

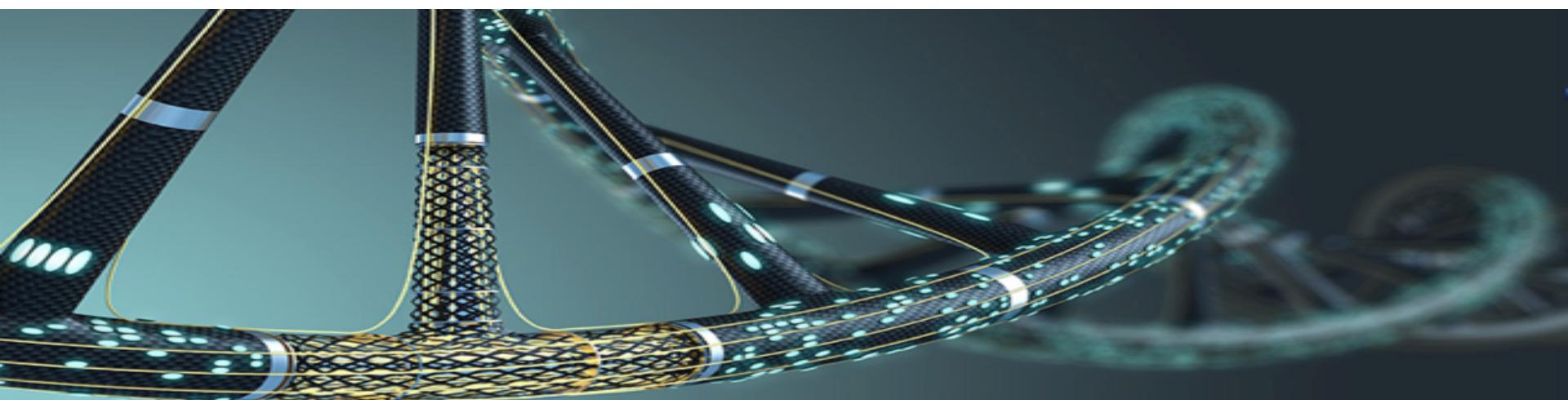


Майор по биоинформатике

Семестр 2

Лекция 5

Мария Попцова

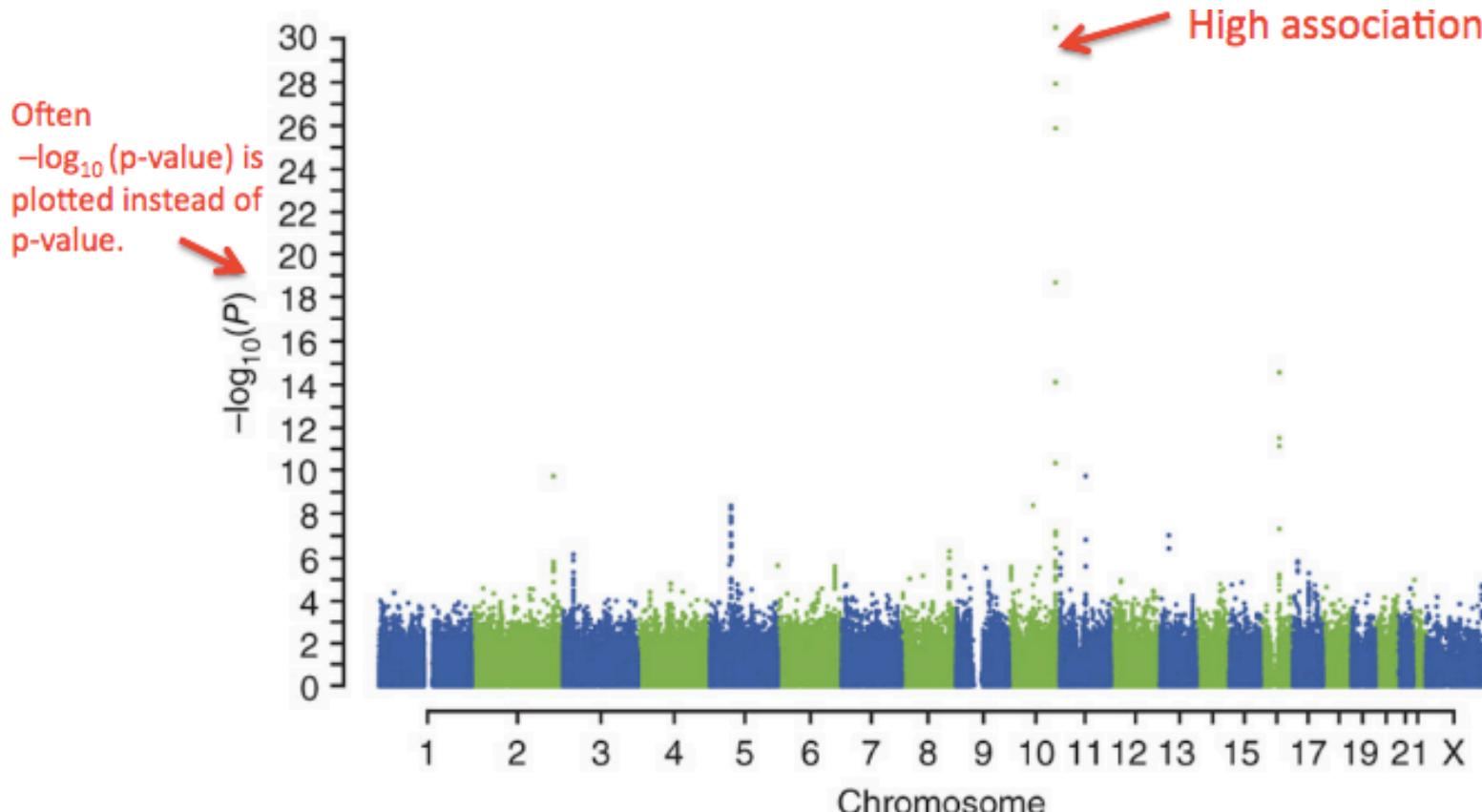


SNP association with a disease

- Мы умеем построить таблицу сопряженности (contingency table) и проверить верность гипотезы о наличии связи между снипом и эффектом методом хи-квадрат
- Мы принимаем или отвергаем нулевую гипотезу (отсутствие связи) с некоторым p-value
- Если $p\text{-value} < 0.05$, то вероятность нулевой гипотезы очень мала, и мы принимаем альтернативную гипотезу о наличии связи между снипом и признаком

Manhattan Plot of p-values from Breast Cancer GWAS

- Analysis of 582,886 SNPs for 3,659 cases with family history and 4,897 controls



Correcting for Multiple Testing

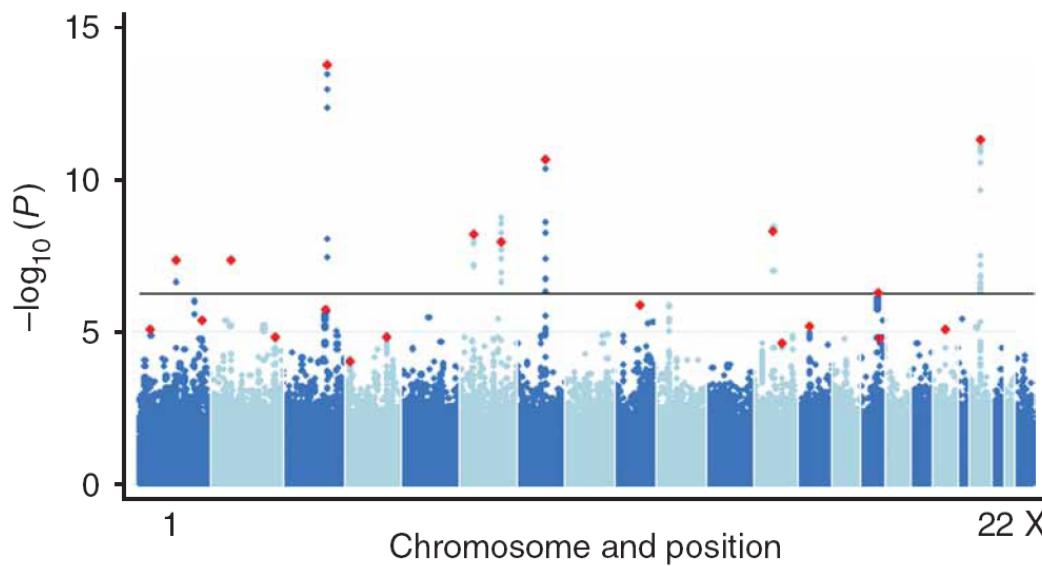
- What happens when we scan the genome of 1 million markers for association with $p = 0.05$?
 - 50,000 ($=1 \text{ million} \times 0.05$) SNPs are expected to be found significant just by chance
 - We need to be more conservative when we decide a given marker is significantly associated with the trait.
- Correction methods
 - Bonferroni correction

Bonferroni Correction

- if N markers are tested, we correct the significance level as $p' = p/N$
 - Assumes the N tests are independent, although this is not true because of the linkage disequilibrium.
 - $0.05 / 1\ 000\ 000 \sim 5 * 10^{-8}$

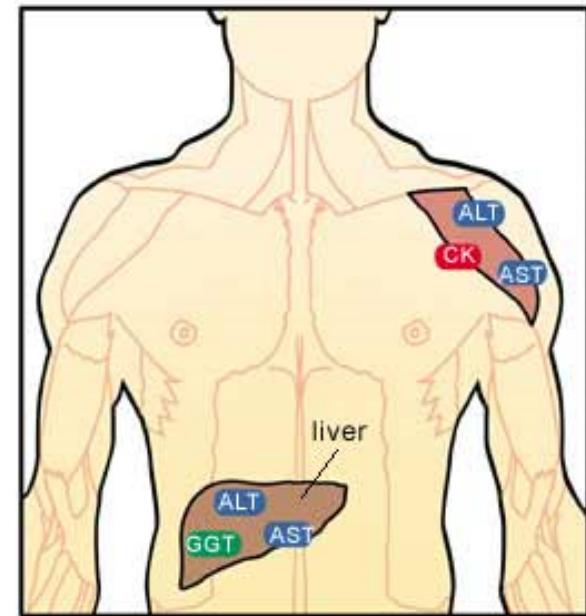
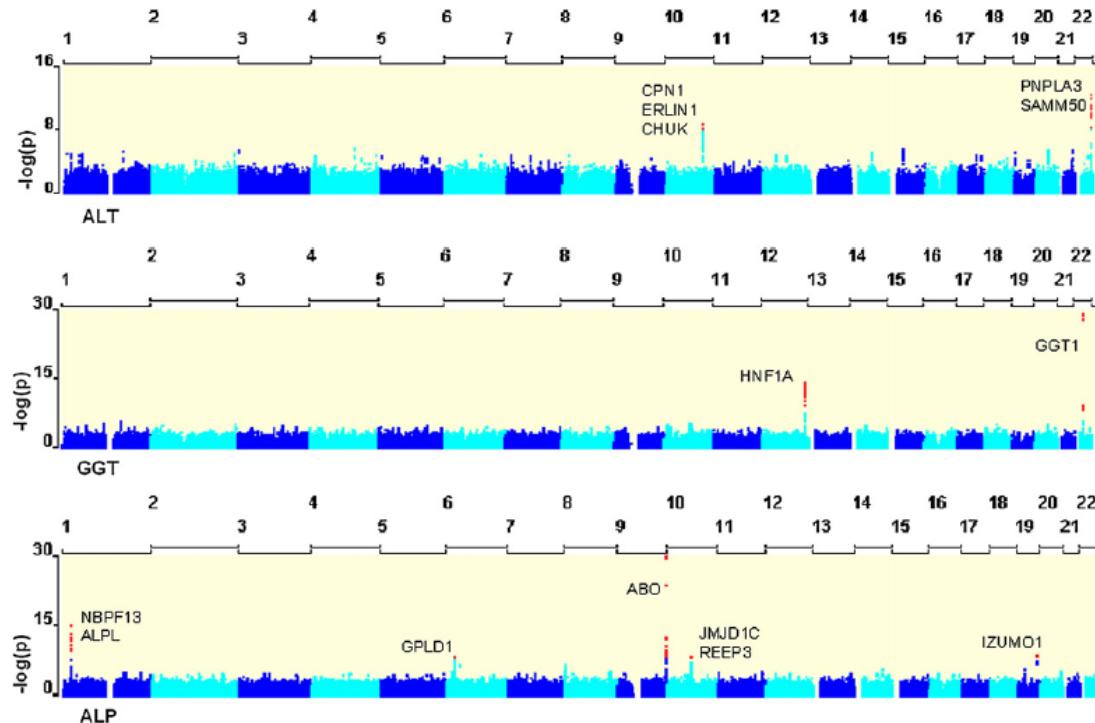
Genome-wide association analysis identifies 20 loci that influence adult height

Michael N Weedon^{1,2,23}, Hana Lango^{1,2,23}, Cecilia M Lindgren^{3,4}, Chris Wallace⁵, David M Evans⁶, Massimo Mangino⁷, Rachel M Freathy^{1,2}, John R B Perry^{1,2}, Suzanne Stevens⁷, Alistair S Hall⁸, Nilesh J Samani⁷, Beverly Shields², Inga Prokopenko^{3,4}, Martin Farrall⁹, Anna Dominiczak¹⁰, Diabetes Genetics Initiative²¹, The Wellcome Trust Case Control Consortium²¹, Toby Johnson^{11–13}, Sven Bergmann^{11,12}, Jacques S Beckmann^{11,14}, Peter Vollenweider¹⁵, Dawn M Waterworth¹⁶, Vincent Mooser¹⁶, Colin N A Palmer¹⁷, Andrew D Morris¹⁸, Willem H Ouwehand^{19,20}, Cambridge GEM Consortium²², Mark Caulfield⁵, Patricia B Munroe⁵, Andrew T Hattersley^{1,2}, Mark I McCarthy^{3,4} & Timothy M Frayling^{1,2}



Population-Based Genome-wide Association Studies Reveal Six Loci Influencing Plasma Levels of Liver Enzymes

Xin Yuan,¹ Dawn Waterworth,¹ John R.B. Perry,³ Noha Lim,¹ Kijoung Song,¹ John C. Chambers,⁴ Weihua Zhang,⁴ Peter Vollenweider,⁵ Heide Stirnadel,² Toby Johnson,^{6,7,8} Sven Bergmann,^{6,8} Noam D. Beckmann,⁶ Yun Li,¹² Luigi Ferrucci,⁹ David Melzer,³ Dena Hernandez,¹⁰ Andrew Singleton,¹⁰ James Scott,¹¹ Paul Elliott,⁴ Gerard Waeber,⁵ Lon Cardon,¹ Timothy M. Frayling,³ Jaspal S. Kooner,¹¹ and Vincent Mooser^{1,*}



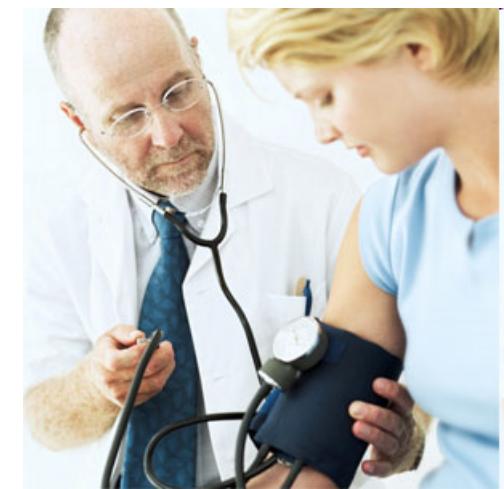
Common variants near MC4R are associated with fat mass, weight and risk of obesity

Ruth J F Loos^{*,1,2,73}, Cecilia M Lindgren^{3,4,73}, Shengxu Li^{1,2,73}, Eleanor Wheeler⁵, Jing Hua Zhao^{1,2}, Inga Prokopenko^{3,4}, Michael Inouye⁵, Rachel M Freathy^{6,7}, Antony P Attwood^{5,8}, Jacques S Beckmann^{9,10}, Sonja I Berndt¹¹, The Prostate, Lung, Colorectal, and Ovarian (PLCO) Cancer Screening Trial⁷¹, Sven Bergmann^{9,12}, Amanda J Bennett^{3,4}, Sheila A Bingham¹³, Murielle Bochud¹⁴, Morris Brown¹⁵, Stéphane Cauchi¹⁶, John M Connell¹⁷, Cyrus Cooper¹⁸, George Davey Smith¹⁹, Ian Day¹⁸, Christian Dina¹⁶, Subhajyoti De²⁰, Emmanouil T Dermitzakis⁵, Alex S F Doney²¹, Katherine S Elliott³, Paul Elliott^{22,23}, David M Evans^{3,19}, I Sadaf Farooqi^{2,24}, Philippe Froguel^{16,25}, Jilur Ghori⁵, Christopher J Groves^{3,4}, Rhian Gwilliam⁵, David Hadley²⁶, Alistair S Hall²⁷, Andrew T Hattersley^{6,7}, Johannes Hebebrand²⁸, Iris M Heid^{29,30}, KORA⁷¹, Blanca Herrera^{3,4}, Anke Hinney²⁸, Sarah E Hunt⁵, Marjo-Riitta Jarvelin^{22,23,31}, Toby Johnson^{9,12,14}, Jennifer D M Jolley⁸, Fredrik Karpe⁴, Andrew Keniry⁵, Kay-Tee Khaw³², Robert N Luben³², Massimo Mangino³³, Jonathan Marchini³⁴, Wendy L McArdle³⁵, Ralph McGinnis⁵, David Meyre¹⁶, Patricia B Munroe³⁶, Andrew D Morris²¹, Andrew R Ness³⁷, Matthew J Neville⁴, Alexandra C Nica⁵, Ken K Ong^{1,2}, Stephen O'Rahilly^{2,24}, Katharine R Owen⁴, Colin N A Palmer³⁸, Konstantinos Papadakis²⁶, Simon Potter⁵, Anneli Pouta^{31,39}, Lu Qi⁴⁰, Nurses' Health Study⁷¹, Joshua C Randall^{3,4}, Nigel W Rayner^{3,4}, Susan M Ring³⁵, Manjinder S Sandhu^{1,32}, André Scherag⁴¹, Matthew A Sims^{1,2}, Kijoung Song⁴², Nicole Soranzo⁵, Elizabeth K Speliotes^{43,44}, Diabetes Genetics Initiative⁷¹, Holly E Syddall¹⁸, Sarah A Teichmann²⁰, Nicholas J Timpson^{3,19}, Jonathan H Tobias⁴⁵, Manuela Uda⁴⁶, The SardiNIA Study⁷¹, Carla I Ganz Vogel²⁸, Chris Wallace³⁶, Dawn M Waterworth⁴², Michael N Weedon^{6,7}, The Wellcome Trust Case Control Consortium⁷², Cristen J Willer⁴⁷, FUSION⁷¹, Vicki L Wright^{2,24}, Xin Yuan⁴², Eleftheria Zeggini³, Joel N Hirschhorn^{44,48–51}, David P Strachan²⁶, Willem H Ouwehand⁸, Mark J Caulfield³⁶, Nilesh J Samani³³, Timothy M Frayling^{6,7}, Peter Vollenweider⁵², Gerard Waeber⁵², Vincent Mooser⁴², Panos Deloukas⁵, Mark I McCarthy^{3,4,73}, Nicholas J Wareham^{1,2,73} & Inês Barroso^{5,73}

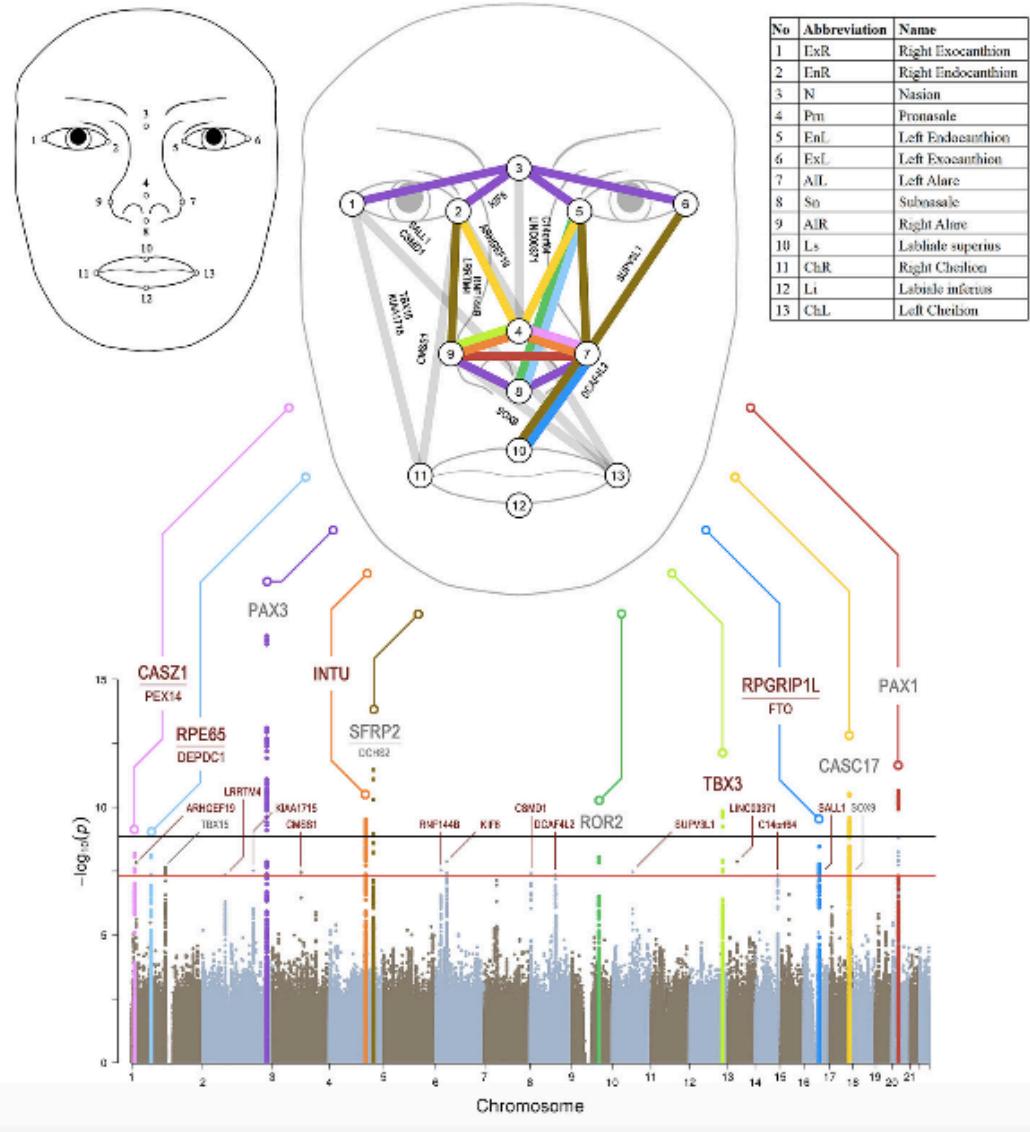


Genome-wide association study identifies eight loci associated with blood pressure

Christopher Newton-Cheh^{1–3,94†}, Toby Johnson^{4–6,94}, Vesela Gateva^{7,94}, Martin D Tobin^{8,94}, Murielle Bochud⁵, Lachlan Coin⁹, Samer S Najjar¹⁰, Jing Hua Zhao^{11,12}, Simon C Heath¹³, Susana Eyheramendy^{14,15}, Konstantinos Papadakis¹⁶, Benjamin F Voight^{1,3}, Laura J Scott⁷, Feng Zhang¹⁷, Martin Farrall^{18,19}, Toshiko Tanaka^{20,21}, Chris Wallace^{22–24}, John C Chambers⁹, Kay-Tee Khaw^{12,25}, Peter Nilsson²⁶, Pim van der Harst²⁷, Silvia Polidoro²⁸, Diederick E Grobbee²⁹, N Charlotte Onland-Moret^{29,30}, Michiel L Bots²⁹, Louise V Wain⁸, Katherine S Elliott¹⁹, Alexander Teumer³¹, Jian'an Luan¹¹, Gavin Lucas³², Johanna Kuusisto³³, Paul R Burton⁸, David Hadley¹⁶, Wendy L McArdle³⁴, Wellcome Trust Case Control Consortium⁹³, Morris Brown³⁵, Anna Dominiczak³⁶, Stephen J Newhouse^{22,23}, Nilesh J Samani³⁷, John Webster³⁸, Eleftheria Zeggini^{19,39}, Jacques S Beckmann^{4,40}, Sven Bergmann^{4,6}, Noha Lim⁴¹, Kijoung Song⁴¹, Peter Vollenweider⁴², Gerard Waeber⁴², Dawn M Waterworth⁴¹, Xin Yuan⁴¹, Leif Groop^{43,44}, Marju Orho-Melander²⁶, Alessandra Allione²⁸, Alessandra Di Gregorio^{28,45}, Simonetta Guarrera²⁸, Salvatore Panico⁴⁶, Fulvio Ricceri²⁸, Valeria Romanazzi^{28,45}, Carlotta Sacerdote⁴⁷, Paolo Vineis^{9,28}, Inès Barroso^{12,39}, Manjinder S Sandhu^{11,12,25}, Robert N Luben^{12,25}, Gabriel J Crawford³, Pekka Jousilahti⁴⁸, Markus Perola^{48,49}, Michael Boehnke⁷, Lori L Bonnycastle⁵⁰, Francis S Collins⁵⁰, Anne U Jackson⁷, Karen L Mohlke⁵¹, Heather M Stringham⁷, Timo T Valle⁵², Cristen J Willer⁷, Richard N Bergman⁵³, Mario A Morken⁵⁰, Angela Döring¹⁵, Christian Gieger¹⁵, Thomas Illig¹⁵, Thomas Meitinger^{54,55}, Elin Org⁵⁶, Arne Pfeufer^{54,55}, H Erich Wichmann^{15,57}, Sekar Kathiresan^{1–3}, Jaume Marrugat³², Christopher J O'Donnell^{58,59}, Stephen M Schwartz^{60,61}, David S Siscovick^{60,61}, Isaac Subirana^{32,62}, Nelson B Freimer⁶³, Anna-Liisa Hartikainen⁶⁴, Mark I McCarthy^{19,65,66}, Paul F O'Reilly⁹, Leena Peltonen^{39,49}, Anneli Pouta^{64,67}, Paul E de Jong⁶⁸, Harold Snieder⁶⁹, Wiek H van Gilst²⁷, Robert Clarke⁷⁰, Anuj Goel^{18,19}, Anders Hamsten⁷¹, John F Peden^{18,19}, Udo Seedorf⁷², Ann-Christine Svänen⁷³, Giovanni Tognoni⁷⁴, Edward G Lakatta¹⁰, Serena Sanna⁷⁵, Paul Scheet⁷⁶, David Schlessinger⁷⁷, Angelo Scuteri⁷⁸, Marcus Dörr⁷⁹, Florian Ernst³¹, Stephan B Felix⁷⁹, Georg Homuth³¹, Roberto Lorbeer⁸⁰, Thorsten Reffelmann⁷⁹, Rainer Rettig⁸¹, Uwe Völker³¹, Pilar Galan⁸², Ivo G Gut¹³, Serge Hercberg⁸², G Mark Lathrop¹³, Diana Zelenika¹³, Panos Deloukas^{12,39}, Nicole Soranzo^{17,39}, Frances M Williams¹⁷, Guangju Zhai¹⁷, Veikko Salomaa⁴⁸, Markku Laakso³³, Roberto Elosua^{32,62}, Nita G Forouhi¹¹, Henry Völzke⁸⁰, Cuno S Uiterwaal²⁹, Yvonne T van der Schouw²⁹, Mattijs E Numans²⁹, Giuseppe Matullo^{28,45}, Gerjan Navis⁶⁸, Göran Berglund²⁶, Sheila A Bingham^{12,83}, Jaspal S Kooner⁸⁴, John M Connell³⁶, Stefania Bandinelli⁸⁵, Luigi Ferrucci²¹, Hugh Watkins^{18,19}, Tim D Spector¹⁷, Jaakkko Tuomilehto^{52,86,87}, David Altshuler^{1,3,88,89}, David P Strachan¹⁶, Maris Laan⁵⁶, Pierre Meneton⁹⁰, Nicholas J Wareham^{11,12}, Manuela Uda⁷⁵, Marjo-Rüütta Jarvelin^{9,67,91}, Vincent Mooser⁴¹, Olle Melander²⁶, Ruth JF Loos^{11,12}, Paul Elliott^{9,94}, Gonçalo R Abecasis^{92,94}, Mark Caulfield^{22,23,94} & Patricia B Munroe^{22,23,94}



Novel genetic loci affecting facial shape variation in humans



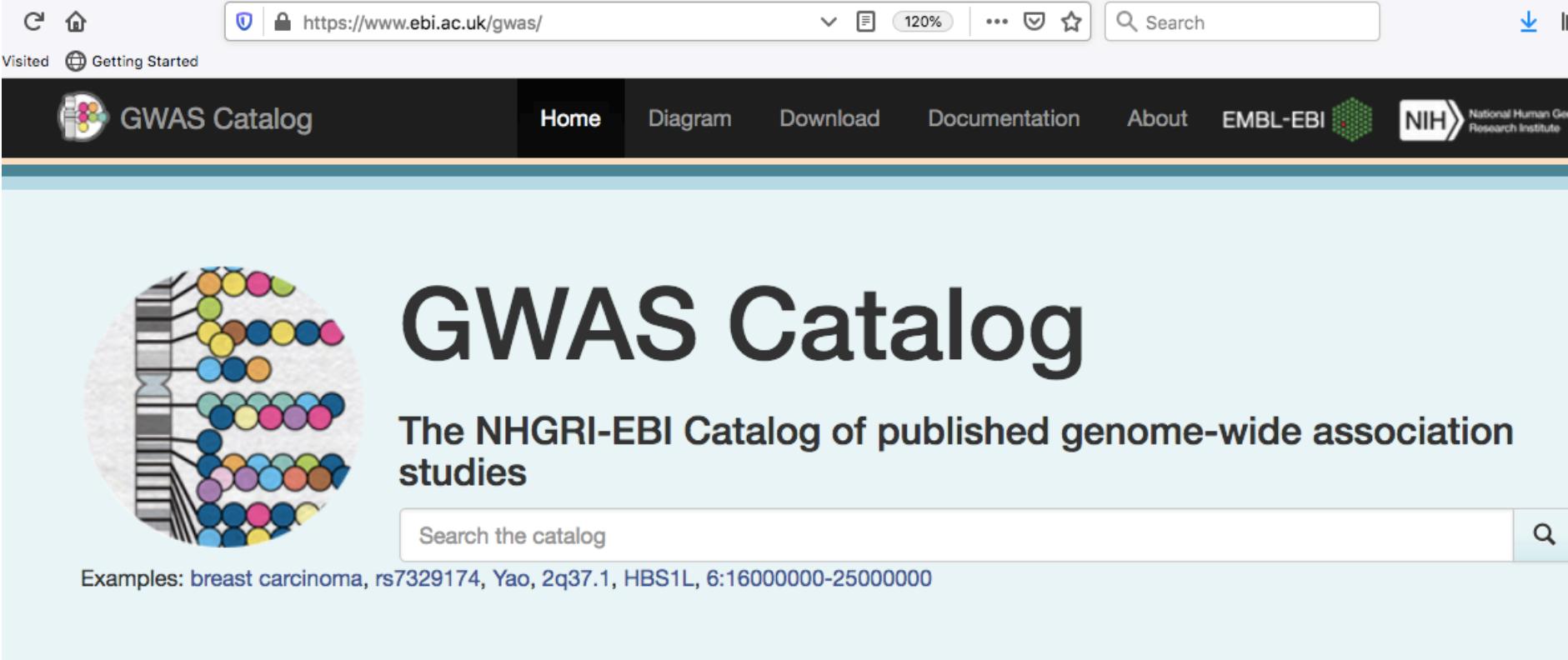
SNPs significantly associated ($p < 5 \times 10^{-8}$) with facial shape phenotypes from European discovery GWAS meta-analysis (RS, TwinksUK, ALSPAC, and PITT), and their multi-ethnic replication (UYG, CANDELA and QIMR).

Region	SNP	Nearest Gene	EA	OA	Trait	Discovery meta-analysis		Replication	
						Beta	P	(N = 10,115)	(N = 7,917)
Novel face-associated loci									
1p36.22	rs143353512	CASZ1	A	G	Prn-AIL	-0.29	6.44×10^{-9}	0.73	0.0001
1p36.13	rs200243292	ARHGEF19	I	T	EnR-ChL	-0.11	1.46×10^{-8}	0.54	0.0640
1p31.2									
2p12									
2q31.1									
3q12.1									
4q28.1									
6p22.3									
6p21.2									
8p23.2									
8q21.3									
10q22.									
12q24.									
13q14.									
14q32.2	rs1989285	C14orf64	G	C	N-Prn	-0.10	4.54×10^{-8}	0.82	0.0019
16q12.1	rs16949899	SALL1	T	G	ExR-ChL	-0.13	3.35×10^{-8}	0.76	0.8912
16q12.2	rs7404301	RPGRIP1L	G	A	AIL-Ls	-0.09	3.49×10^{-9}	0.43	2.87×10^{-7}

Genotyping was carried out using the Infinium II HumanHap 550K GenotypingBeadChip version 3 (Illumina, San Diego, California USA).

We then set our study-wide significant threshold at $p < 1.2 \times 10^{-9}$ using Bonferroni correction of 43 phenotypes, considering significant threshold for single phenotype GWAS as $P < 5 \times 10^{-8}$.

<https://www.ebi.ac.uk/gwas/>



The screenshot shows the GWAS Catalog homepage. At the top, there is a navigation bar with links for Home, Diagram, Download, Documentation, About, EMBL-EBI, and NIH. Below the navigation bar, the main title "GWAS Catalog" is displayed in large, bold letters. To the left of the title is a circular graphic featuring a stylized DNA helix composed of colored circles (blue, green, yellow, red) and vertical bars. Below the title, the subtitle "The NHGRI-EBI Catalog of published genome-wide association studies" is shown. A search bar with the placeholder "Search the catalog" and a magnifying glass icon is located below the subtitle. At the bottom of the page, there is a text block providing information about the catalog's creation and history.

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GWAS Catalog

The NHGRI-EBI Catalog of published genome-wide association studies

Search the catalog

Examples: breast carcinoma, rs7329174, Yao, 2q37.1, HBS1L, 6:16000000-25000000

It was created by the [National Human Genome Research Institute](#) (NHGRI) in 2008 and have become a collaborative project between the NHGRI and the [European Bioinformatics Institute](#) (EBI) since 2010.^[1] As of September 2018, it has included 71,673 [SNP](#)-trait associations in 3,567 publications.



This diagram shows all SNP-trait associations with $p\text{-value} \leq 5.0 \times 10^{-8}$, published in the GWAS Catalog

Как рассчитать риск заболевания?

См семинар

	Здоровы	Больны
A	55	22
G	21	0
AA	22	11
AG	11	0
GG	5	0
AA / AG	22	11
GG / AG	16	0
AA / AG	5	0
GG / AA	33	11

38 – здоровы
11 – больны
49 – всего людей

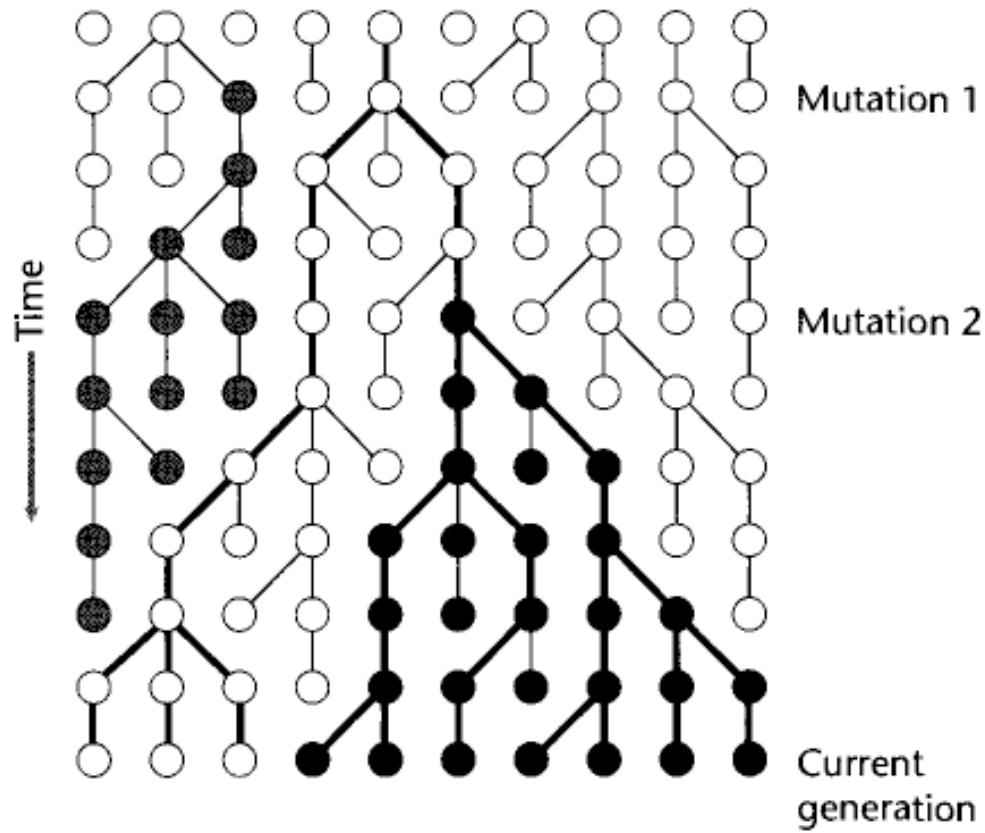
$11/49 = 0.22$ –
риск
заболевания без
генотипирования

Для AA
22 – здоровы
11 – больны
33 – всего людей

$11/33 = 0.33$ –
риск
заболевания с
аллелем AA

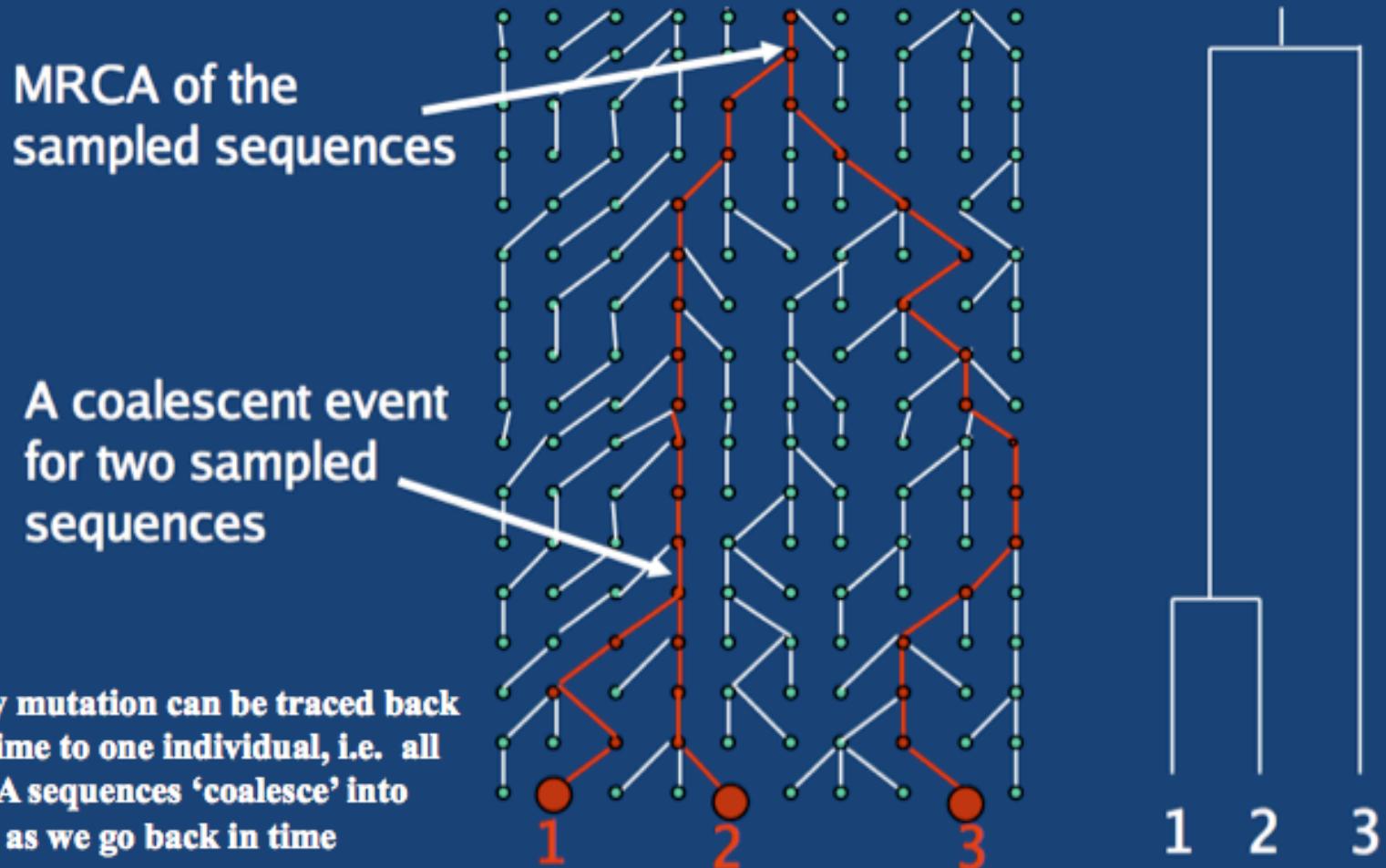
Увеличение риска заболевания при наличии аллели AA – $0.33/0.22=1.5$

Популяционная генетика



Альбрехт Дюрер “Адам и Ева”

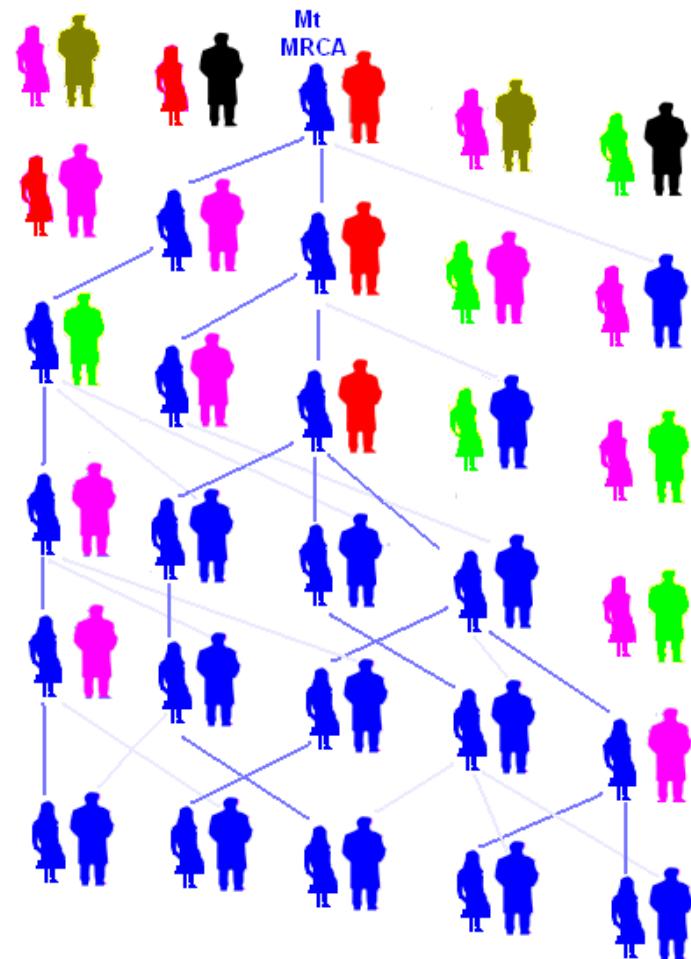
Теория коалесценции

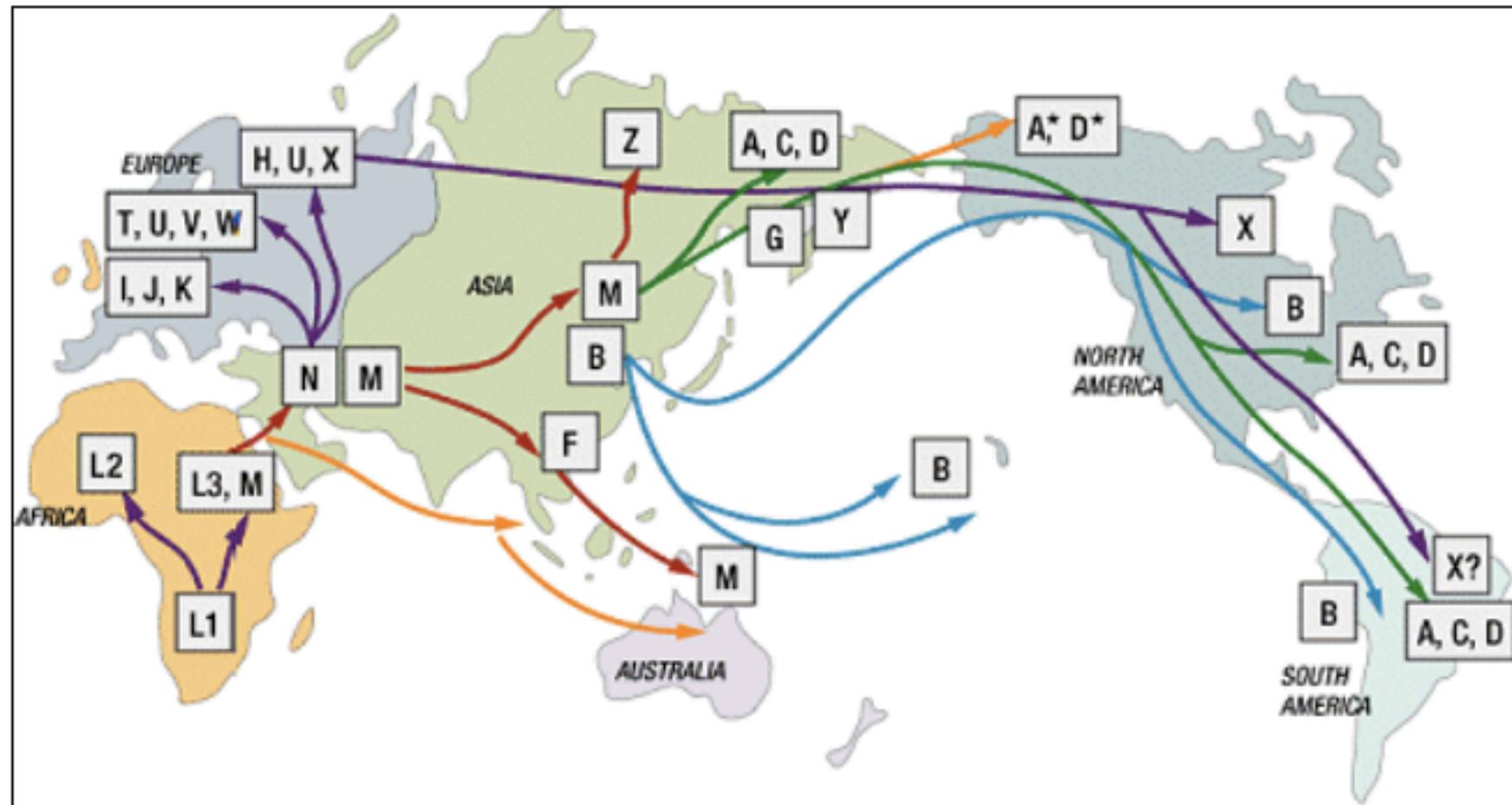


Митохондриальная Ева (Mt MRCA)

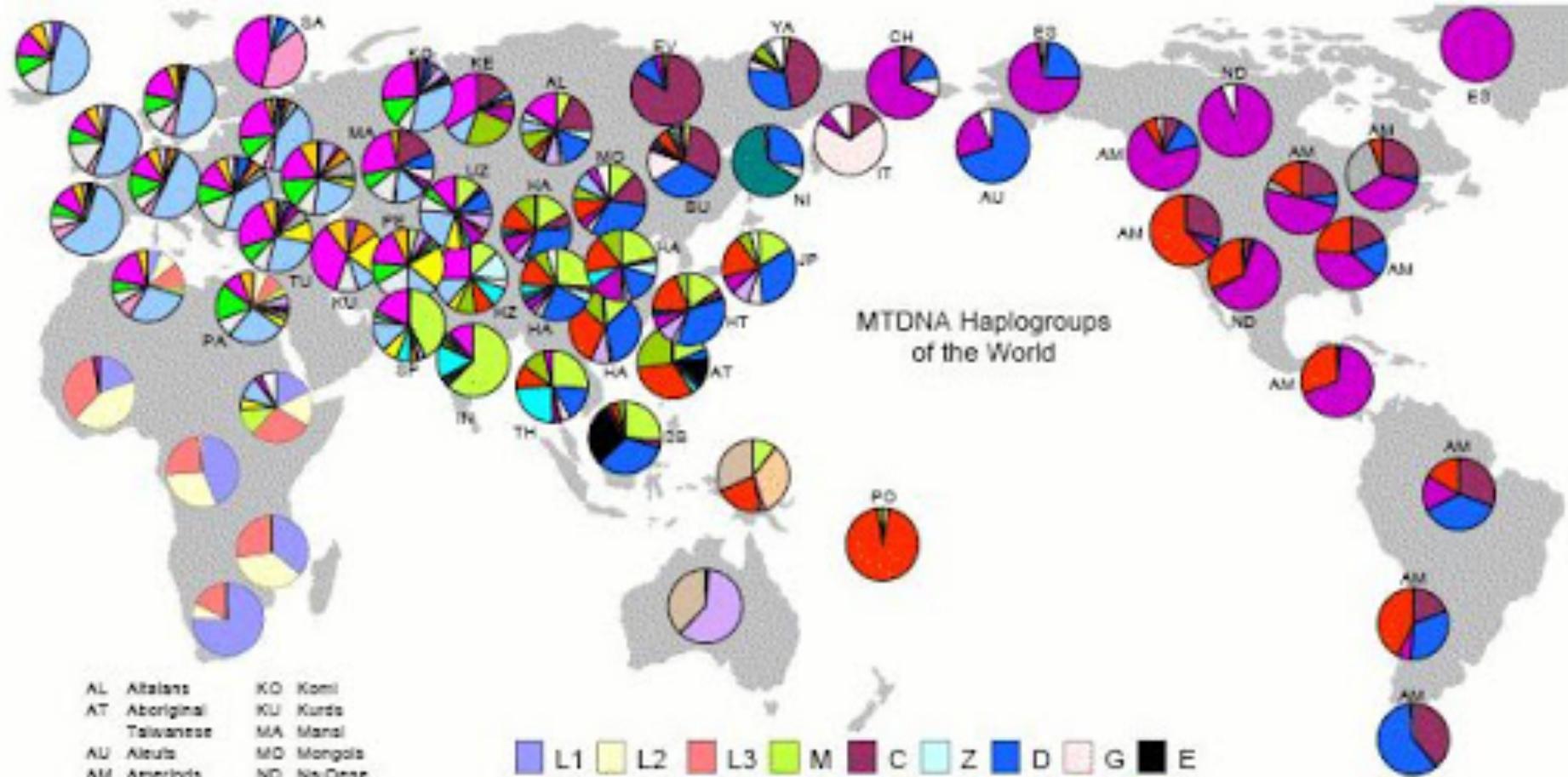
MRCA - most recent common ancestor

- Жила около 200,000 лет назад, возможно, в Восточной Африке
- Жила гораздо раньше, чем, по оценкам. произошла миграция из Африки - где-то между 95,000 и 45,000.





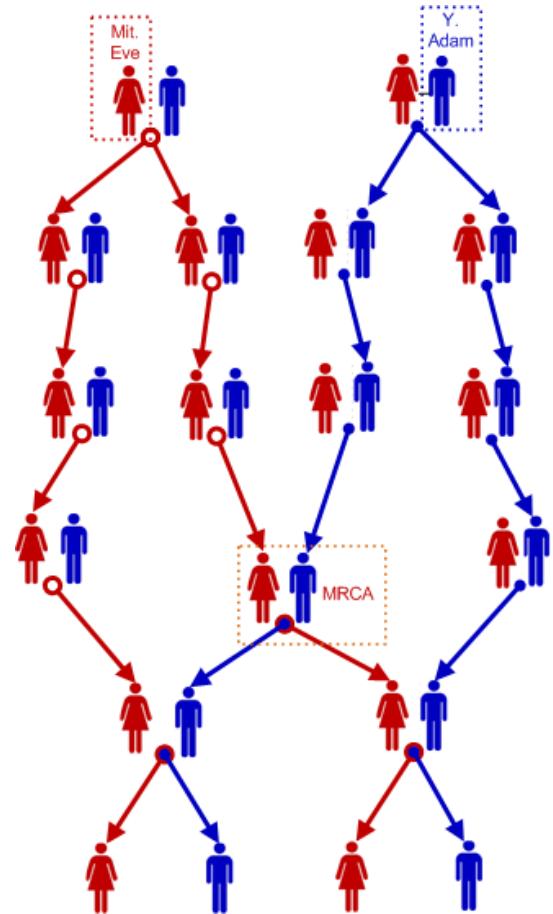

FamilyTreeDNA
 mtDNA Migrations Map



Specific tribes or locations are shown at left. Unlabelled sites are for general population in the area. African, American, and especially Polynesian areas are very large. The data in this chart is supposed to represent the situation before the recent European expansion beginning about 1500 AD. Assignments in Australia are somewhat iffy.

Y-хромосомный Адам (Y-MRCA)

- 60,000 years ago



- Map shows first migratory routes taken by humans, based on surveys of different types of the male Y chromosome.
"Adam" represents the common ancestor from which all Y chromosomes descended
- Research based on DNA testing of 10,000 people from indigenous populations around the world

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New binary polymorphisms reshape and increase resolution of the human Y chromosomal haplogroup tree

Y-Chromosome Phylogenetic Tree

Files in this Data Supplement:

- Y-Chromosome Phylogenetic Tree Poster

This Article

Published in Advance April 2, 2008, doi:
[10.1101/gr.7172008](https://doi.org/10.1101/gr.7172008)

Genome Res. May 2008 vol. 18 no. 5 830–838

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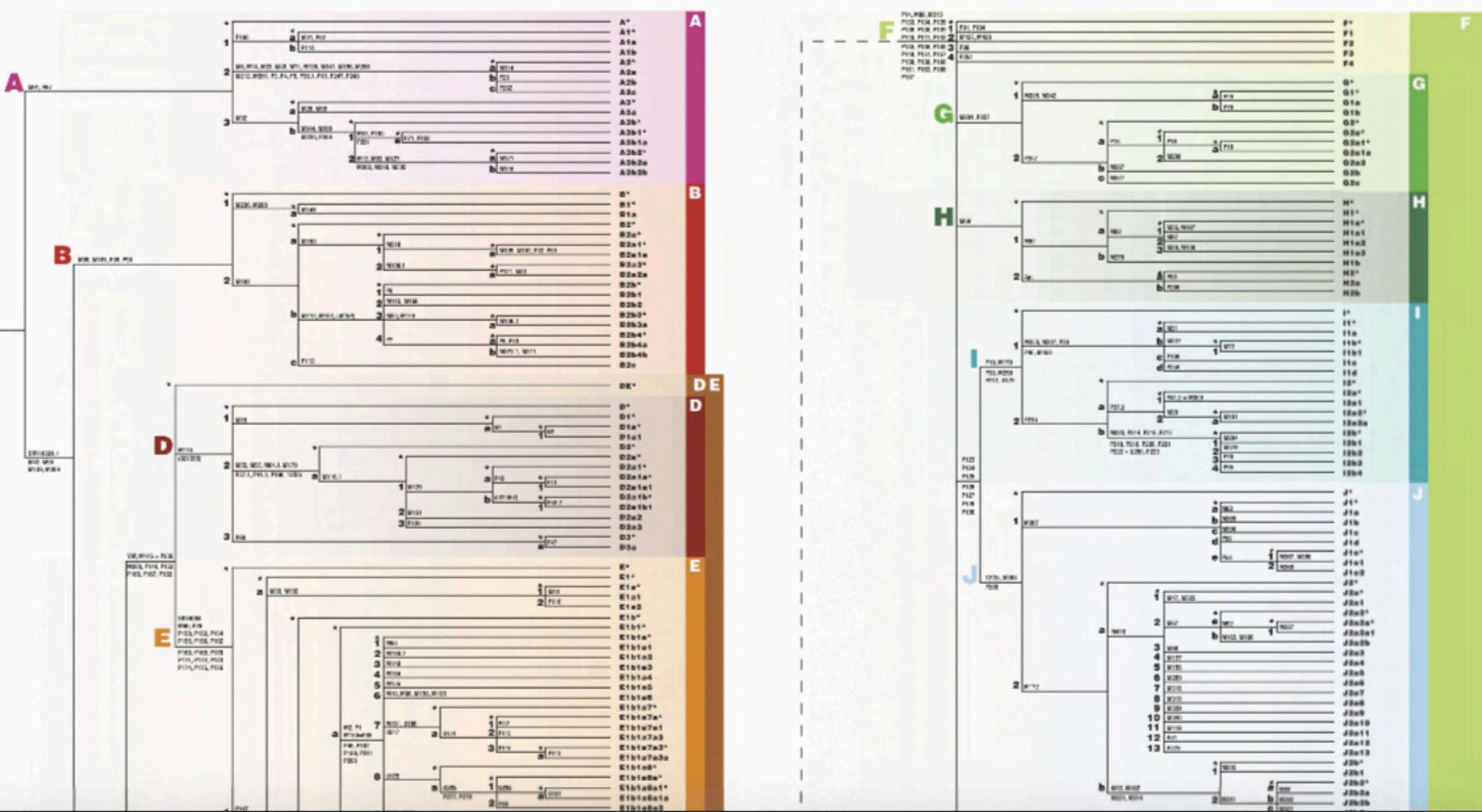
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Supplemental Research Data

» Y-Chromosome Phylogenetic Tree Poster

Y-Chromosome Phylogenetic Tree





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Y-DNA haplogroups

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Origins, spread and ethnic association of European haplogroups and subclades



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[MtDNA Haplogroups](#)

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- [Haplogroups J & T](#)

- [Haplogroup W](#)
- [Haplogroup I](#)

- [Haplogroup X](#)
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Author: [Maciamo Hay](#). Last update August 2017.

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2

t

- **The influence of environmental factors:** The risk for breast and ovarian cancer is only partially determined by genetics. Environmental factors, including but not limited to diet and lifestyle, also play significant roles.
- **This is not the entire genetic picture:** The mutations reported by 23andMe account for only a portion of the entire genetic contribution to breast and ovarian cancer. There are other known mutations, including many in BRCA1 and BRCA2, for which 23andMe does not provide data. If you are concerned about these, you should consult a medical professional about taking specific tests that offer a more complete assessment of these two genes. There are also unidentified genetic factors that affect breast cancer risk.
- **Your ancestry affects your chances of having these mutations:** Though extremely rare in the general population, these mutations are much more common in families with Ashkenazi Jewish ancestry.
- **The mutations described here cannot predict definitively whether you will develop breast or ovarian cancer:** Though having these mutations greatly increases the risk for both diseases, many people who have them will never get the disease. Conversely, lacking these mutations does not substantially reduce your breast or ovarian cancer risk.
- **These mutations are also relevant to men:** Although men are not at risk for ovarian cancer and are at very low risk for breast cancer, BRCA1 and BRCA2 mutations can increase a man's risk for prostate cancer and male breast cancer. Men who carry one of these mutations have a 50% chance of passing it on to their daughters, who would then be at increased risk for breast and ovarian cancer. The mothers and sisters of men who carry one of these mutations also have a 50% chance of being carriers.
- **The wishes of members in your account:** You are about to unlock results for everyone in your account, including the following individuals:

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Disease Risks (12)

	Type 1 Diabetes
	Type 2 Diabetes
	Rheumatoid Arthritis
	Crohn's Disease
	Age-related Macular Degeneration
	See all 12 risk reports...

Carrier Status (21)

Alpha-1 Antitrypsin Deficiency	Variant Absent
BRCA Cancer Mutations (Selected)	Variant Absent
Bloom's Syndrome	Variant Absent
Canavan Disease	Variant Absent
Connexin 26-Related Sensorineural Hearing Loss	Variant Absent
	See all 21 carrier status...

Traits (10)

Alcohol Flush Reaction		Does Not Flush
Bitter Taste Perception		Can Taste
Earwax Type		Wet
Eye Color		Likely Brown
Lactose Intolerance		Likely Tolerant
		See all 10 traits...

Drug Response (8)

Clopidogrel (Plavix®) Efficacy	Greatly Reduced
Alcohol Consumption, Smoking and Risk of Esophageal Cancer	new
Response to Hepatitis C Treatment	new
Abacavir Hypersensitivity	Typical
Fluorouracil Toxicity	Typical
	See all 8 drug response...

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Elevated Risk

Name	Absolute Risk	Relative Risk	Last Updated
No diseases in this category.			

Decreased Risk

Name	Absolute Risk	Relative Risk	Last Updated
Celiac Disease	0.03%	0.26	Jul 7, 2009
Age-related Macular Degeneration	2.3%	0.33	May 21, 2008
Crohn's Disease	0.3%	0.50	Jul 16, 2009
Rheumatoid Arthritis	1.4%	0.59	Aug 6, 2009
Type 2 Diabetes	17%	0.70	Feb 2, 2009
Type 1 Diabetes	0.8%	0.78	Jul 30, 2009

Typical Risk

Name	Absolute Risk	Relative Risk	Last Updated
Prostate Cancer	18%	1.03	Oct 22, 2009
Parkinson's Disease	1.6%	0.98	Sep 29, 2008
Venous Thromboembolism	12%	0.96	Jul 30, 2009
Psoriasis	9.9%	0.87	Jul 7, 2009
Atrial Fibrillation	23%	0.85	Oct 29, 2009
Breast Cancer	Not Applicable		Feb 18, 2010

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Research Reports give you information from research that has not yet gained enough scientific consensus to be included in our Clinical Reports. It also includes established research that does not have a dramatic influence on a person's risk for a disease.

Sort groups by: [Name](#) | [Research Confidence](#) | [Last Updated Date](#)

Elevated Risk [?](#)

Name	Research Confidence	Last Updated
Colorectal Cancer	★★★★	Jul 16, 2009
Exfoliation Glaucoma	★★★★	Jun 25, 2009
Restless Legs Syndrome	★★★★	Jul 16, 2009
Abdominal Aortic Aneurysm	★★★	Nov 21, 2008
Ankylosing Spondylitis	★★★	Feb 21, 2008
Asthma	★★★	May 12, 2008
Brain Aneurysm	★★★	Nov 21, 2008
Celiac Disease: Preliminary Research	★★★	Apr 9, 2008
Chronic Lymphocytic Leukemia	★★★	Nov 21, 2008
Neuroblastoma	★★★	May 9, 2008
Tuberculosis	★★★	Apr 23, 2009
Cleft Lip and Cleft Palate	★★	Jun 18, 2009
Developmental Dyslexia	★★	Feb 21, 2008
Gout	★★	Apr 21, 2008



23andMe Carrier Status

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Name	Status	Last Updated
Alpha-1 Antitrypsin Deficiency	Variant.Absent	Apr 9, 2009
BRCA Cancer Mutations (Selected)	Variant.Absent	Feb 12, 2009
Bloom's Syndrome	Variant.Absent	Jan 7, 2009
Canavan Disease	Variant.Absent	Nov 19, 2009
Connexin 26-Related Sensorineural Hearing Loss	Variant.Absent	Nov 19, 2009
Cystic Fibrosis	Variant.Absent	Nov 19, 2009
Factor XI Deficiency	Variant.Absent	Nov 19, 2009
Familial Dysautonomia	Variant.Absent	Nov 19, 2009
Fanconi Anemia (FANCC-related)	Variant.Absent	Nov 19, 2009
G6PO Deficiency	Variant.Absent	Aug 27, 2009
Gaucher Disease	Variant.Absent	Nov 19, 2009
Glycogen Storage Disease Type 1a	Variant.Absent	Jan 7, 2009
Hemochromatosis	Variant.Absent	Dec 18, 2008
Limb-girdle Muscular Dystrophy	Variant.Absent	Nov 19, 2009
Maple Syrup Urine Disease Type 1B	Variant.Absent	Nov 19, 2009
Mucolipidosis IV	Variant.Absent	Nov 19, 2009
Niemann-Pick Disease Type A	Variant.Absent	Nov 19, 2009
Rhizomelic Chondrodysplasia Punctata Type 1 (RCDP1)	Variant.Absent	Nov 19, 2009
Sickle Cell Anemia & Malaria Resistance	Variant.Absent	Sep 3, 2008
Tay-Sachs Disease	Variant.Absent	Nov 19, 2009
Torsion Dystonia	Variant.Absent	Nov 19, 2009

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Name	Outcome	Last Updated
Alcohol Flush Reaction	Does Not Flush	Dec 19, 2007
Bitter Taste Perception	Can Taste	Nov 19, 2007
Earwax Type	Wet	Nov 19, 2007
Eye Color	Likely Brown	Mar 25, 2008
Lactose Intolerance	Likely Tolerant	Nov 19, 2007
Malaria Resistance (Duffy Antigen)	Not Resistant	Feb 28, 2008
Muscle Performance	Likely Sprinter	Nov 19, 2007
Non-ABO Blood Groups	See Report	Mar 25, 2008
Norovirus Resistance	Not Resistant	Jul 23, 2008
Resistance to HIV/AIDS	Not Resistant	Jan 27, 2008

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maternal line

Your mitochondrial DNA determines your maternal haplogroup. [What is a haplogroup?](#)

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Maternal Haplogroup: U5

Locations of haplogroup U5 circa 500 years ago, before the era of intercontinental travel.



Haplogroup U5 arose among early colonizers of Europe around 40,000 years ago; maternal descendants of those early colonizers persist in the region to this day. After the last Ice Age two subgroups of U5 expanded across Europe and into northern Africa and the Near East. Today, one subgroup, U5b1b, is shared by groups as diverse as the northern African desert-dwelling Berbers and the Scandinavian Arctic-dwelling Saami, also known as the Lapps.

Haplogroup: U5, a subgroup of [U](#)

Age: 40,000 years

Region: Europe, Near East, North Africa

Populations: Basques, Saami (Lapps) of northern Scandinavia

Highlight: Though primarily a European haplogroup, U5 was recently found in mitochondrial DNA extracted from the remains of a 6th-century AD Chinese chieftain.

Your Family and Friends

[K1a1b1a](#) Simone Brutlag

[U5b2](#) Douglas Brutlag

[L3e](#) Nigerian Man

[D5a2](#) Chinese Man

[D4e2](#) Japanese Man



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paternal line

Your Y chromosome DNA determines your paternal haplogroup. [What is a haplogroup?](#)

[Map](#) [History](#) [Haplogroup Tree](#)

Paternal Haplogroup: E3b1a

Locations of haplogroup E3b1a circa 500 years ago, before the era of intercontinental travel.



E3b is most common in northern Africa and southern Europe. It arose about 17,000 years ago in eastern Africa and spread into the Mediterranean region after the Ice Age. E3b1a, a subgroup of E3b, expanded out of the Near East 8,000 years ago into northern Africa and southern Europe. Today it is one of the most common haplogroups in those regions.

Haplogroup: E3b1a, a subgroup of [E3b](#)

Age: 14,000 years

Region: Northern Africa, Southern Europe

Populations: Berbers, Iberians, Balkans

Highlight: Two different migrations brought E3b1a into Europe.

Your Family and Friends

[N](#) Chinese Man

[E3a8a](#) Nigerian Man

[E3b1a](#) Douglas Brutlag

[D2](#) Japanese Man

[N/A](#) Simone Brutlag



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ancestry painting

Trace the ancestry of your chromosomes, one segment at a time. Last updated April 23th, 2008.

Chromosome View

Solid segments indicate that both chromosomes come from the same geographic region. See a Cambodian Woman's painting.
Dual-colored segments indicate chromosomes from different geographic regions. See an African American Man's painting.

Select a person: Douglas Brutlag



Douglas Brutlag

Europe 100%
Asia 0%
Africa 0%
Not Genotyped

Worldwide Examples

Click on the icons in the map below to see sample paintings of individuals from across the globe.



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...why only three populations are used.
...why it says I'm European/African/Asian when I'm really an American/Australian/South African.
...how the percentages are calculated.
...where the X and Y chromosomes are.

Генотек



https://www.genotek.ru

67%



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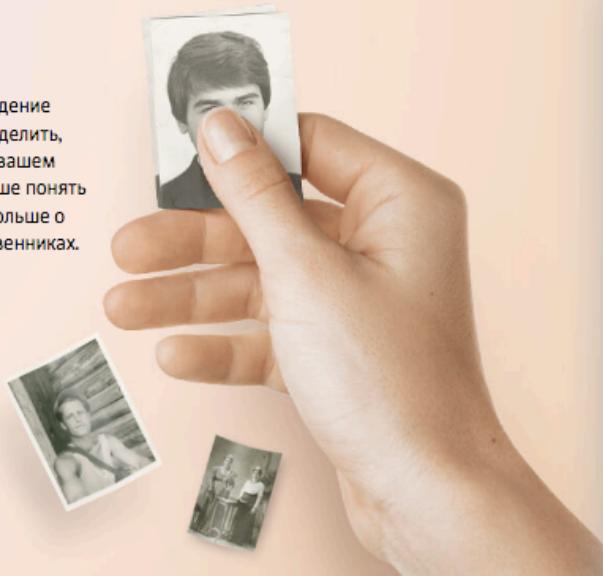
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