100 person. These results indicate your odds of developing this condition.

Individuals from the average population are estimated to develop this condition in 6 out of every 100 person. These results apply to an average sample of european

# Summary of your genetic results

ancestry.

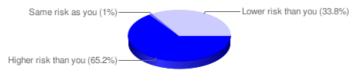
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

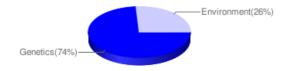
Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs4420638	APOC1	AA	0.57	0.6724
rs429358	ApoE	ТТ	0.96	0.9794
Source : Examp	le Data			

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

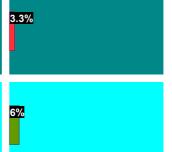


#### Factors responsible for Alzheimer disease









### Aneurysm

An aneurysm is a localized, blood-filled dilation of a blood vessel caused by disease or a weakening of the vessel wall. As the size of an aneurysm increases, there is an increased risk of rupture, which can result in severe hemorrhage or other complications including sudden death.

#### Understand your risks high

Your estimated lifetime risk

condition

ancestry

Your genetic predisposition towards this disease is high. Note that for your risk to be considered high, either of these two requirements must be met: Either your risk of developing the condition is higher than 20% and higher than the general population by 1.2x - or - your risk of developing the condition is three times higher than the general population.

Individuals with your genetic variants are estimated to develop this condition in 26.8 out of every 100 person. These results indicate your odds of developing this

Individuals from the average population are estimated to develop this condition in 17 out of every 100 person. These results apply to an average sample of european

Summary of your genetic results

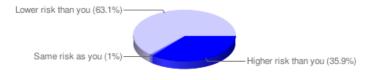
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs10958409	intergenic	AG	1.22	0.2688
rs1333040	intergenic	ТТ	1.23	0.3136
rs700651	BOLL	GA	1.05	0.42

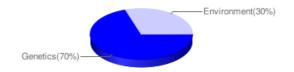
### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Aneurysm

Although your risks of developing this condition may seem high to you, keep in mind that your environment is a significantly responsible factor as well. This gives you a degree of control over your odds.







European

17%

26.8%

# Atrial fibrillation

Atrial fibrillation (AF) is the most common cardiac abnormal heart rhythm. It can often be identified by taking a pulse and observing that the heartbeats do not occur at regular intervals. Risk increases with age, with 8% of people over 80 having AF.

#### Understand your risks high

Your estimated lifetime risk

condition

Your genetic predisposition towards this disease is high. Note that for your risk to be considered high, either of these two requirements must be met: Either your risk of developing the condition is higher than 20% and higher than the general population by 1.2x - or - your risk of developing the condition is three times higher than the general population.

Individuals with your genetic variants are estimated to develop this condition in **30.7** out of every **100** person. These results indicate your odds of developing this

Individuals from the average population are estimated to develop this condition in 25

out of every 100 person. These results apply to an average sample of european ancestry.

#### Summary of your genetic results

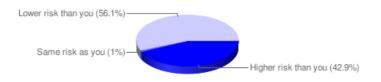
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

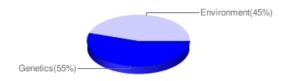
	cus	Your genotype	Relative Risk	Genotype frequency
rs10033464 inter	ergenic (	GG	0.96	0.81
rs2200733 LOC	C729065	тс	1.28	0.2112

Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

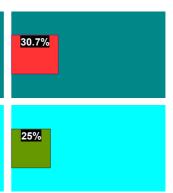


#### Factors responsible for Atrial fibrillation









#### Bladder cancer

Bladder cancer is a disease in which abnormal cells multiply without control in the bladder. It is the fourth most common type of cancer in men and the ninth most common cancer in women. Untreated, superficial tumors may gradually begin to infiltrate the muscular wall of the bladder.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 4.6 out of every 100 person. These results indicate your odds of developing this condition.

Individuals from the average population are estimated to develop this condition in **4.1 out of every 100** person. These results apply to an average sample of european

# ancestry.

#### Summary of your genetic results

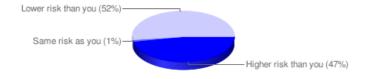
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

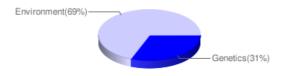
Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs9642880	intergenic	GT	1.01	0.4902
rs710521	TP63	AA	1.1	0.4761
Source : Examp		~~	1.1	0.4701

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

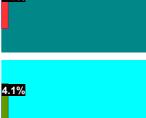


#### Factors responsible for Bladder cancer









#### **Breast cancer**

Breast cancer is a cancer that starts in the cells of the breast. Worldwide, breast cancer is the second most common type of cancer after lung cancer and the fifth most common cause of cancer death.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 0.4 out of every 100 person. These results indicate your odds of developing this condition.

The average lifetime risk Individuals from the average population are estimated to develop this condition in **0.4 out of every 100** person. These results apply to an average sample of european ancestry.

### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

0.4%

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs4986761	ATM	TT	1	1
rs3218695	ATM	CC	1	1
rs3092856	ATM	CC	1	1
rs3218707	ATM	GG	1	1
rs1800058	ATM	CC	1	0.9025
rs1042522	TP53	GC	0.97	0.3542
rs1801673	ATM	AA	0.91	0
rs1800056	ATM	TT	1	0.9604
rs1800057	ATM	CC	1	0.9409
rs1800057 Source : Example		CC	1	0.9409

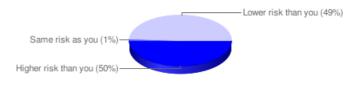
Breast cancer (cont'd)



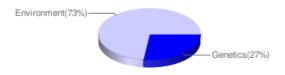


# Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Breast cancer



### Celiac disease

Celiac disease is an autoimmune disorder of the small intestine that occurs in genetically predisposed people of all ages from middle infancy upwards. Symptoms include chronic diarrhea, failure to thrive (in children) and fatigue.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 0.7 out of every 100 person. These results indicate your odds of developing this condition.

The average lifetime risk Individuals from the average population are estimated to develop this condition in 1 out of every 100 person. These results apply to an average sample of european ancestry.

### Summary of your genetic results

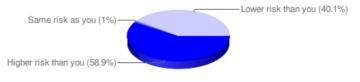
To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

1%

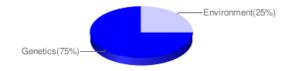
Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs6822844	IL21	GG	0.97	0.7225
rs2187668	HLA	GG	0.74	0.8464
Source : Examp	le Data			

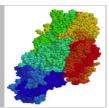
#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



### Factors responsible for Celiac disease







### **Colorectal cancer**

Colorectal cancer includes cancerous growths in the colon, rectum, and appendix. It is the second most common form of cancer related death in the Western world. Many colorectal cancers are thought to arise from adenomatous polyps in the colon.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 5.4 out of every 100 person. These results indicate your odds of developing this condition.

#### The average lifetime risk Individuals from the average

Individuals from the average population are estimated to develop this condition in 6 **out of every 100** person. These results apply to an average sample of european ancestry.

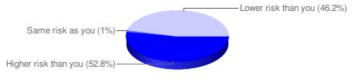
### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

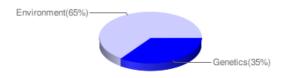
Marker	Locus	Your genotype	Relative Risk	Genotype frequency	
rs12953717	SMAD7	СТ	1	0.4712	
rs6983267	intergenic	ТТ	0.74	0.2601	
rs4939827	SMAD7	ТТ	1.17	0.2209	
rs4464148	SMAD7	СТ	1.03	0.4118	

### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

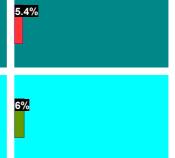


#### Factors responsible for Colorectal cancer









### Coronary heart disease

Coronary heart disease is the leading cause of death in the United States, the United Kingdom, and Canada, killing one person every 30 seconds in these countries alone. Even when medical intervention is successful, the physical capabilities of the body are often severely impaired.

#### Understand your risks high

Your estimated lifetime risk

condition

ancestry.

Your genetic predisposition towards this disease is high. Note that for your risk to be considered high, either of these two requirements must be met: Either your risk of developing the condition is higher than 20% and higher than the general population by 1.2x - or - your risk of developing the condition is three times higher than the general population.

Individuals with your genetic variants are estimated to develop this condition in **74.2** out of every **100** person. These results indicate your odds of developing this

Individuals from the average population are estimated to develop this condition in **49 out of every 100** person. These results apply to an average sample of european

### Summary of your genetic results

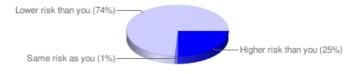
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

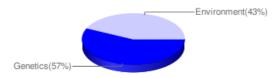
Marker	Locus	Your genotype	Relative Risk	Genotype frequency	
rs10757274	intergenic	GG	1.15	0.1296	
rs2383206	intergenic	GG	1.22	0.2809	
rs2383207	intergenic	GG	1.08	0.2304	
rs10757278	intergenic	GA	1	0.5	

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

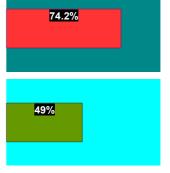


#### Factors responsible for Coronary heart disease









#### Gastric cancer

Stomach or gastric cancer can develop in any part of the stomach and may spread throughout the stomach and to other organs; particularly the esophagus, lungs and the liver. Stomach cancer is the fourth most common cancer worldwide.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

 Your estimated lifetime risk
 2%

 Individuals with your genetic variants are estimated to develop this condition in 2 out of every 100 person. These results indicate your odds of developing this condition.
 2%

 The average lifetime risk
 100 person. These results indicate your odds of develop this condition.
 2%

 Individuals from the average population are estimated to develop this condition in 2.3 out of every 100 person. These results apply to an average sample of european ancestry.
 2.3%

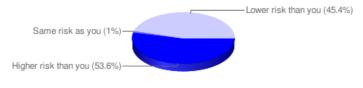
#### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

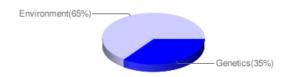
Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs1801133	MTHFR	CC	0.89	0.4761
Source : Examp	le Data			

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Gastric cancer







#### Graves' disease

Graves' disease is an autoimmune disease. It most commonly affects the thyroid, causing it to grow to twice its size or more (goiter), be overactive, with related hyperthyroid symptoms such as increased heartbeat, muscle weakness, disturbed sleep, and irritability.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk 0.2% Individuals with your genetic variants are estimated to develop this condition in 0.2 out of every 100 person. These results indicate your odds of developing this condition.

The average lifetime risk Individuals from the average population are estimated to develop this condition in 1 out of every 100 person. These results apply to an average sample of european ancestry.

### Summary of your genetic results

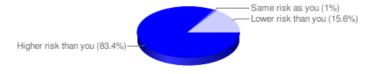
To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition

1%

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs1800630	TNF	CC	0.64	0.7225
rs1800629	TNF	GG	0.62	0.6889
rs1799964	LTA	TT	0.57	0.6241

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



### Factors responsible for Graves disease









### Lung cancer

Lung cancer is a disease of uncontrolled cell growth in tissues of the lung. This growth may lead to metastasis, which is the invasion of adjacent tissue and infiltration beyond the lungs. Lung cancer is the most common cause of cancer-related death in men and the second most common in women.

#### Understand your risks high

Your genetic predisposition towards this disease is high. Note that for your risk to be considered high, either of these two requirements must be met: Either your risk of developing the condition is higher than 20% and higher than the general population by 1.2x - or - your risk of developing the condition is three times higher than the general population.

Individuals with your genetic variants are estimated to develop this condition in **34.9** out of every **100** person. These results indicate your odds of developing this

Individuals from the average population are estimated to develop this condition in **17.2 out of every 100** person. These results apply to an average sample of

The average lifetime risk

Your estimated lifetime risk

condition

#### Summary of your genetic results

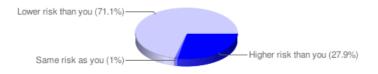
european ancestry.

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs1051730	CHRNA3	TT	1.42	0.1521
rs951266	CHRNA5	TT	1.43	0.1521

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

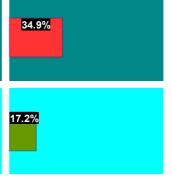


#### Factors responsible for Lung cancer









#### Lupus

Lupus is a chronic autoimmune connective tissue disease that can affect any part of the body. As occurs in other autoimmune diseases, the immune system attacks the body's cells and tissues, resulting in inflammation and tissue damage. There is currently no cure. Lupus can be fatal, although with recent medical advances, fatalities are becoming increasingly rare.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

 Your estimated lifetime risk
 0.2%

 Individuals with your genetic variants are estimated to develop this condition in 0.2 out of every 100 person. These results indicate your odds of developing this condition.
 0.2%

 The average lifetime risk
 Individuals from the average population are estimated to develop this condition in 0.2 out of every 100 person. These results apply to an average sample of european ancestry.
 0.25%

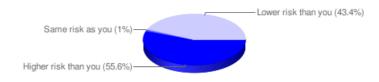
### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

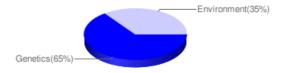
Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs10488631	TNPO3	TC	1.63	0.2112
rs7574865	STAT4	GT	1.24	0.3542
rs9888739	ITGAM	CC	0.94	0.81
rs1800630	TNF	CC	0.82	0.7225
rs2187668	HLA	GG	0.83	0.8464
rs1800629	TNF	GG	0.73	0.6889
Source : Example	Data			

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Lupus







### Migraine

Migraine is a neurological syndrome characterized by altered bodily perceptions, headaches, and nausea. The typical migraine headache is unilateral and pulsating, lasting from 4 to 72 hours. Symptoms include nausea, vomiting, photophobia (increased sensitivity to bright light), and hyperacusis (increased sensitivity to sound).

#### Understand your risks medium

Your genetic predisposition towards this disease is medium. Note that for your risk to be considered medium, this requirement must be met: Your risk of developing the condition is higher than 7% (and your risk does not qualify as high).

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 23.2 out of every 100 person. These results indicate your odds of developing this condition.

Individuals from the average population are estimated to develop this condition in **20 out of every 100** person. These results apply to an average sample of european

# Summary of your genetic results

ancestry.

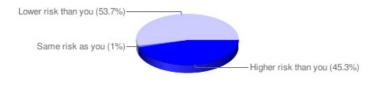
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs1801133	MTHFR	CC	1.16	0.4761
Source · Examp	le Data			

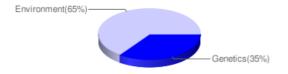
### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



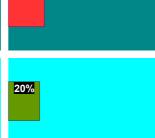
### Factors responsible for Migraine

Although your risks of developing this condition may seem high to you, keep in mind that your environment is a significantly responsible factor as well. This gives you a degree of control over your odds.









23.2%

### Multiple sclerosis

Multiple sclerosis (MS) is an autoimmune condition in which the immune system attacks the central nervous system. The disease onset usually occurs in young adults, and it is more common in females. There is no known cure for MS. Treatments attempt to return function after an attack, prevent new attacks, and prevent disability.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk<br/>Individuals with your genetic variants are estimated to develop this condition in 0.4<br/>out of every 100 person. These results indicate your odds of developing this<br/>condition.0.4%The average lifetime risk<br/>Individuals from the average population are estimated to develop this condition in 0.2<br/>out of every 100 person. These results apply to an average sample of european0.2%

### Summary of your genetic results

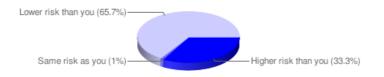
ancestry.

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs12722489	IL2RA	GG	1.06	0.6889
rs6897932	IL7R	CC	1.07	0.5776
rs3135388	HLA	СТ	1.67	0.3078

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Multiple sclerosis

Although your risks of developing this condition may seem high to you, keep in mind that your environment is a significantly responsible factor as well. This gives you a degree of control over your odds.

Environment(60%) Genetics(40%)





### Obesity

Obesity is a medical condition in which excess body fat has accumulated to the extent that it may have an adverse affect on health, leading to reduced life expectancy. Obesity is a leading preventable cause of death worldwide, and it is viewed as one of the most serious public health problems of present times.

#### Understand your risks medium

Your genetic predisposition towards this disease is medium. Note that for your risk to be considered medium, this requirement must be met: Your risk of developing the condition is higher than 7% (and your risk does not qualify as high).

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 25 out of every 100 person. These results indicate your odds of developing this condition.

Individuals from the average population are estimated to develop this condition in **25** out of every **100** person. These results apply to an average sample of european

# Summary of your genetic results

ancestry.

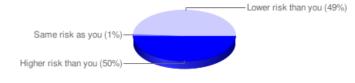
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

Marker	Locus	Your genotype	Relative Risk	Genotype frequency	
rs3764220	SCG3	AA	1	1	
Source : Examp	le Data				

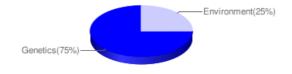
### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Obesity

Although your risks of developing this condition may seem high to you, keep in mind that your environment is a significantly responsible factor as well. This gives you a degree of control over your odds.







25%

25%

### Osteoarthritis

Osteoarthritis, is a group of diseases and mechanical abnormalities that causes degradation of joints, including articular cartilage and the subchondral bone next to it. Osteoarthritis is the most common form of arthritis and the leading cause of chronic disability. Osteoarthritis affects nearly 33 million people in North America, accounting for 25% of visits to primary care physicians.

#### Understand your risks medium

Your estimated lifetime risk

Your genetic predisposition towards this disease is medium. Note that for your risk to be considered medium, this requirement must be met: Your risk of developing the condition is higher than 7% (and your risk does not qualify as high).

condition.
The average lifetime risk
Individuals from the average population are estimated to develop this condition in 43

Individuals with your genetic variants are estimated to develop this condition in **40.9 out of every 100** person. These results indicate your odds of developing this

out of every 100 person. These results apply to an average sample of european ancestry.

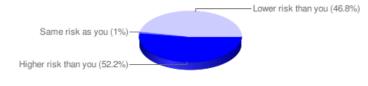
### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

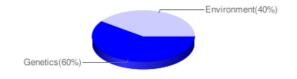
Marker	Locus	Your genotype	Relative Risk	Genotype frequency	
rs4140564	PTGS2	TT	0.95	0.8464	
Source · Examp	le Data				

### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

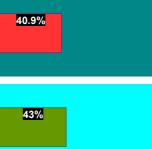


### Factors responsible for Osteoarthritis









### Peripheral vascular disease

Peripheral vascular disease includes all diseases caused by the obstruction of large arteries in the arms and legs. The prevalence of peripheral vascular disease in people aged over 55 years is 10%-25% and increases with age; 70%-80% of affected individuals are asymptomatic; only a minority ever require revascularisation or amputation.

#### Understand your risks high

Your estimated lifetime risk

condition

Your genetic predisposition towards this disease is high. Note that for your risk to be considered high, either of these two requirements must be met: Either your risk of developing the condition is higher than 20% and higher than the general population by 1.2x - or - your risk of developing the condition is three times higher than the general population.

Individuals with your genetic variants are estimated to develop this condition in 20.4 out of every 100 person. These results indicate your odds of developing this

Individuals from the average population are estimated to develop this condition in **14.5 out of every 100** person. These results apply to an average sample of

### Summary of your genetic results

european ancestry.

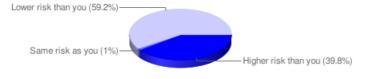
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

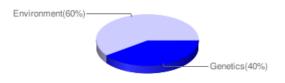
Marker	Locus	Your genotype	Relative Risk	Genotype frequency	
rs951266	CHRNA5	TT	1.41	0.1521	
Source : Exan	nple Data				

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

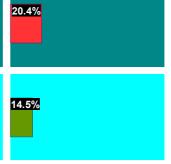


# Factors responsible for Peripheral vascular disease









#### Prostate cancer

Prostate cancer is a disease in which cancer develops in the prostate, a gland in the male reproductive system. Rates of prostate cancer vary widely across the world. It is more common in Europe and most common in the United States. It is the most common type of cancer in men in the United States and the second most common in the United Kingdom, after lung cancer.

#### Understand your risks medium

Your genetic predisposition towards this disease is medium. Note that for your risk to be considered medium, this requirement must be met: Your risk of developing the condition is higher than 7% (and your risk does not qualify as high).

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 10.5 out of every 100 person. These results indicate your odds of developing this condition.

#### The average lifetime risk Individuals from the average

Individuals from the average population are estimated to develop this condition in **16 out of every 100** person. These results apply to an average sample of european ancestry.

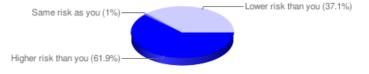
### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

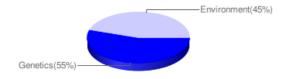
Marker	Locus	Your genotype	Relative Risk	Genotype frequency	
rs6983267	intergenic	TT	0.79	0.2601	
rs16901979	intergenic	CC	0.97	0.9409	
rs1859962	intergenic	GT	0.94	0.4982	
rs1447295	intergenic	CC	0.91	0.8649	

### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

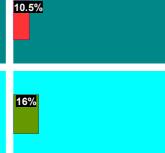


### Factors responsible for Prostate cancer









### Psoriasis

Psoriasis is a chronic, non-contagious autoimmune disease which affects the skin and joints. It commonly causes red scaly patches to appear on the skin. Psoriasis affects both sexes equally and can occur at any age, although it most commonly appears for the first time between the ages of 15 and 25 years. The prevalence of psoriasis in Western populations is estimated to be around 2 to 3%.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 2.3 out of every 100 person. These results indicate your odds of developing this condition.

Individuals from the average population are estimated to develop this condition in **11.4 out of every 100** person. These results apply to an average sample of

Summary of your genetic results

european ancestry.

The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs1800629	TNF	GG	0.76	0.6889
rs4112788	intergenic	CT	0.94	0.48
rs1265159	intergenic	CC	0.28	0.5929

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.

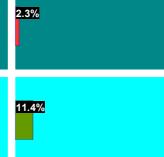


### Factors responsible for Psoriasis









# Rheumatoid arthritis

Rheumatoid arthritis is a chronic, systemic inflammatory disorder that attacks the joints and often progresses to destruction of the articular cartilage of the joints. About 1% of the world's population is afflicted by rheumatoid arthritis with women three times more likely than men to be affected. Onset is most frequent in 40 to 50 years, but no age is immune.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 0.6 out of every 100 person. These results indicate your odds of developing this condition.

Individuals from the average population are estimated to develop this condition in 1 out of every 100 person. These results apply to an average sample of european

# Summary of your genetic results

ancestry.

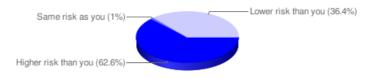
The average lifetime risk

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

Locus	Your genotype	Relative Risk	Genotype frequency	
TRAF1	AA	0.82	0.2704	
STAT4	GT	1.12	0.3542	
MHC	СТ	0.83	0.4992	
PTPN22	GG	0.82	0.7744	
MMEL1	GA	1	0.4422	
	TRAF1 STAT4 MHC PTPN22	TRAF1 AA STAT4 GT MHC CT PTPN22 GG	TRAF1         AA         0.82           STAT4         GT         1.12           MHC         CT         0.83           PTPN22         GG         0.82	TRAF1         AA         0.82         0.2704           STAT4         GT         1.12         0.3542           MHC         CT         0.83         0.4992           PTPN22         GG         0.82         0.7744

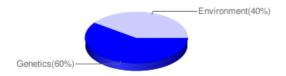
#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



### Factors responsible for Rheumatoid arthritis

Although your risks of developing this condition may seem high to you, keep in mind that your environment is a significantly responsible factor as well. This gives you a degree of control over your odds.







0.6%

1%

#### Skin cancer

Skin cancer is a malignant growth on the skin. There is over 1,100,000 cases reported in North America each year resulting in 1,100 deaths. The majority of these are called Basal Cell Carcinomas. Carcinomas are usually localized growths caused by excessive cumulative exposure to the sun and do not tend to spread.

#### Understand your risks medium

Your genetic predisposition towards this disease is medium. Note that for your risk to be considered medium, this requirement must be met: Your risk of developing the condition is higher than 7% (and your risk does not qualify as high).

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 31.2 out of every 100 person. These results indicate your odds of developing this condition.

#### The average lifetime risk Individuals from the average population are estimated to develop this condition in 35 out of every 100 person. These results apply to an average sample of european ancestry.

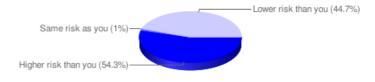
#### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs7538876	PADI6	GG	0.81	0.3364
rs401681	CLPTM1	TC	1.03	0.4902
rs801114	intergenic	GT	1.07	0.4488

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Skin cancer

Although your risks of developing this condition may seem high to you, keep in mind that your environment is a significantly responsible factor as well. This gives you a degree of control over your odds.

Environment(60%) Genetics(40%)





35%

# Type 1 diabetes

Type 1 diabetes is an autoimmune disease that results in destruction of insulin-producing beta cells of the pancreas. Type 1 is lethal unless treated with exogenous insulin. Injection is the traditional and still most common method for administering insulin.

#### Understand your risks low

Your genetic predisposition towards this disease is low. Note that for your risk to be considered low, this requirement must be met: All conditions with a lifetime risk lower than 7% are included, except for those that are already considered of high or medium risk.

Your estimated lifetime risk Individuals with your genetic variants are estimated to develop this condition in 1.3 out of every 100 person. These results indicate your odds of developing this condition.

Individuals from the average population are estimated to develop this condition in 1

out of every 100 person. These results apply to an average sample of european ancestry.

### Summary of your genetic results

The average lifetime risk

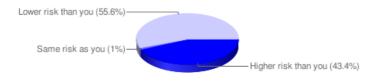
To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

1%

Marker	Locus	Your genotype	Relative Risk	Genotype frequency
rs7574865	STAT4	GT	1.2	0.3542
rs3087243	CTLA4	GG	1.1	0.2916
rs725613	CLEC16A	CA	0.97	0.4352

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



### Factors responsible for Type 1 diabetes







# Type 2 diabetes

Type 2 diabetes is a disorder that is characterized by high blood glucose in the context of insulin resistance and relative insulin deficiency. There are an estimated 26.6 million people in the United States living with diabetes (7.8% of the population). Of these, only 18.9 million have received a diagnosis - 90% of which fall in to the type 2 category.

#### Understand your risks medium

Your genetic predisposition towards this disease is medium. Note that for your risk to be considered medium, this requirement must be met: Your risk of developing the condition is higher than 7% (and your risk does not qualify as high).

condition.

Individuals with your genetic variants are estimated to develop this condition in 21.4 out of every 100 person. These results indicate your odds of developing this

The average lifetime risk

Your estimated lifetime risk

Individuals from the average population are estimated to develop this condition in 24 out of every 100 person. These results apply to an average sample of european ancestry.

### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition

Locus	Your genotype	Relative Risk	Genotype frequency
SLC30A8	CC	1.07	0.5776
CDKAL1	AA	0.93	0.4356
intergenic	ТТ	1.01	0.64
TCF7L2	СТ	1.17	0.4032
IGF2BP2	TT	1.09	0.09
HHEX	AG	0.8	0.4872
intergenic	CA	0.87	0.2112
	SLC30A8 CDKAL1 intergenic TCF7L2 IGF2BP2 HHEX	SLC30A8CCCDKAL1AAintergenicTTTCF7L2CTIGF2BP2TTHHEXAG	SLC30A8         CC         1.07           CDKAL1         AA         0.93           intergenic         TT         1.01           TCF7L2         CT         1.17           IGF2BP2         TT         1.09           HHEX         AG         0.8

Type 2 diabetes (cont'd)







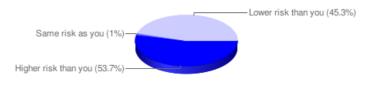
21.4%

24%

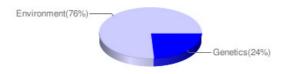
# Type 2 diabetes (cont'd)

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Type 2 diabetes



# Venous thromboembolism

Venous thromboembolism is the formation of a blood clot (thrombus) inside a blood vessel, obstructing the flow of blood through the circulatory system. It can result in deprivation of oxygen in the adjacent area, and/or infarction, a mode of cell death.

#### Understand your risks medium

Your genetic predisposition towards this disease is medium. Note that for your risk to be considered medium, this requirement must be met: Your risk of developing the condition is higher than 7% (and your risk does not qualify as high).

Your estimated lifetime risk<br/>Individuals with your genetic variants are estimated to develop this condition in 23.3<br/>out of every 100 person. These results indicate your odds of developing this<br/>condition.23.3%The average lifetime risk<br/>Individuals from the average population are estimated to develop this condition in 25<br/>out of every 100 person. These results apply to an average sample of european<br/>ancestry.25%

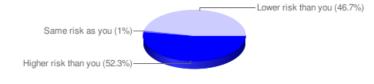
### Summary of your genetic results

To evaluate your risks, we looked at the following markers in your DNA. The column named "Relative risk" indicates the individual contribution of each marker to your lifetime risk. If the relative risk is higher than 1, then the genetic marker increases your risk of developing this condition.

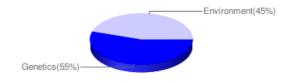
Marker	Locus	Your genotype	Relative Risk	Genotype frequency		
rs6025	F5	GG	0.93	0.9604		
Source · Example Data						

#### Your risks compared to a similar group of individuals

Here is another way of interpreting your results. The information that we use for this comparison comes in part from the international HapMap Project, the largest publicly available database of human genome variation, and in part from the Center for Disease Control and Prevention.



#### Factors responsible for Venous thromboembolism







# Glossary

#### Arteries

Arteries are blood vessels that carry blood away from the heart. All arteries, with the exception of the pulmonary and umbilical arteries, carry oxygenated blood. The circulatory system is extremely important for sustaining life. Its proper functioning is responsible for the delivery of oxygen and nutrients to all cells, as well as the removal of carbon dioxide and waste products, maintenance of optimum pH, and the mobility of the elements, proteins and cells of the immune system. In developed countries, the two leading causes of death, myocardial infarction and stroke each may directly result from an arterial system that has been slowly and progressively compromised by years of deterioration.

#### Autoimmunity

Autoimmunity is the failure of an organism to recognize its own constituent parts as *self*, which allows an immune response against its own cells and tissues. Any disease that results from such an aberrant immune response is termed an **autoimmune disease**.

#### Basal cell carcinoma

**Basal cell carcinoma** is the most common type of skin cancer. It rarely metastasizes or kills, but it is still considered malignant because it can cause significant destruction and disfigurement by invading surrounding tissues. Statistically, approximately 3 out of 10 Caucasians develop a basal cell cancer within their lifetime. In 80 percent of all cases, basal cell cancers are found on the head and neck. There appears to be an increase in the incidence of basal cell cancer of the trunk in recent years.

#### Cartilage

**Cartilage** is a type of dense connective tissue existing within many joints. It is composed of specialized cells that produce a large amount of extracellular matrix composed of collagen fibers, abundant ground substance rich in proteoglycan, and elastin fibers. Cartilage is classified in three types, **elastic cartilage**, **hyaline cartilage** and **fibrocartilage**, which differ in the relative amounts of these three main components.

#### Dementia

**Dementia** is a cognitive impairment. It may be static, the result of a unique global brain injury or progressive, resulting in long-term decline in cognitive function due to damage or disease in the body beyond what might be expected from normal aging. Although dementia is far more common in the geriatric population, it may occur in any stage of adulthood. This age cutoff is defining, as similar sets of symptoms due to a dysfunction, are given different names in populations younger than adult.

#### Disease

An abnormal condition of an organism that impairs bodily functions, associated with specific symptoms and signs. It may be caused by external factors, such as invading organisms, or it may be caused by internal dysfunctions, such as autoimmune diseases. In human beings, "disease" is often used more broadly to refer to any condition that causes extreme pain, dysfunction, distress, social problems, and/or death to the person afflicted, or similar problems for those in contact with the person. In this broader sense, it sometimes includes injuries, disabilities, disorders, syndromes, infections, isolated symptoms, deviant behaviors, and atypical variations of structure and function, while in other contexts and for other purposes these may be considered distinguishable categories.

#### **Disease incidence**

Incidence is a measure of the risk of developing some new condition within a specified period of time.

#### Deoxyribonucleic acid

A nucleic acid that contains the genetic instructions used in the development and functioning of all known living organisms and some viruses. The main role of DNA molecules is the long-term storage of information. DNA is often compared to a set of blueprints or a recipe, or a code, since it contains the instructions needed to construct other components of cells, such as proteins and RNA molecules. The DNA segments that carry this genetic information are called genes, but other DNA sequences have structural purposes, or are involved in regulating the use of this genetic information.

#### **DNA** marker

A gene or DNA sequence with a known location on a chromosome and associated with a particular gene or trait. It can be described as a variation, which may arise due to mutation or alteration in the genomic loci, that can be observed. A genetic marker may be a short DNA sequence, such as a sequence surrounding a single base-pair change (single nucleotide polymorphism, SNP), or a long one, like minisatellites.

#### Epidemiology

**Epidemiology** is the study of factors affecting the health and illness of populations, and serves as the foundation and logic of interventions made in the interest of public health and preventive medicine. It is considered a cornerstone methodology of public health research, and is highly regarded in evidence-based medicine for identifying risk factors for disease and determining optimal treatment approaches to clinical practice.

#### Esophagus

The **esophagus**, sometimes known as the **gullet**, is an organ in vertebrates which consists of a muscular tube through which food passes from the pharynx to the stomach.

#### Exogenous

In biology, an exogenous factor is any material that is present and active in an individual organism or living cell but that originated outside of that organism, as opposed to an endogenous factor.

#### Gene

A gene is the basic unit of heredity in a living organism. All living things contain genes. Genes hold the information to build and maintain their cells and pass genetic traits to offspring.

#### **Genetic Counselling**

The process by which patients or relatives, at risk of an inherited disorder, are advised of the consequences and nature of the disorder, the probability of developing or transmitting it, and the options open to them in management and family planning in order to prevent, avoid or ameliorate it. This complex process can be seen from diagnostic (the actual estimation of risk) and supportive aspects.

#### Genotype

The **genotype** is the genetic constitution of a cell, an organism, or an individual (i.e. the specific allele makeup of the individual) usually with reference to a specific character under consideration. For instance, the human albino gene has two allele forms, dominant A and recessive a, and there are three possible genotypes- AA (homozygous dominant), Aa (heterozygous), and aa (homozygous recessive).

#### HapMap project

The **International HapMap Project** is an organization whose goal is to develop a haplotype map of the human genome (**HapMap**), which will describe the common patterns of human genetic variation. The HapMap is expected to be a key resource for researchers to find genetic variants affecting health, disease and responses to drugs and environmental factors. The information produced by the project is made freely available to researchers around the world.

#### Hemorrhage

**Bleeding**, technically known as **hemorrhaging** is the loss of blood from the circulatory system. Bleeding can occur internally, where blood leaks from blood vessels inside the body or externally, either through a natural opening such as the vagina, mouth, nose, or anus, or through a break in the skin. The complete loss of blood is referred to as exsanguination, and desanguination is a massive blood loss. Typically, a healthy person can endure a loss of 10-15% of the total blood volume without serious medical difficulties, and blood donation typically takes 8-10% of the donor's blood volume.

#### Human genome

The **human genome** is the genome of *Homo sapiens*, which is stored on 23 chromosome pairs. Twenty-two of these are autosomal chromosome pairs, while the remaining pair is sex-determining. The haploid human genome occupies a total of just over 3 billion DNA base pairs. The Human Genome Project (HGP) produced a reference sequence, which is used worldwide in biomedical sciences.

#### **Hyperacusis**

**Hyperacusis** is a health condition characterized by an over-sensitivity to certain frequency ranges of sound (a collapsed tolerance to normal environmental sound). A person with severe hyperacusis has difficulty tolerating everyday sounds, some of which may seem unpleasantly loud to that person but not to others. The most common sound to appear unpleasant in hyperacusis is people eating or clicking their fingers.

#### Infarction

In medicine, an **infarction** is the process of tissue death (necrosis) caused by blockage of the tissue's blood supply. The supplying artery may be blocked by an obstruction (e.g. an embolus, thrombus, or atherosclerotic plaque), may be mechanically compressed (e.g. tumor, volvulus, or hernia), ruptured by trauma (e.g. atherosclerosis or vasculitides), or vasoconstricted (e.g. cocaine vasoconstriction leading to myocardial infarction).

#### Inflammation

**Inflammation** is the complex biological response of vascular tissues to harmful stimuli, such as pathogens, damaged cells, or irritants. It is a protective attempt by the organism to remove the injurious stimuli as well as initiate the healing process for the tissue. Inflammation is not a synonym for infection.

#### Insulin

**Insulin** is a hormone that has extensive effects on metabolism and other body functions, such as vascular compliance. Insulin causes cells in the liver, muscle, and fat tissue to take up glucose from the blood, storing it as glycogen in the liver and muscle, and stopping use of fat as an energy source. When insulin is absent (or low), glucose is not taken up by body cells, and the body begins to use fat as an energy source, for example, by transfer of lipids from adipose tissue to the liver for mobilization as an energy source. As its level is a central metabolic control mechanism, its status is also used as a control signal to other body systems (such as amino acid uptake by body cells). It has several other anabolic effects throughout the body. When control of insulin levels fails, diabetes mellitus results.

#### Malignant growth

**Cancer** (medical term: malignant neoplasm, **malignant growth**) is a class of diseases in which a group of cells display *uncontrolled growth* (division beyond the normal limits), *invasion* (intrusion on and destruction of adjacent tissues), and sometimes *metastasis* (spread to other locations in the body via lymph or blood). These three malignant properties of cancers differentiate them from benign tumors, which are self-limited, and do not invade or metastasize. Most cancers form a tumor but some, like leukemia, do not. The branch of medicine concerned with the study, diagnosis, treatment, and prevention of cancer is oncology.

#### **Metastasis**

**Metastatic disease**, sometimes abbreviated **mets**, is the spread of a disease from one organ or part to another non-adjacent organ or part. Only malignant tumor cells and infections have the established capacity to **metastasize**; however, this is recently reconsidered by new research.

#### Molecular test

A laboratory test based on the analysis of DNA or large molecules.

#### **Mutation**

In biology, **mutations** are changes to the nucleotide sequence of the genetic material of an organism. Mutations can be caused by copying errors in the genetic material during cell division, by exposure to ultraviolet or ionizing radiation, chemical mutagens, viruses, or can be induced by the organism itself, by cellular processes such as hypermutation.

#### Neurology

**Neurology** is a medical specialty dealing with disorders of the nervous system. Specifically, it deals with the diagnosis and treatment of all categories of disease involving the central, peripheral, and autonomic nervous systems, including their coverings, blood vessels, and all effector tissue, such as muscle. The corresponding surgical specialty is neurosurgery. A **neurologist** is a physician (not a surgeon) who specializes in neurology, and is trained to investigate, or diagnose and treat, neurological disorders.

#### Pancreas

The **pancreas** is a gland organ in the digestive and endocrine system of vertebrates. It is both an endocrine gland producing several important hormones, including insulin, glucagon, and somatostatin, as well as an exocrine gland, secreting pancreatic juice containing digestive enzymes that pass to the small intestine. These enzymes help in the further breakdown of the carbohydrates, protein, and fat in the chyme.

#### Photophobia

Photophobia is a symptom of excessive sensitivity to light and the aversion to sunlight or well-lit places. In ordinary medical terms photophobia is not a morbid fear, but an experience of discomfort or pain to the eyes due to light exposure.

#### Predisposition

A genetic predisposition is a genetic effect which influences the phenotype of an organism but which can be modified by the environmental conditions. Genetic testing is able to identify individuals who are genetically predisposed to certain health problems.

#### Revascularization

**Revascularization** is "a surgical procedure for the provision of a new, additional, or augmented blood supply to a body part or organ." The term derives from the prefix re-, in this case meaning "restoration" and vasculature, which refers to the circulatory structures of an organ. Revascularization involves a thorough analysis and diagnosis and treatment of the existing diseased vasculature of the affected organ, and can be aided by the use of different imaging modalities such as magnetic resonance imaging, PET scan, CT scan, and X ray fluoroscopy.

#### SNP (Single Nucleotide Polymorphism)

A DNA sequence variation occurring when a single nucleotide - A, T, C, or G - in the genome (or other shared sequence) differs between members of a species (or between paired chromosomes in an individual). For example, two sequenced DNA fragments from different individuals, AAGC**C**TA to AAGC**T**TA, contain a difference in a single nucleotide. In this case we say that there are two *alleles* : C and T. Almost all common SNPs have only two alleles.

#### Subchondral bone

The **epiphysis** is the rounded end of a long bone, at its joint with adjacent bone(s). Between the epiphysis and diaphysis (the long midsection of the long bone) lies the metaphysis, including the epiphyseal plate (growth plate). At the joint, the epiphysis is covered with articular cartilage; below that covering is a zone similar to the epiphyseal plate, known as **subchondral bone**.

#### Syndrome

In medicine and psychology, the term **syndrome** refers to the association of several clinically recognizable features, signs (observed by a physician), symptoms (reported by the patient), phenomena or characteristics that often occur together, so that the presence of one feature alerts the physician to the presence of the others. In recent decades the term has been used outside of medicine to refer to a combination of phenomena seen in association.

#### Thrombus

A **thrombus**, or **blood clot**, is the final product of the blood coagulation step in hemostasis. It is achieved via the aggregation of platelets that form a platelet plug, and the activation of the humoral coagulation system (i.e. clotting factors). A thrombus is normal in cases of injury, but pathologic in instances of thrombosis.

#### Thyroid

The **thyroid** is one of the largest endocrine glands in the body. This gland is found in the neck inferior to (below) the thyroid cartilage (also known as the Adam's apple in men) and at approximately the same level as the cricoid cartilage. The thyroid controls how quickly the body burns energy, makes proteins, and how sensitive the body should be to other hormones.

#### Variation (genetics)

**Genetic diversity** is a level of biodiversity that refers to the total number of genetic characteristics in the genetic makeup of a species. It is distinguished from genetic variability, which describes the tendency of genetic characteristics to vary.

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