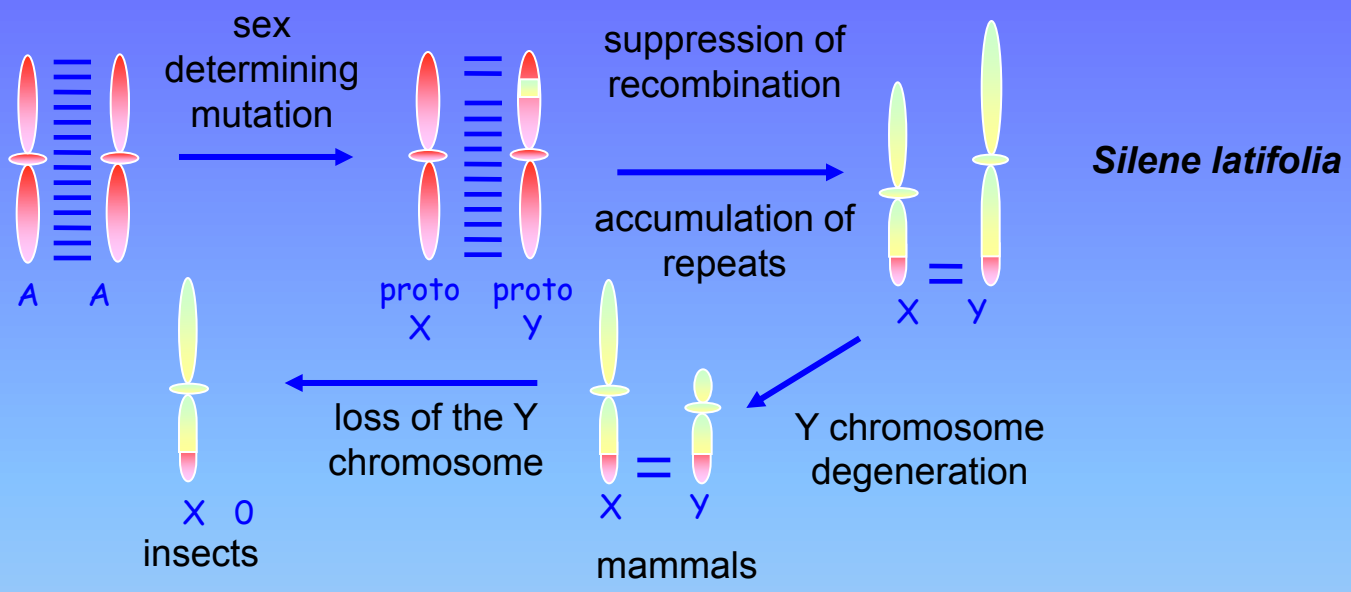
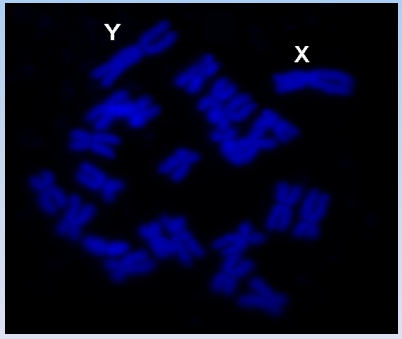
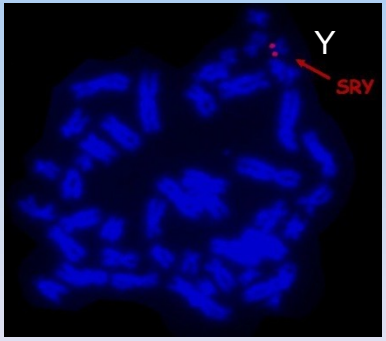
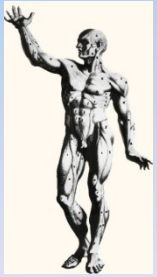
Osud skrytý v DNA aneb proč jsme ještě nevymřeli





vs.





Silene latifolia (10-20 MYA)

Homo sapiens (240-320 MYA)

Sex chromosomes - history

1891 Hermann Henking – spermatogenesis in Pyrrhocoris apterus (firebug)

Henking saw that some sperm cells had 12 chromosomes, while others had only 11.

Henking noticed that the mysterious twelfth chromosome looked different from all the others.

He thus named this chromosome the "X element," to represent its unknown nature.

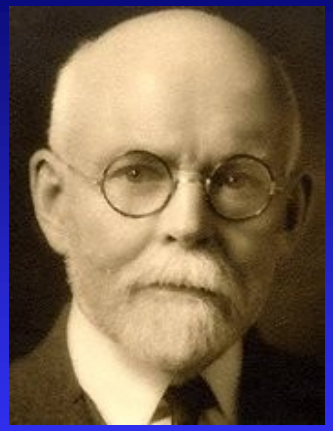
1902 Clarence Erwin **McClung -THE ACCESSORY CHROMOSOME**

Nettie Stevens

Tenebrio molitor, Coleoptera

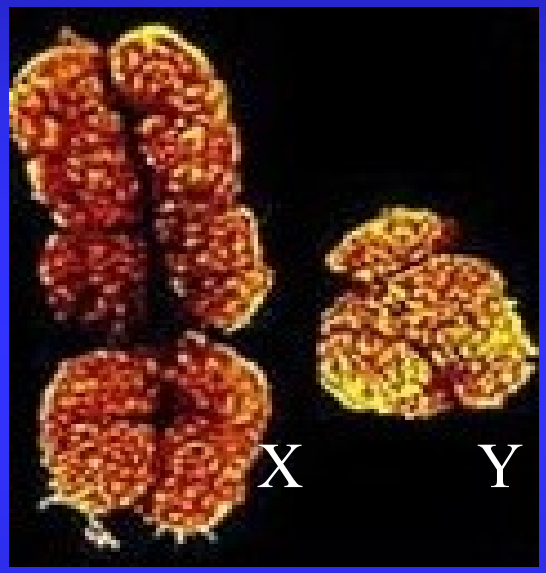
(F = AAXX, M = AAXY)

mealworm



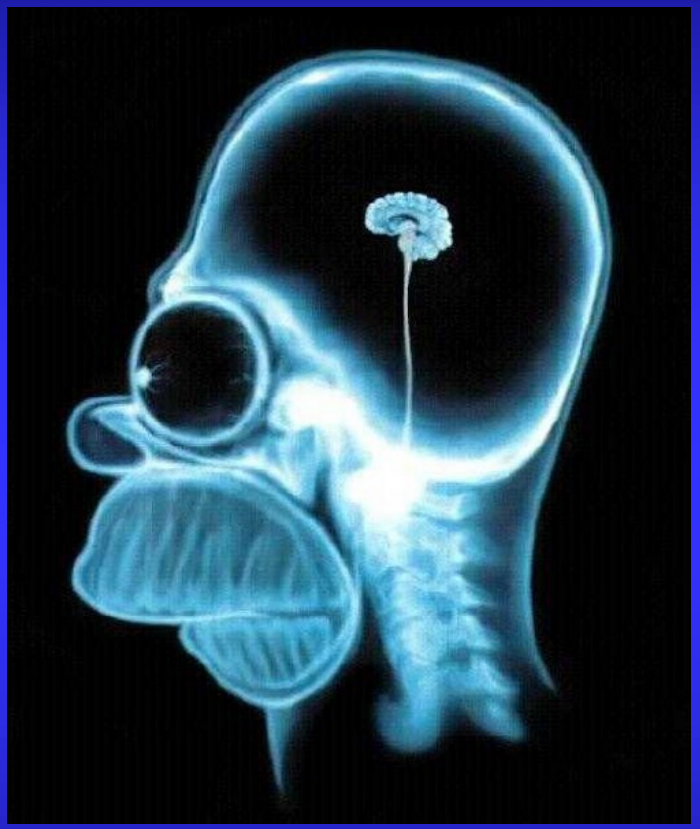
Edmund Wilson

Past, present and future of sex chromosomes



at the current rate of degradation, human Y chromosome will likely lose its last 45 genes in just 10 million years

Accumulation of „brain specific“ genes on the X chromosome



The presence of genes on the X chromosome that get expressed in the brain may allow more rapid selection for favorable genetic variations which enhance cognitive function

Human X chromosome and cancer genes

- Human X chromosome is almost depleted of cancer genes due to selection

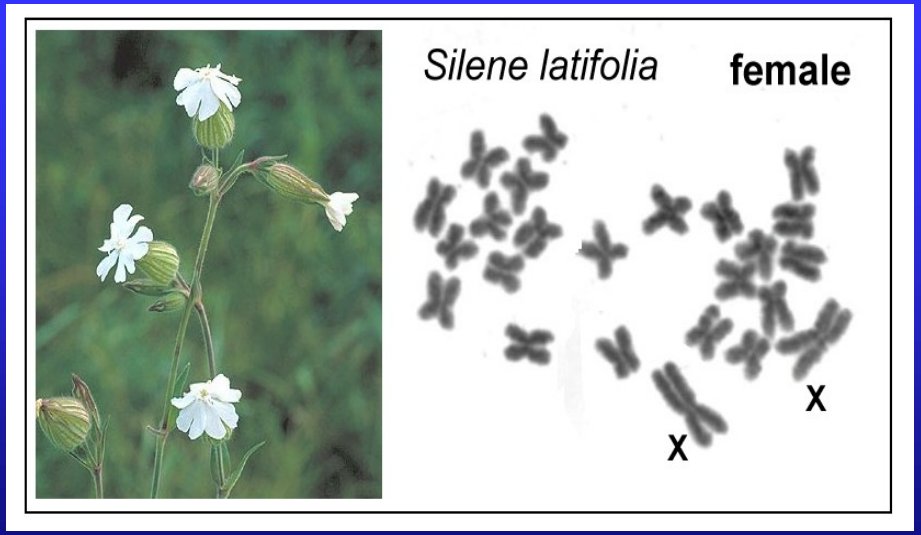
The day after Y chromosome disappears

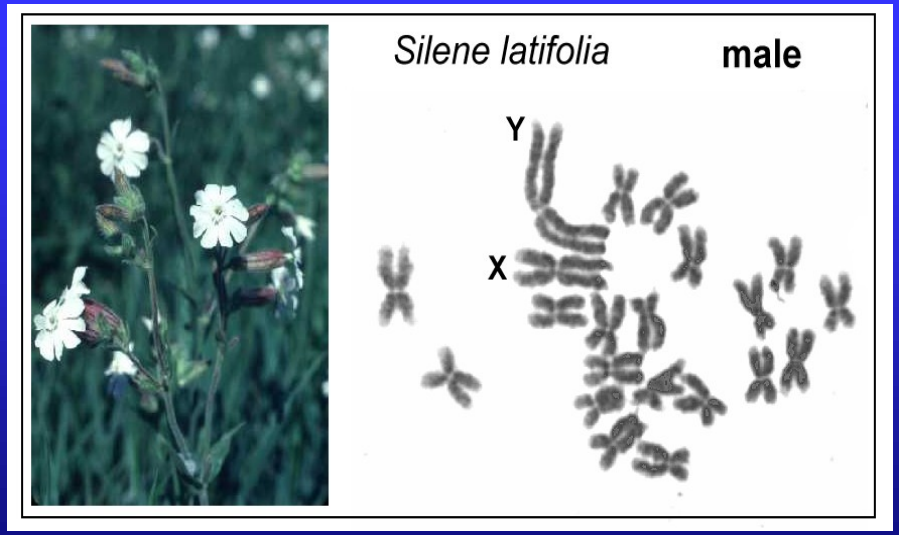
SRY has been lost in some rare rodents
and replaced by a new sex determining gene

So as the human Y runs out of options, new sex determining genes may evolve, leading to evolution of different hominid species.

Sex chromosomes in *S. latifolia*

Gregor Mendel





Gregor Johann Mendel

GREGOR MENDEL'S
LETTERS TO CARL NÄGELI

Carl Wilhelm von Nägeli

Finally, let me report on a curiosity in the numerical ratios in which the male and the female plants of the hybrid *Lychnis diurna*+*L. vespertina occur.* I fertilized three flowers of *L. diurna* and planted the seeds of each capsule separately. The

Brünn, 27 September, 1870

23andMe

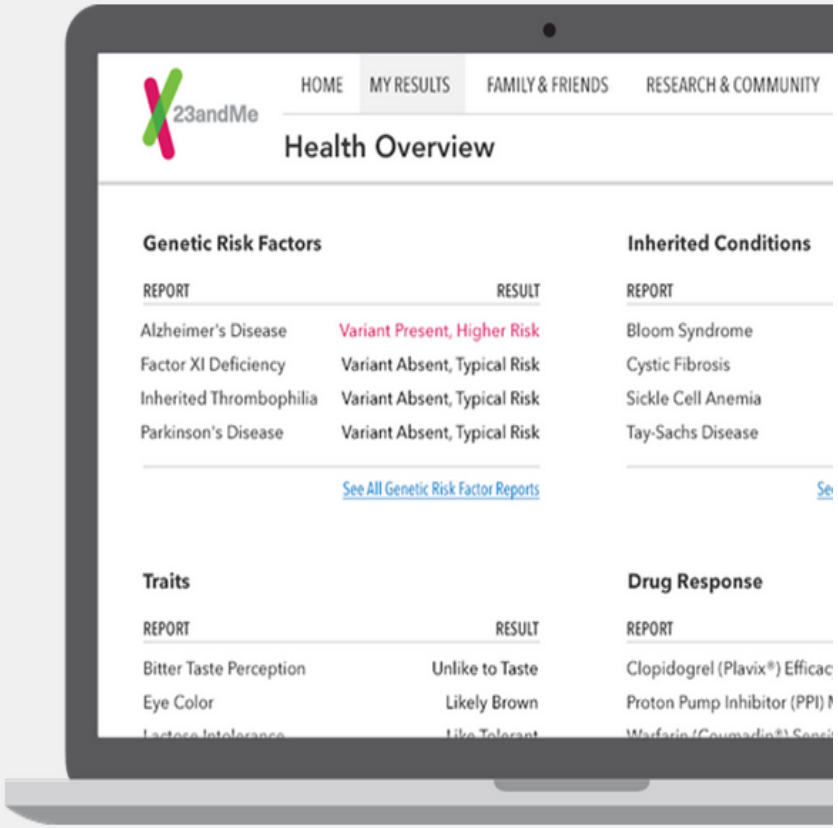
welcome | health | ancestry | research | how it works | buy | help

Learn how your DNA may affect your health.

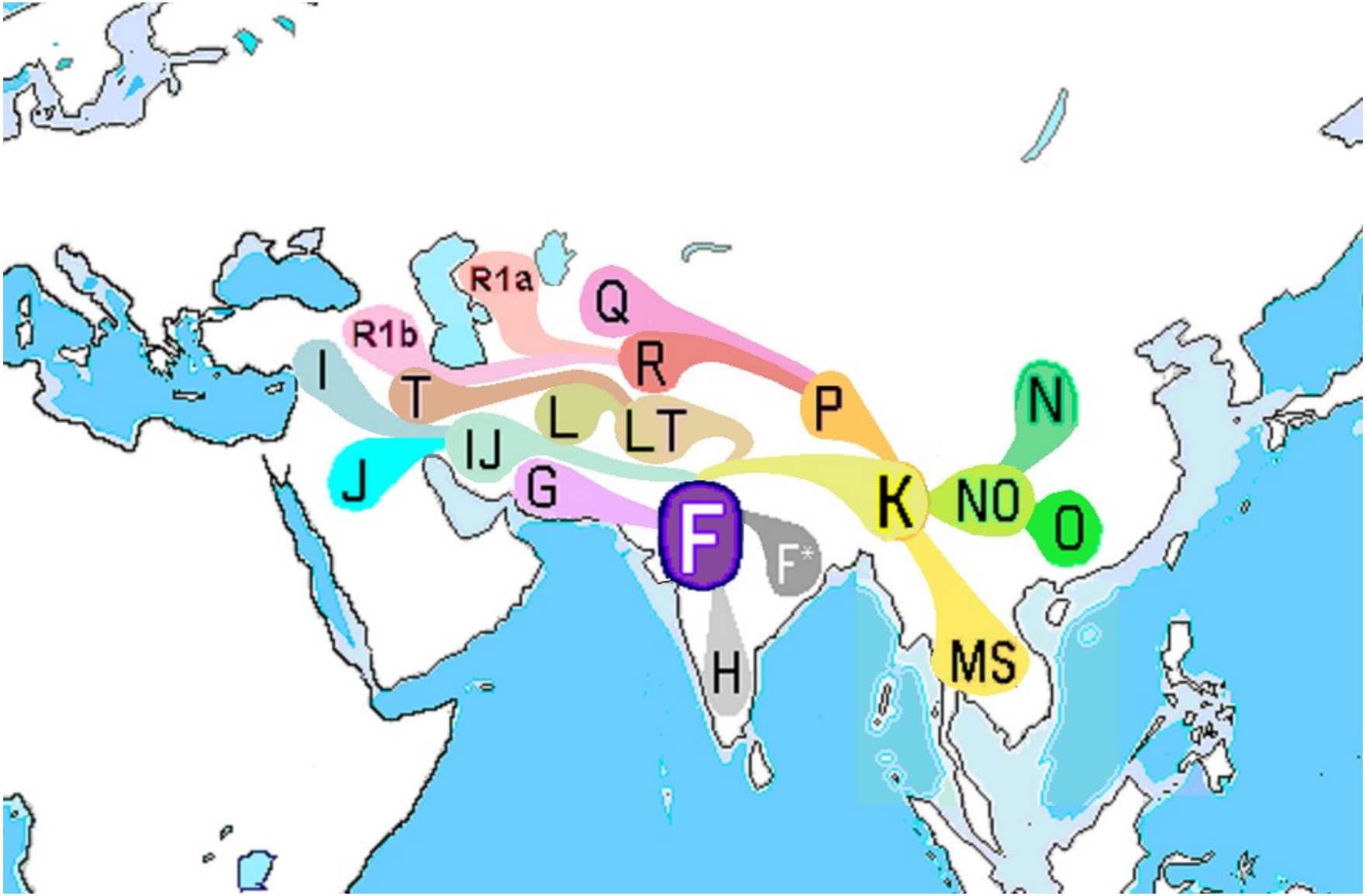
Our genes are a part of who we are, so naturally they impact our health. By knowing more about your DNA, you may be able to take steps towards living a healthier life.

Keep in mind that many conditions and traits are influenced by multiple factors. Our reports are intended for informational purposes only and do not diagnose disease or illness.

- **Plan for the future.**
  Learn if you are a carrier for certain inherited conditions, so you and your family can be prepared.
- **Stay one step ahead.**
  Find out if you have certain genetic risk factors, so you can make better lifestyle choices and appropriately monitor your health.
- **Engage in your health care.**
  Understand how your DNA may affect your health and response to




23andMe
DNA Genetek
BIO-SPECIMEN BAG


R1a
Q
R1b
R
I
T
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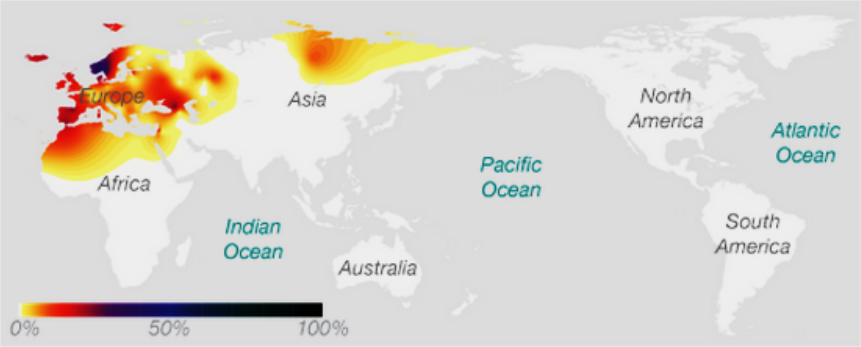
LT
P
N
IJ
J
G
F
F*
K
NO
O
H
MS

HOME | MY RESULTS | FAMILY & FRIENDS | RESEARCH & COMMUNITY

MATERNAL LINE: H1

Overview | History | Haplogroup Tree | Community

Locations of haplogroup H1 before the widespread migrations of the past few hundred years.



Haplogroup H1 is widespread in Europe, especially the western part of the continent. It originated about 13,000 years ago, not long after the Ice Age ended.

Maternal haplogroups are families of mitochondrial DNA types that all trace back to a single mutation at a specific place and time. By looking at the geographic distribution of mtDNA types, we learn how our ancient female ancestors migrated throughout the world.

Haplogroup: H1, a subgroup of H
Age: 13,000 years
Region: Europe, Near East, Central Asia, Northwestern Africa
Example Populations: Spanish,Berbers,Lebanese
Highlight: H1 appears to have been common in Doggerland, an ancient land now flooded by the North Sea.

PATERNAL LINE: I1*

I1* is a subgroup of I1

Locations of haplogroup I1 before the widespread migrations of the past few hundred years.

Haplogroup I1 can be found at levels of 10% and higher in many parts of Europe, due to its expansion with men who migrated northward after the end of the Ice Age about 12,000 years ago. It reaches its highest levels in Denmark and the southern parts of Sweden and Norway.

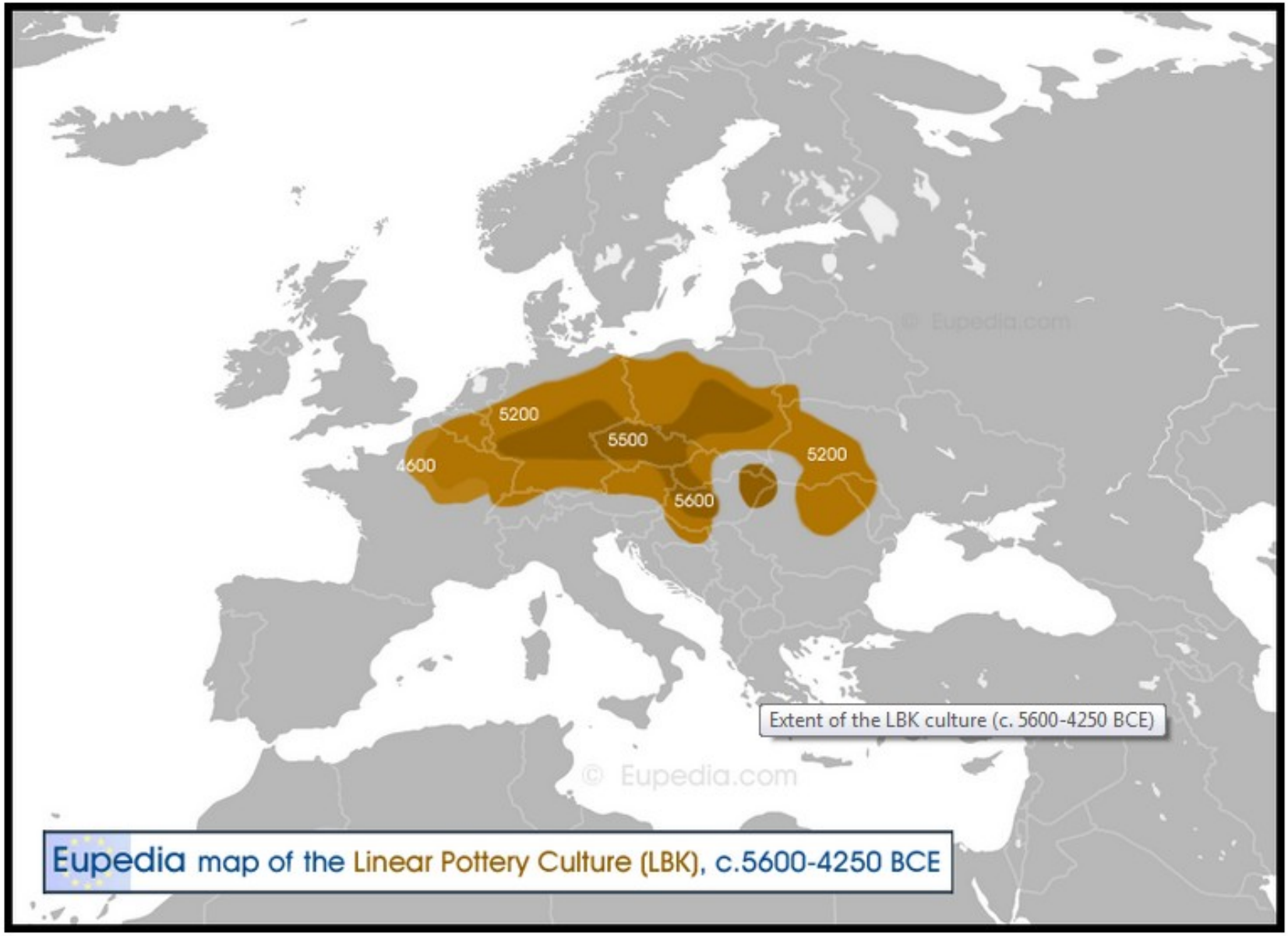
Paternal haplogroups are families of Y chromosomes that all trace back to a single mutation at a specific place and time. By looking at the geographic distribution of these related lineages, we learn how our ancient male ancestors migrated throughout the world.

Haplogroup: I1, a subgroup of I
Age: 28,000 years
Region: Northern Europe
Example Populations: Finns, Norwegians, Swedes
Highlight: Haplogroup I1 reaches highest frequencies in Scandinavia.

Haplogroups of You and Your Connections

I1*	Roman Hobza

Haplogroups of Example Profiles


5200
5500
4600
5200
5600
Extent of the LBK culture (c. 5600-4250 BCE)
© Eupedia.com
© Eupedia.com
Eupedia map of the Linear Pottery Culture (LBK), c.5600-4250 BCE

SHOW RESULTS FOR Roman Hobza ▾

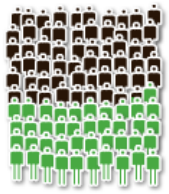
SEE NEW AND RECENTLY UPDATED REPORTS »

These reports provide information about your possible risk for developing certain health conditions based on genetics. Environmental and lifestyle factors also often play a large role in your risk for developing these conditions.

Elevated Risk

NAME	CONFIDENCE	YOUR RISK	AVG. RISK	COMPARED TO AVERAGE
Venous Thromboembolism	★★★★	41.8%	12.3%	3.39x
Gout	★★★★	35.7%	22.8%	1.57x
Melanoma	★★★★	4.0%	2.9%	1.38x
Restless Legs Syndrome	★★★★	2.5%	2.0%	1.25x
Exfoliation Glaucoma	★★★★	2.2%	0.7%	2.90x
Esophageal Squamous Cell Carcinoma (ESCC)	★★★★	0.43%	0.36%	1.21x
Stomach Cancer (Gastric Cardia Adenocarcinoma)	★★★★	0.28%	0.23%	1.22x
Primary Biliary Cirrhosis	★★★★	0.11%	0.08%	1.43x
Scleroderma (Limited Cutaneous Type)	★★★★	0.08%	0.07%	1.24x

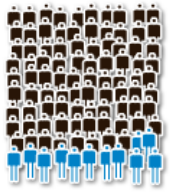
Show information for Roman Hobza ▾ assuming European ▾ ethnicity and an age range of 0-79 ▾



Roman Hobza

41.8 out of 100

men of European ethnicity who share Roman Hobza's genotype will develop Venous Thromboembolism between the ages of 0 and 79.



Average

12.3 out of 100

men of European ethnicity will develop Venous Thromboembolism between the ages of 0 and 79.

What does the Odds Calculator show me?

Use the ethnicity and age range selectors above to see the estimated incidence of Venous Thromboembolism due to genetics for men with **Roman Hobza**'s genotype. The 23andMe Odds Calculator assumes that a person is free of the condition at the lower age in the range. You can use the name selector above to see the estimated incidence of Venous Thromboembolism for the genotypes of other people in your account.

The 23andMe Odds Calculator only takes into account effects of markers with known associations that are also on our genotyping chip. Keep in mind that aside from genetics, environment and lifestyle may also contribute to one's risk for Venous Thromboembolism.

55 %
Attributable to Genetics

Understanding Your Results

The heritability of venous thromboembolism is estimated to be 55%. This means that genetics (including unknown factors and known ones such as the SNPs we describe here) and environment play nearly equal roles in this condition. There are a number of environmental factors of various strengths that contribute to venous thromboembolism. Strong risk factors include hip or leg fractures, hip or knee replacement, major surgery or trauma, and spinal cord injury or surgery. Moderate risk factors include arthroscopic knee surgery, having central venous lines, congestive heart or respiratory failure, hormone replacement or oral contraceptive use, cancer, pregnancy, paralytic stroke, previous venous thromboembolism, and thrombophilia. Weak risk factors include bed rest for more than three days, immobility due to sitting (such as a long car or plane trip), specific types of chemotherapy, increasing age, laparoscopic surgery, obesity, and varicose veins. (sources)

What You Can Do

Assuming the ethnicity setting above is correct, your test results indicate you are at increased risk for venous thromboembolism based on genetics. Note that family history and non-genetic factors can also influence your risk for venous thromboembolism. Below are some steps you can take to reduce your risk.

Gene or region: F5
SNP: rs6025

	SNP used	Genotype	Adjusted Odds Ratio*
Roman Hobza	rs6025	CT	European: 4.69
** Odds ratios are reported for all available ethnicities.*			

Factor V is the last clotting factor in the pathway before the activation step that turns prothrombin into thrombin. Clotting is usually kept from spiraling out of control by a feedback loop, similar to the way a thermostat operates. Once enough thrombin has been activated, it binds to a protein called "protein C." Protein C then inactivates factor V, thus cutting off activation of prothrombin into thrombin.

The SNP in the F5 gene causes a change in the protein sequence of factor V that prevents protein C from inactivating it. Since this version of factor V can still participate in the activation of thrombin, a situation results in which thrombin can be turned on but cannot be turned off. Once the clotting cascade is set off (whether appropriately or not), the riskier version of the SNP makes it more difficult to shut it off.

The riskiness of the T version of this SNP is further increased for women who also take hormonal birth control.

(The riskier version of this gene is also sometimes called Factor V Leiden, after the city in the Netherlands where this SNP and its effects on factor V's role in clotting were first discovered.)

The studies whose data we report as applicable to those of "European" ancestry confirmed the association between this SNP and VTE in samples from the Netherlands, Sweden, the United Kingdom, Brazil, Italy, and France.

African and Asian populations appear to have only one version of the SNP, meaning that association studies are very difficult to perform.

Citations

Rosendaal et al. (1995) . "High risk of thrombosis in patients homozygous for factor V Leiden (activated protein C resistance)." *Blood* 85(6):1504-8.

Smith et al. (2007) . "Association of genetic variations with nonfatal venous thrombosis in postmenopausal women." *JAMA* 297(5):489-98.

Emmerich et al. (2001) . "Combined effect of factor V Leiden and prothrombin 20210A on the risk of venous thromboembolism--pooled analysis of 8 case-control studies including 2310 cases and 3204 controls. Study Group for Pooled-Analysis in Venous Thromboembolism." *Thromb Haemost* 86(3):809-16.

Bertina et al. (1994) . "Mutation in blood coagulation factor V associated with resistance to activated protein C." *Nature* 369(6475):64-7.

Lane et al. (2000) . "Role of hemostatic gene polymorphisms in venous and arterial thrombotic disease." *Blood* 95(5):1517-32.

Gene or region: F2
SNP: i3002432

Decreased Risk

NAME	CONFIDENCE	YOUR RISK	AVG. RISK	COMPARED TO AVERAGE
Type 2 Diabetes	★★★★	17.7%	25.7%	0.69x
Alzheimer's Disease	★★★★	4.3%	7.2%	0.60x
Rheumatoid Arthritis	★★★★	1.6%	2.4%	0.68x
Parkinson's Disease	★★★★	1.2%	1.6%	0.73x
Age-related Macular Degeneration	★★★★	0.92%	6.55%	0.14x
Crohn's Disease	★★★★	0.31%	0.53%	0.58x
Multiple Sclerosis	★★★★	0.24%	0.34%	0.69x
Type 1 Diabetes	★★★★	0.12%	1.02%	0.12x
Celiac Disease	★★★★	0.05%	0.12%	0.44x

BRCA Cancer Mutations (Selected)	★★★★	Variant Absent
Beta Thalassemia	★★★★	Variant Absent
Bloom's Syndrome	★★★★	Variant Absent
Canavan Disease	★★★★	Variant Absent
Congenital Disorder of Glycosylation Type 1a (PMM2-CDG)	★★★★	Variant Absent
Connexin 26-Related Sensorineural Hearing Loss	★★★★	Variant Absent
Cystic Fibrosis	★★★★	Variant Absent
D-Bifunctional Protein Deficiency	★★★★	Variant Absent
DPD Deficiency	★★★★	Variant Absent
Dihydrolipoamide Dehydrogenase Deficiency	★★★★	Variant Absent
Factor XI Deficiency	★★★★	Variant Absent
Familial Dysautonomia	★★★★	Variant Absent
Familial Hypercholesterolemia Type B	★★★★	Variant Absent
Familial Hyperinsulinism (ABCC8-related)	★★★★	Variant Absent
Familial Mediterranean Fever	★★★★	Variant Absent
Fanconi Anemia (FANCC-related)	★★★★	Variant Absent
G6PD Deficiency	★★★★	Variant Absent

Reading Ability	★★★	Typical Nonword Reading Score
Response to Diet	★★★	See Report
Response to Exercise	★★★	See Report
Sex Hormone Regulation	★★★	See Report
Sweet Taste Preference	★★★	See Report
Tooth Development	★★★	See Report
Tuberculosis Susceptibility	★★★	See Report
Breast Morphology	★★★	Not Applicable
Menarche	★★★	Not Applicable
Menopause	★★★	Not Applicable
Eating Behavior	★★	Greater tendency to overeat
HIV Progression	★★	See Report
Hair Thickness	★★	Typical, if European or African
Longevity	★★	See Report
Measures of Intelligence	★★	Lower Non-Verbal IQ
Memory	★★	Typical Episodic Memory
Odor Detection	★★	Typical Sensitivity to Sweaty Odor
Pain Sensitivity	★★	Increased
Avoidance of Errors	★	See Report

Centrum strukturní a funkční genomiky

Oddělení vývojové genetiky rostlin

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Vašek Bačovský Roman Gogela Vojta Hudzieczek

Roman Hobza Bohouš Janoušek Zdeňek Kubat

Wojtek Jesionek Markéta Palovská Janka Puterová

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