



# Л е к ц и я № 7

## Полногеномный анализ ассоциаций

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Кафедра фармакологии и биоинформатики

Для студентов, обучающихся по направлению 06.03.01 «Биология»  
профили Биохимия, Генетика  
при изучении дисциплины «Биоинформатика»

# П л а н л е к ц и и

- **Полногеномный анализ ассоциаций GWAS**
- **Моногенные заболевания**
- **Однонуклеотидный полиморфизм SNP**
- **Мультифакторные заболевания**

# Характеристики генома

- 24 хромосомы
- ~3.2 млрд. пар нуклеотидов
- ~25 тыс. генов, ~1.3% генома
- Один ген — 1000-3000 оснований
- Два генома идентичны на ~99.9 %
- Фенотипические признаки и все заболевания — ~0.1 % генома, т.е. его 1/1000 мутированная часть

# Факторы заболеваний

## Deciphering Complex Disease

A systems biology approach using the SOLiD™ System

Detect and Sequence Insertions and Deletions

Discover and Quantitate Non-Coding RNA

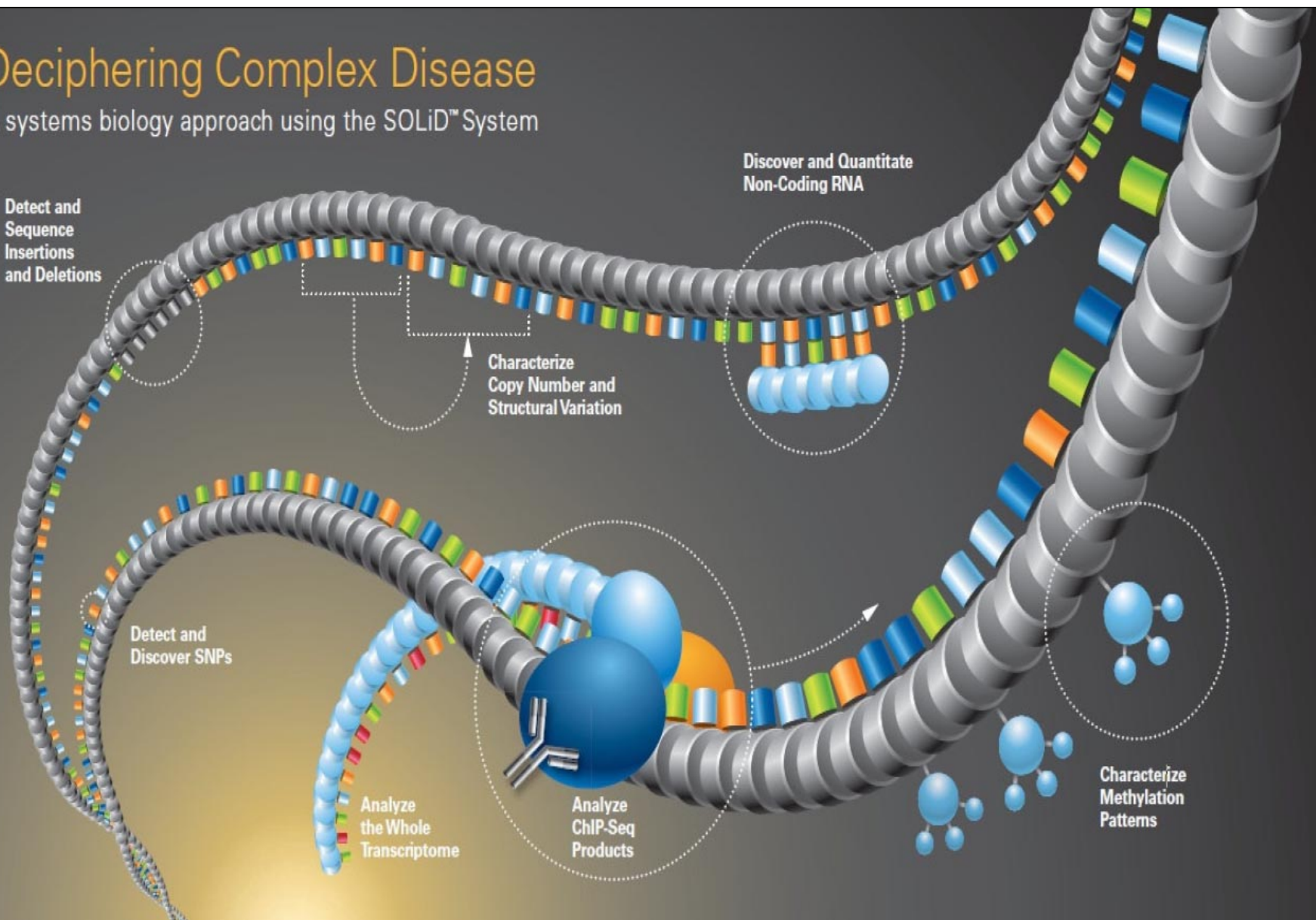
Characterize Copy Number and Structural Variation

Detect and Discover SNPs

Analyze the Whole Transcriptome

Analyze ChIP-Seq Products

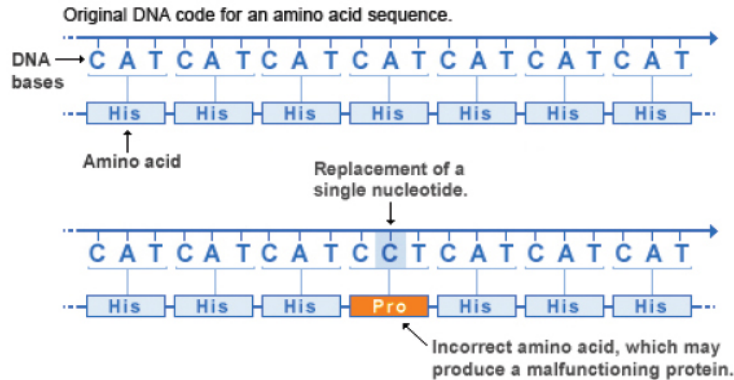
Characterize Methylation Patterns



# Мутации

## MISSENSE MUTATION

## Замены

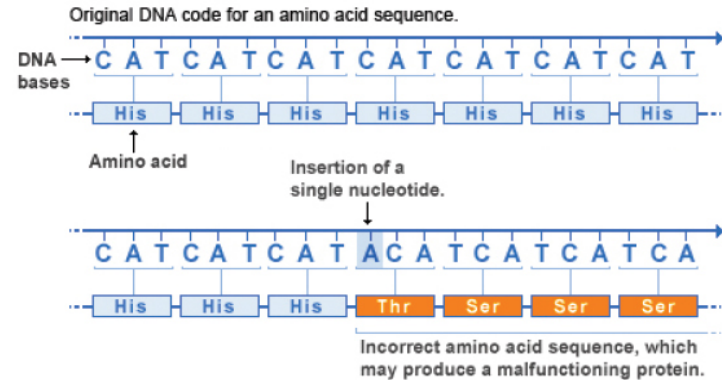


U.S. National Library of Medicine

This type of mutation is a change in one DNA base pair that results in the substitution of one amino acid for another in the protein made by a gene.

## INSERTION

## Вставки

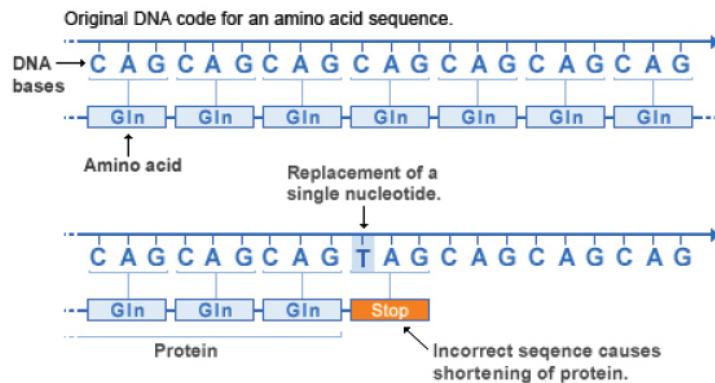


U.S. National Library of Medicine

An insertion changes the number of DNA bases in a gene by adding a piece of DNA. As a result, the protein made by the gene may not function properly.

## NONSENSE MUTATION

## Нонсенсы

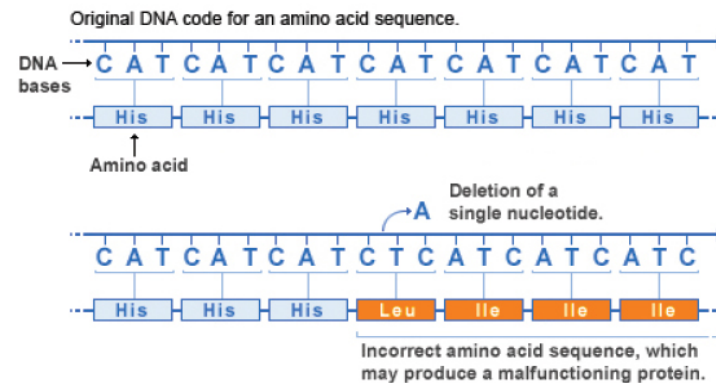


U.S. National Library of Medicine

A nonsense mutation is also a change in one DNA base pair. Instead of substituting one amino acid for another, however, the altered DNA sequence prematurely signals the cell to stop building a protein. This type of mutation results in a shortened protein that may function improperly or not at all.

## DELETION

## Делеции



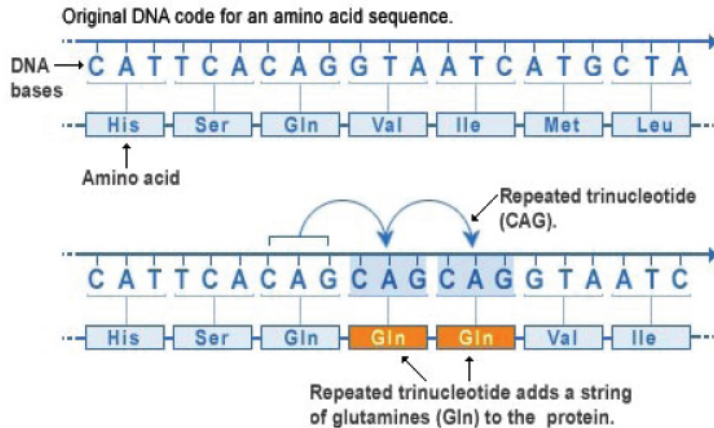
U.S. National Library of Medicine

A deletion changes the number of DNA bases by removing a piece of DNA. Small deletions may remove one or a few base pairs within a gene, while larger deletions can remove an entire gene or several neighboring genes. The deleted DNA may alter the function of the resulting protein(s).

# Мутации

## REPEAT EXPANSION

## Повторы

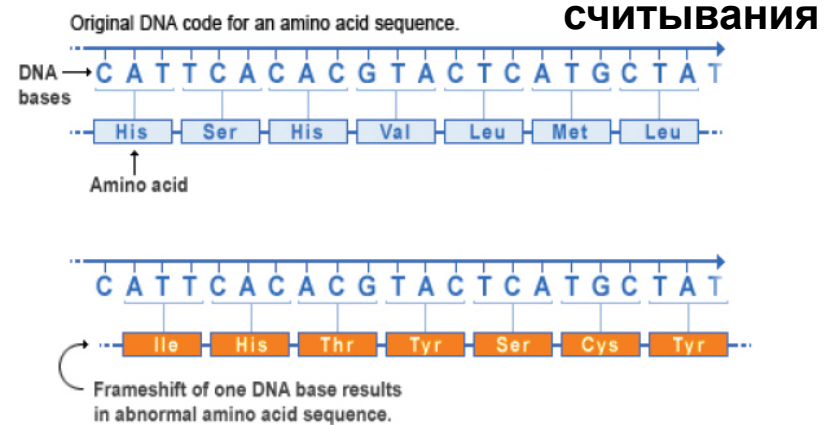


U.S. National Library of Medicine

Nucleotide repeats are short DNA sequences that are repeated a number of times in a row. For example, a trinucleotide repeat is made up of 3-base-pair sequences, and a tetranucleotide repeat is made up of 4-base-pair sequences. A repeat expansion is a mutation that increases the number of times that the short DNA sequence is repeated. This type of mutation can cause the resulting protein to function improperly.

## FRAMESHIFT MUTATION

## Изменение рамки



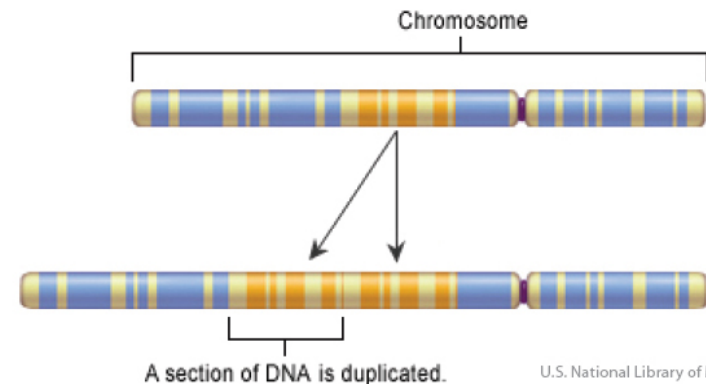
U.S. National Library of Medicine

This type of mutation occurs when the addition or loss of DNA bases changes a gene's reading frame. A reading frame consists of groups of 3 bases that each code for one amino acid. A frameshift mutation shifts the grouping of these bases and changes the code for amino acids. The resulting protein is usually nonfunctional. Insertions, deletions, and duplications can all be frameshift mutations.

## DUPLICATION

## Дублирование

A duplication consists of a piece of DNA that is abnormally copied one or more times. This type of mutation may alter the function of the resulting protein.



U.S. National Library of Medicine

Source: <http://ghr.nlm.nih.gov/handbook/mutationsanddisorders/possiblemutations>

# **GWAS**

**Genome-Wide Association Studies**

**Полногеномный анализ ассоциаций**

# **GDA<sub>s</sub>**

**Gene-Disease Associations**

**Ассоциации ген – болезнь**

# **VDA<sub>s</sub>**

**Variant-Disease Associations**

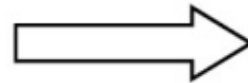
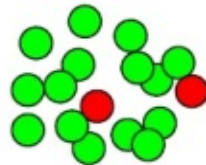
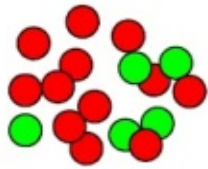
**Ассоциации варианты (SNP) – болезнь**

# GWAS

**Выявление «генов предрасположенности» к заболеванию проводится путем сопоставлений частот генотипов у больных и здоровых**

Группа больных

Контроль (здоровые)



● - генотип, указывающий на предрасположенность к заболеванию.

$P_{\text{больные}}$

$\gg$

$P_{\text{контроль}}$

**OR** – количественная мера предрасположенности (**Odd Ratio**), показывает во сколько раз повышена вероятность заболеть для носителя «плохого» генотипа

$$\text{OR} = \frac{P_{\text{больные}} (1 - P_{\text{контроль}})}{P_{\text{контроль}} (1 - P_{\text{больные}})}$$



# Международный проект HapMap

International  
HapMap  
Project



International HapMap Project

[Home](#) | [About the Project](#) | [Data](#) | [Publications](#)

2002 — 2010

[中文](#) | [English](#) | [Français](#) | [日本語](#) | [Yoruba](#)

The International HapMap Project is a partnership of scientists and funding agencies from Canada, China, Japan, Nigeria, the United Kingdom and the United States to develop a public resource that will help researchers find genes associated with human disease and response to pharmaceuticals. See "[About the International HapMap Project](#)" for more information.

## Project Information

[About the Project](#)

[HapMap Publications](#)

[HapMap Tutorial](#)

[HapMap Mailing List](#)

[HapMap Project Participants](#)

[HapMap Mirror Site in Japan](#)

## Project Data

[HapMap Genome Browser \(B35 - full data set\)](#)

[HapMap Genome Browser \(B36 - genotypes & frequencies only\)](#)

[HapMart](#)

[Bulk Data Download](#)

[Data Freezes for Publication](#)

[ENCODE Project](#)

[Guidelines For Data Use](#)

## Useful Links

## News

- 2008-02-21: **Incorrect position for merged SNPs in rel #23**

The position of ~24,500 SNPs was inadvertently entered incorrectly in HapMap release #23 bulk files (genotypes and frequencies). A complete list of affected SNPs can be found [here](#). Errors are being corrected and new genotypes and frequency files will be made available shortly under HapMap release #23a.

- 2008-02-06: **HapMap inferred genotypes (rel#22)**

Genotypes were inferred for 73 CEPH children using the method of Burdick et al. (Nat Genet 38:1002-4) and based on the genotypes of 60 CEPH individuals in the same families. Data and list of CEPH samples are available [here](#).

- 2008-01-31: **HapMap Public Release #23**

Genotypes and frequency data for this release are now available for [bulk download](#). These datasets include SNPs in the Affymetrix 6.0 array, as well as SNPs excluded from release #22 which have now been merged into new rs# identifiers in dbSNP 126 (NCBI b36). Lists of merged SNPs may be found [here](#). Please refer to the [release notes](#) for additional information.

- 2007-12-20: **Official release of HapMap Phased Haplotypes in NCBI b36 coordinates**

HapMap release #22 phased autosomes are now available for [bulk download](#).

- 2007-12-12: **Genotype imputation using MACH1 software now available on HapMap Genome Browser**

Impute genotypes for all HapMap SNPs in a given region by providing a subset of genotypes on HapMap SNPs. Browse a region of interest, upload your own

## Карта гаплотипа генома человека

Описание общих закономерностей наследственной генетической изменчивости людей

Генетические варианты, влияющие на здоровье, болезни и реакции на лекарственные препараты и факторы окружающей среды

# 1000 Genomes

IGSR: The International Genome Sample Resource

Supporting open human variation data

2008 — 2015 — .....

[Home](#) [About](#) [Data](#) [Portal](#) [Analysis](#) [Contact](#) [Browser](#) [FAQ](#)

Search IGSR



## IGSR and the 1000 Genomes Project



## Links

[Announcements](#)

[IGSR Sample Collection Principles](#)

[1000 Genomes Project Publications](#)

[File formats](#)

[Software tools](#)

[Download data](#)

<http://www.internationalgenome.org/>

2,504 генома и результаты их выравниваний  
по референсному геному GRCh38

# Референсный геном человека

Full Report ▾

Send to: ▾

[Download Assembly](#)

See [Genome](#) information for **Homo sapiens**

There are 917 assemblies for this organism  
[See more](#)

**Access the data**

- Genome Data Viewer
- RefSeq Annotation Report
- BLAST the assembly
- Full sequence report
- Statistics report
- Regions report
- FTP directory for RefSeq assembly

**GRCh38.p13**  
**Description:** Genome Reference Consortium Human Build 38 patch release 13 (GRCh38.p13)  
**Organism name:** [Homo sapiens \(human\)](#)  
**BioProject:** [PRJNA31257](#)  
**Submitter:** Genome Reference Consortium  
**Date:** 2019/02/28  
**Assembly type:** haploid-with-alt-loci  
**Release type:** patch  
**Assembly level:** Chromosome  
**Genome representation:** full  
**RefSeq category:** reference genome  
**GenBank assembly accession:** GCA\_000001405.28 (latest)  
**RefSeq assembly accession:** GCF\_000001405.39 (latest)  
**RefSeq assembly and GenBank assembly identical:** no ([hide details](#))

- Only in GenBank: 1 unplaced scaffold (in primary assembly-unit)
- Data displayed for RefSeq version

IDs: 2334371 [UID] 8687898 [GenBank] 8765528 [RefSeq]

**History** ([Show revision history](#))


**Comment**

The DNA sequence is composed of genomic sequences, primarily finished clones that were sequenced as part of the Human Genome Project (HGP).


**Всего доступно  
917 различных сборок**

**Сборка GRCh38.p13 — февраль 2019  
Genome Reference Consortium Human Build 38 patch release 13**

# HGMD — Human Gene Mutation Database



**The Human Gene Mutation Database**  
at the Institute of Medical Genetics in Cardiff



Home Search help Statistics New genes What is new Background Publications Contact Register Login L SDBs Other

2007 — .....

The Human Gene Mutation Database (HGMD®) represents an attempt to collate all known (published) gene lesions responsible for human inherited disease and is maintained in Cardiff by D.N. Cooper, E.V. Ball, P.D. Stenson, A.D. Phillips, K. Evans, S. Heywood, M.J. Hayden, M.M. Chapman, M.E. Mort, L. Azevedo and D.S. Millar.

Get HGMD Professional

\*Please note that this less up-to-date public version of our database is freely available only to [registered](#) users from academic institutions/non-profit organisations. All commercial users are required to purchase a license from QIAGEN®, our commercial partner. A license to [HGMD Professional](#) is available to both commercial and academic/non-profit users wishing to access the most up-to-date version of the database (visit QIAGEN® to request a [free trial](#) of HGMD Professional). Read more about how HGMD is [funded](#). You may not copy, store or re-distribute HGMD data without express written permission (i) from the curators or (ii) via your license agreement. Copyright © Cardiff University 2017. All rights reserved.

Register for Public Version

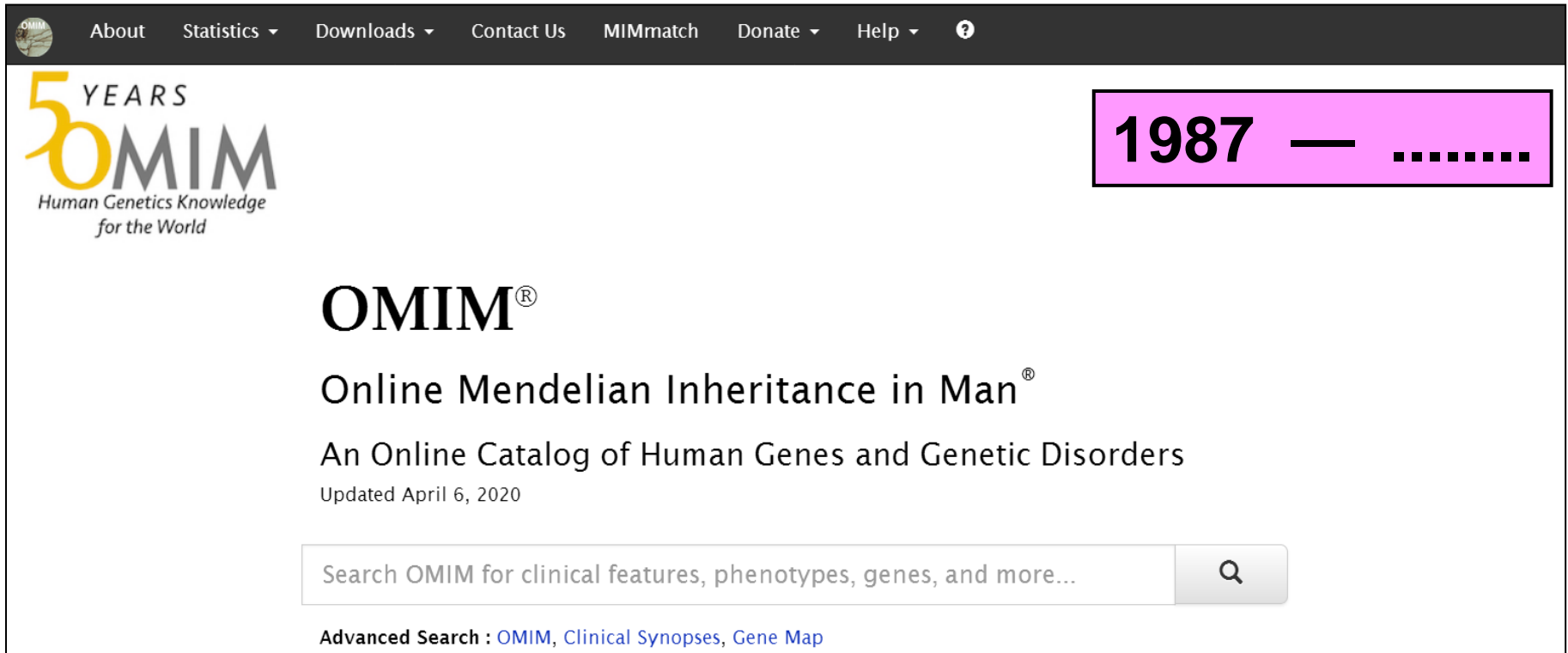
Table:	Description:	Public entries: <small>This site. Academic/non-profit users only</small>	Total entries: <small>HGMD Professional 2019.4</small>
<b>Mutation totals (as of 2020-04-07)</b>		<b>189186</b>	<b>275716</b>
Gene symbol	The gene description, gene symbol (as recommended by the HUGO Nomenclature Committee) and chromosomal location is recorded for each gene. In cases where a gene symbol has not yet been made official, a provisional symbol has been adopted which is denoted by lower-case letters.	7677	10902
cDNA sequence	cDNA reference sequences are provided, numbered by codon.	7729	11079
Genomic coordinates	Genomic (chromosomal) coordinates have been calculated for missense/nonsense, splicing, regulatory, small deletions, small insertions and small indels.	0	250578
HGVS nomenclature	Standard HGVS nomenclature has been obtained for missense/nonsense, splicing, regulatory, small deletions, small insertions and small indels.	0	250862
Missense/nonsense	Single base-pair substitutions in coding regions are presented in terms of a triplet change with an additional flanking base included if the mutated base lies in either the first or third position in the triplet.	106004	159705

<http://www.hgmd.cf.ac.uk/ac/>

Regulatory	mutation relative to the transcriptional initiation site, initiation codon, polyadenylation site or termination codon is given.	3544	4575
Small deletions	Micro-deletions (20 bp or less) are presented in terms of the deleted bases in lower case plus, in upper case, 10 bp DNA sequence flanking both sides of the lesion. The numbered codon is preceded in the given sequence by the caret character (^).	28155	39822
Small insertions	Micro-insertions (20 bp or less) are presented in terms of the inserted bases in lower case plus, in upper case, 10 bp DNA sequence flanking both sides of the lesion. The numbered codon is preceded in the given sequence by the caret character (^).	11745	16881
Small indels	Micro-indels (20 bp or less) are presented in terms of the deleted/inserted bases in lower case plus, in upper case, 10 bp DNA sequence flanking both	2676	2676

Описание 275,716 мутаций ядерных генов,  
 приводящих к наследственным заболеваниям

# OMIM — Online Mendelian Inheritance in Man



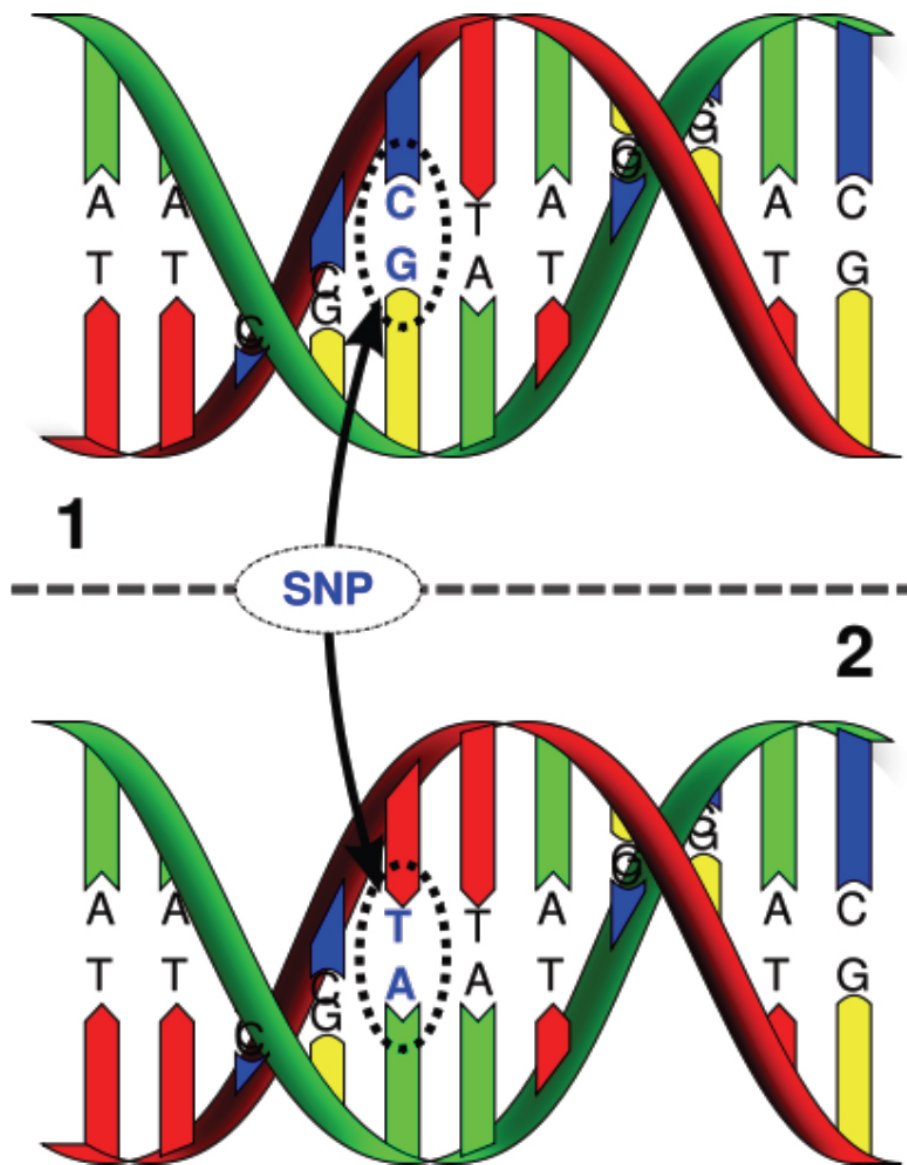
The screenshot shows the OMIM website homepage. At the top is a dark navigation bar with links: About, Statistics, Downloads, Contact Us, MIMmatch, Donate, Help, and a question mark icon. Below the navigation bar is a white header area. On the left is the OMIM 50th anniversary logo, which says '50 YEARS OMIM Human Genetics Knowledge for the World'. On the right is a pink box containing the year '1987' followed by a horizontal line and a series of dots. Below the header is the main content area. It features the OMIM logo, the text 'Online Mendelian Inheritance in Man', and 'An Online Catalog of Human Genes and Genetic Disorders'. Below this is the update date 'Updated April 6, 2020'. A search bar is present with the placeholder text 'Search OMIM for clinical features, phenotypes, genes, and more...'. Below the search bar is a link for 'Advanced Search : OMIM, Clinical Synopses, Gene Map'.

<https://www.omim.org/>

OMIM is supported by a grant from NHGRI, licensing fees, and [generous contributions from people like you.](#)

**Описание 6,632 генетических заболеваний и отклонений  
и 4,252 генов, их вызывающих  
Фенотипические проявления генетических синдромов  
Генетические карты**

# Однонуклеотидный полиморфизм



~10 млрд. SNP

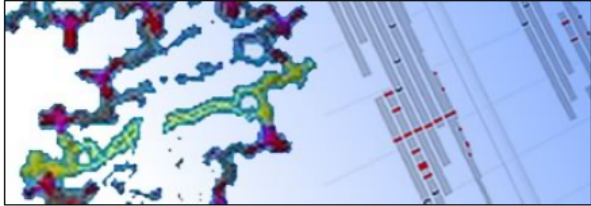
# Базы данных GWAS — dbSNP

NCBI Resources How To Sign in to NCBI

dbSNP SNP Search

Advanced

**1999 — .....**



**dbSNP**

dbSNP contains human single nucleotide variations, microsatellites, and small-scale insertions and deletions along with publication, population frequency, molecular consequence, and genomic and RefSeq mapping information for both common variations and clinical mutations.

**Getting Started**

- [dbSNP 20th Anniversary](#)
- [Overview of dbSNP](#)
- [About Reference SNP \(rs\)](#)
- [Factsheet](#)
- [Entrez Updates \(February 5, 2020\)](#)

**Submission**

- [How to Submit](#)
- [Hold Until Published \(HUP\) Policies](#)
- [Submission Search](#)

**Access Data**

- [Variation Services API](#)
- [FTP Download](#)
- [Tutorials on GitHub](#)

**https://www.ncbi.nlm.nih.gov/snp**

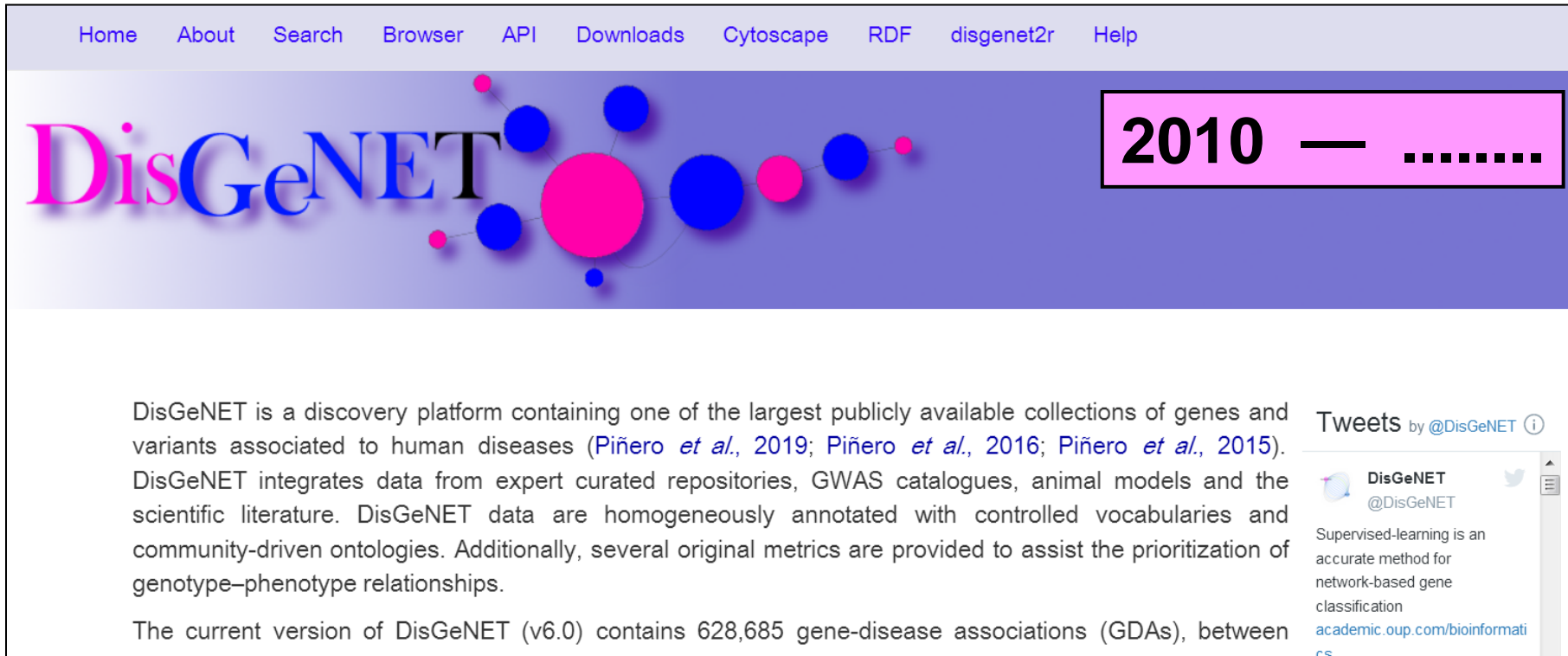
**How to Search dbSNP. Additional search terms are [here](#).**

All of dbSNP (then use filters on results page)	<a href="#">all[sb]</a>
dbSNP RefSNP ID	Single: <a href="#">328</a> ; Multiple <a href="#">328,226,200</a>
Gene	Gene symbol <a href="#">PTEN[Gene Name]</a> or gene ID <a href="#">4023[Gene ID]</a>

Genomic location of a single position or range on GRCh38. See the

**Более 2 млрд. аннотированных SNP человека**

# Базы данных GWAS — DisGeNET



Home About Search Browser API Downloads Cytoscape RDF disgenet2r Help

## DisGeNET

2010 — .....

DisGeNET is a discovery platform containing one of the largest publicly available collections of genes and variants associated to human diseases (Piñero *et al.*, 2019; Piñero *et al.*, 2016; Piñero *et al.*, 2015). DisGeNET integrates data from expert curated repositories, GWAS catalogues, animal models and the scientific literature. DisGeNET data are homogeneously annotated with controlled vocabularies and community-driven ontologies. Additionally, several original metrics are provided to assist the prioritization of genotype–phenotype relationships.

The current version of DisGeNET (v6.0) contains 628,685 gene-disease associations (GDAs), between

Tweets by @DisGeNET

DisGeNET @DisGeNET

Supervised-learning is an accurate method for network-based gene classification  
[academic.oup.com/bioinformatics](http://academic.oup.com/bioinformatics)

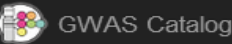
<http://www.disgenet.org/>

The information in DisGeNET can be accessed in several ways:

- 628,685 GDAs между 17,549 генами и 24,166 заболеваниями и отклонениями
- 210,498 VDAs между 117,337 SNPs и 10,358 заболеваниями и фенотипами

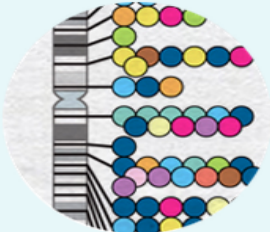


# GWAS Catalog



GWAS Catalog

Home Diagram Download Documentation About EMBL-EBI NIH National Human Genome Research Institute



## GWAS Catalog

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

2008 — .....

Search the catalog

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

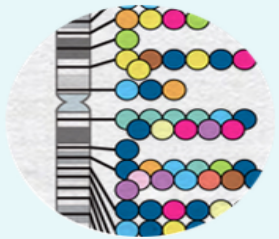
NEW! We can now accept direct submissions of summary statistics for published GWAS through our new [submission page](#)! See the [documentation](#) for detailed instructions.

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

Documentation Diagram Ancestry

179,364 ассоциации

# GWAS Catalog



## GWAS Catalog

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



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

NEW! We can now accept direct submissions of summary statistics for published GWAS through our new [submission page](#)! See the [documentation](#) for detailed instructions.

### Download

Download a full copy of the GWAS Catalog in spreadsheet format as well as current and older versions of the GWAS diagram in SVG format.

### Summary statistics

Documentation and access to full summary statistics for GWAS Catalog studies where available.

### Submit

Submit summary statistics to GWAS Catalog.

### Documentation

Including FAQs, our curation process, training materials, related resources, a list of abbreviations and API documentation.

### Diagram

Explore an interactive visualisation of all SNP-trait associations with genome-wide significance ( $p \leq 5 \times 10^{-8}$ ).

### Ancestry

An introduction to our ancestry curation process.

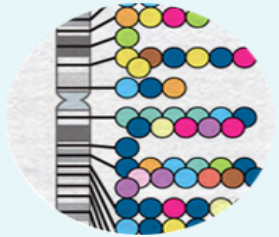
# GWAS Catalog

	A	H	K	M	O	U	Y	AA	AB	AE
1	DATE ADDED	DISEASE/TRAIT	REGION	CHR_POS	MAPPED_GENE	STRONGEST SNP-RISK ALLELE	CONTEXT	RISK ALLELE FREQUENCY	P-VALUE	OR or BETA
494	2008-06-16	Type 2 diabetes	16q12.2	53782363	FTO	rs8050136-A	intron_variant	0.4	7.00E-14	1.23
495	2008-06-16	Type 2 diabetes	3q27.2	185793899	IGF2BP2	rs4402960-T	intron_variant	0.32	9.00E-16	1.14
496	2008-06-16	Type 2 diabetes	8q24.11	117172544	SLC30A8	rs13266634-C	missense_variant	0.69	5.00E-08	1.12
497	2008-06-16	Type 2 diabetes	6p22.3	20668203	CDKAL1	rs10946198-C	intron_variant	0.32	1.00E-08	1.16
498	2008-06-16	Type 2 diabetes	10q23.13	92702202	HNFX - 3_RNA	rs5014180-C	intergenic_variant	0.57	5.00E-06	1.13
499	2008-06-16	Type 2 diabetes	10q25.2	112994529	TCF7L2	rs7901695-C	intron_variant	NR	1.00E-48	1.37
500	2008-06-16	Type 2 diabetes	9p21.3	22029548	AL359922.1, CDKN1A	rs564398-T	3_prime_UTR_variant	0.56	1.00E-06	1.13
501	2008-06-16	Type 2 diabetes	11p15.1	17387083	KCNJ11	rs5245-C	missense_variant	NR	5.00E-11	1.14
502	2008-06-16	Type 2 diabetes	3p25.2	12351626	PPARG	rs1091262-C	missense_variant	NR	2.00E-06	1.14
503	2008-06-16	Type 2 diabetes	9p21.3	22134095	CDKN2B-AS1 - AL1	rs10811634-T	intergenic_variant	0.83	5.00E-06	1.19
525										
529										
530										
531										
532										
533										
534										
535										
661										
662										
663										
664										
665										
666										
667										
668										
669										
670	2011-05-24	Type 2 diabetes	1q32.2	207478831	CR2	rs17045328-G	intron_variant	0.3	7.00E-06	1.38
671	2011-05-24	Type 2 diabetes	6q13	70579486	SDHAF4	rs1048886-G	missense_variant	0.18	3.00E-08	1.54
672	2011-05-24	Type 2 diabetes	6q24.1	139952510	AL050338.2	rs642858-A	intron_variant	0.4	2.00E-06	1.35
673	2011-05-24	Type 2 diabetes	7q22.1	100892456	ACHE	rs7636-A	missense_variant	0.96	5.00E-06	1.95
674	2011-05-24	Type 2 diabetes	10q26.3	131149699	TCERG1L	rs10741243-G	missense_variant	0.3	1.00E-06	1.25
675	2011-05-24	Type 2 diabetes	3q12.3	102484201	ZPLD1 - AC063938	rs2063640-A	missense_variant	0.3	1.00E-06	1.25
676	2011-05-24	Type 2 diabetes	19q13.2	39090097	ACP7	rs472265-G	missense_variant	0.3	1.00E-06	1.25
677	2011-05-24	Type 2 diabetes	3q23	142712158	AC072028.1, PLS1	rs3773506-C	missense_variant	0.3	1.00E-06	1.25
779	2009-04-10	Type 2 diabetes	11p15.4	2837316	KCNQ1	rs2237897-C	missense_variant	0.3	1.00E-06	1.25
780	2009-04-10	Type 2 diabetes	6p22.3	20657634	CDKAL1	rs4712524-G	intron_variant	0.42	3.00E-10	1.22
781	2009-04-10	Type 2 diabetes	3q27.2	185812502	IGF2BP2	rs6769511-C	intron_variant	0.32	1.00E-09	1.23
1416	2008-09-09	Type 2 diabetes	2q12.1	105221141	LINC01918 - GPR45	rs6712932-?	intergenic_variant	NR	6.00E-06	1.52

1. Хромосома 8, длинное плечо, область 2, бэнд 4, суб-бэнд 11
2. Позиция — 117172544
3. Ген SLC30A8 — цинковый транспортер 8, ZNT8
4. Рисксовая аллель С — цитозин
5. Тип мутации — миссенс
6. Общая частота встречаемости рисксовой аллели 0.68 %
7. Частота встречаемости рисксовой аллели у больных —  $5 \cdot 10^{-8}$
8. Мера предрасположенности (Odd Ratio) — 1.12 раза

Сахарный диабет типа 2  
1,705 SNP

# GWAS Catalog



## GWAS Catalog

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



Examples: [breast carcinoma](#), [rs7329174](#), [Yao](#), [2q37.1](#), [HBS1L](#), [6:16000000-25000000](#)

**NEW!** We can now accept direct submissions of summary statistics for published GWAS through our new [submission page](#)! See the [documentation](#) for detailed instructions.

### Download

Download a full copy of the GWAS Catalog in spreadsheet format as well as current and older versions of the GWAS diagram in SVG format.

### Summary statistics

Documentation and access to full summary statistics for GWAS Catalog studies where available.

### Submit

Submit summary statistics to GWAS Catalog.

### Documentation

Including FAQs, our curation process, training materials, related resources, a list of abbreviations and API documentation.

### Diagram

Explore an interactive visualisation of all SNP-trait associations with genome-wide significance ( $p \leq 5 \times 10^{-8}$ ).

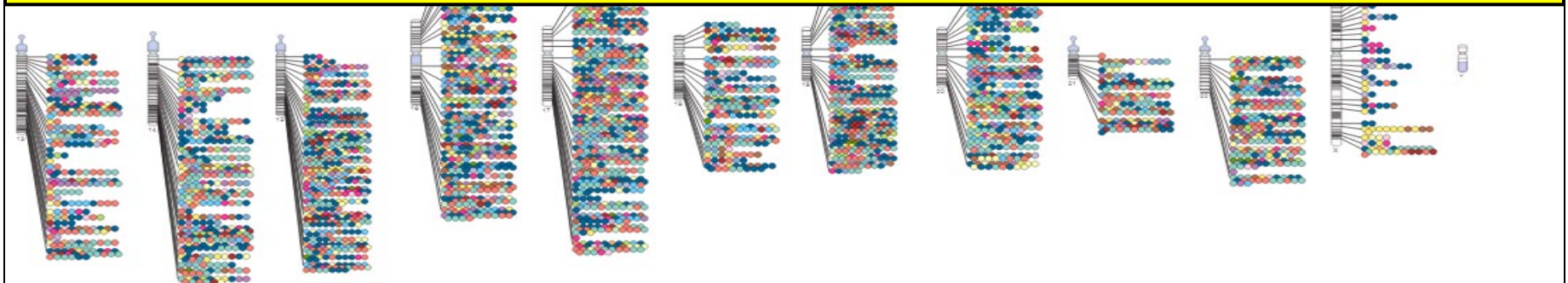
### Ancestry

An introduction to our ancestry curation process.

# GWAS Diagram



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



# GWAS Diagram

Filter the diagram

Filter by trait

Clear

Apply

Show SNPs for

Digestive system disease 372

Cardiovascular disease 1131

Metabolic disease 744

Immune system disease 2090

Nervous system disease 2031

Liver enzyme measurement 170

Lipid or lipoprotein measurement 1190

Inflammatory marker measurement 474

Hematological measurement 5015

Body weights and measures 2622

Download diagram



feedback

# GWAS Diagram

Filter the diagram

Filter by trait

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Show SNPs for

Digestive system disease 372

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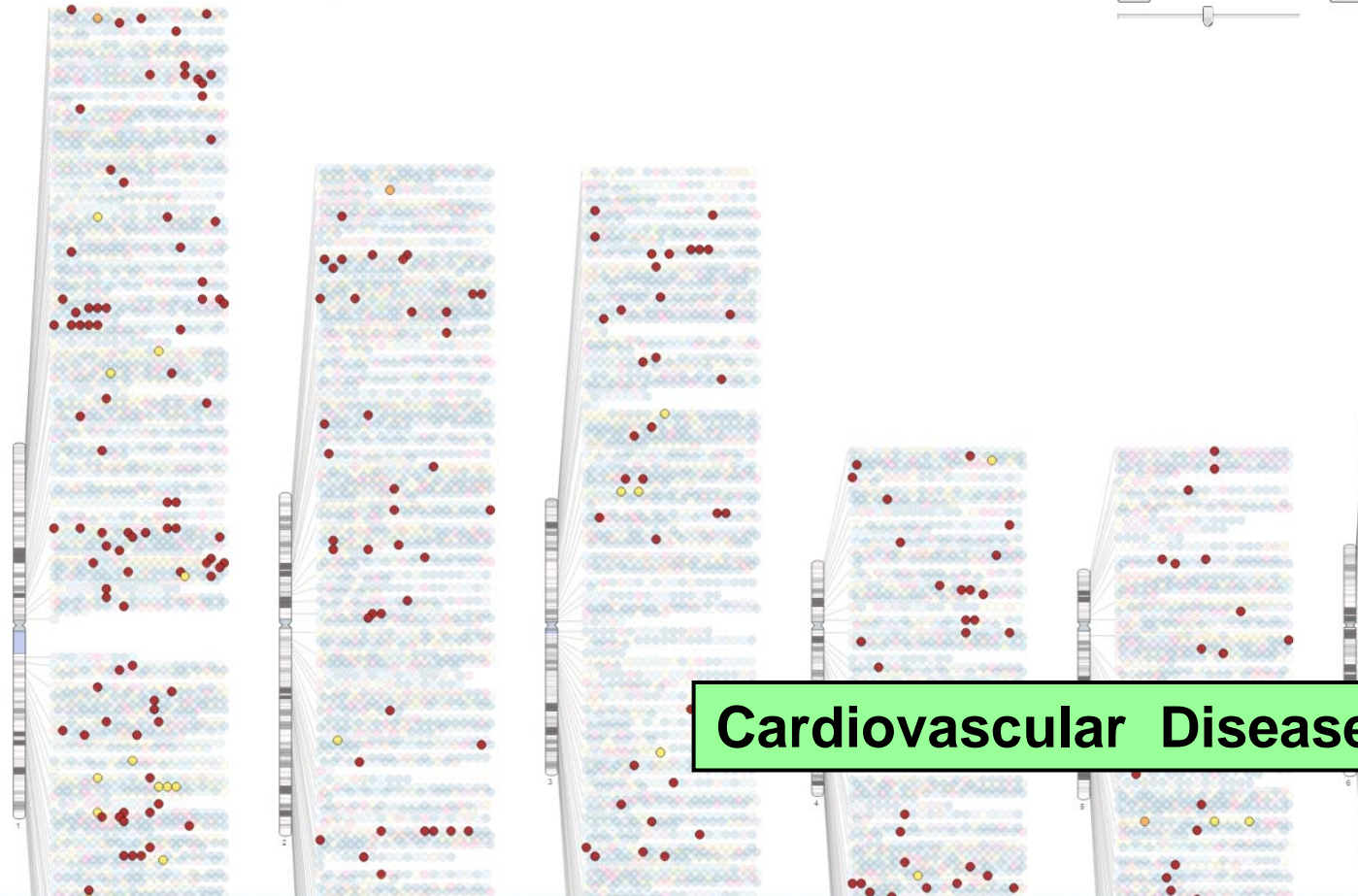
Inflammatory marker measurement 474

Hematological measurement 5015

Body weights and measures 2622

SNP-trait associations for "Cardiovascular disease" on the diagram: 1219

Download diagram



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# GWAS Diagram

Filter the diagram

Filter by trait

Clear

Apply

Show SNPs for

Digestive system disease 372

Cardiovascular disease 1131

Metabolic disease 744

Immune system disease 2090

Nervous system disease 2031

Liver enzyme measurement 170

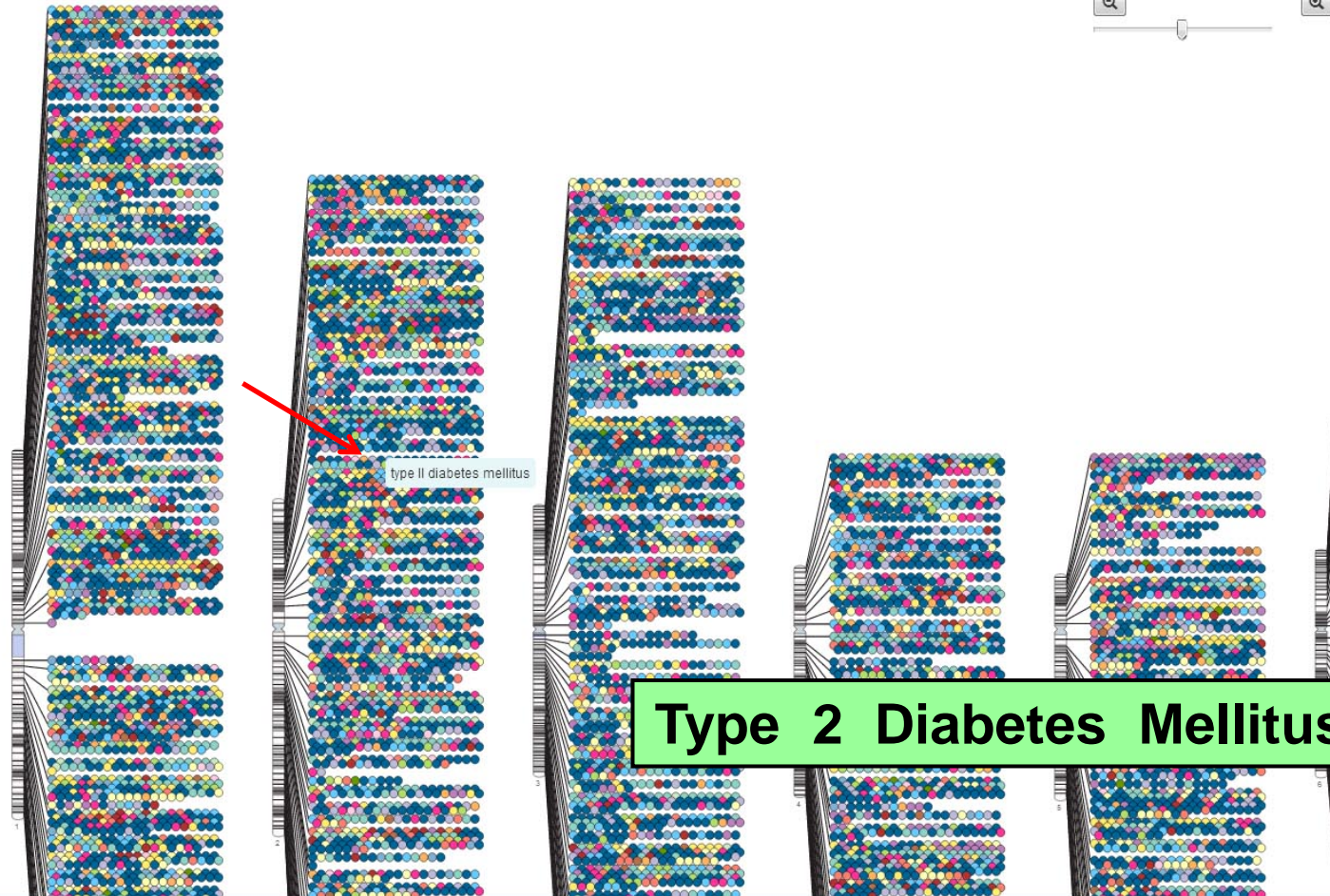
Lipid or lipoprotein measurement 1190

Inflammatory marker measurement 474

Hematological measurement 5015

Body weights and measures 2622

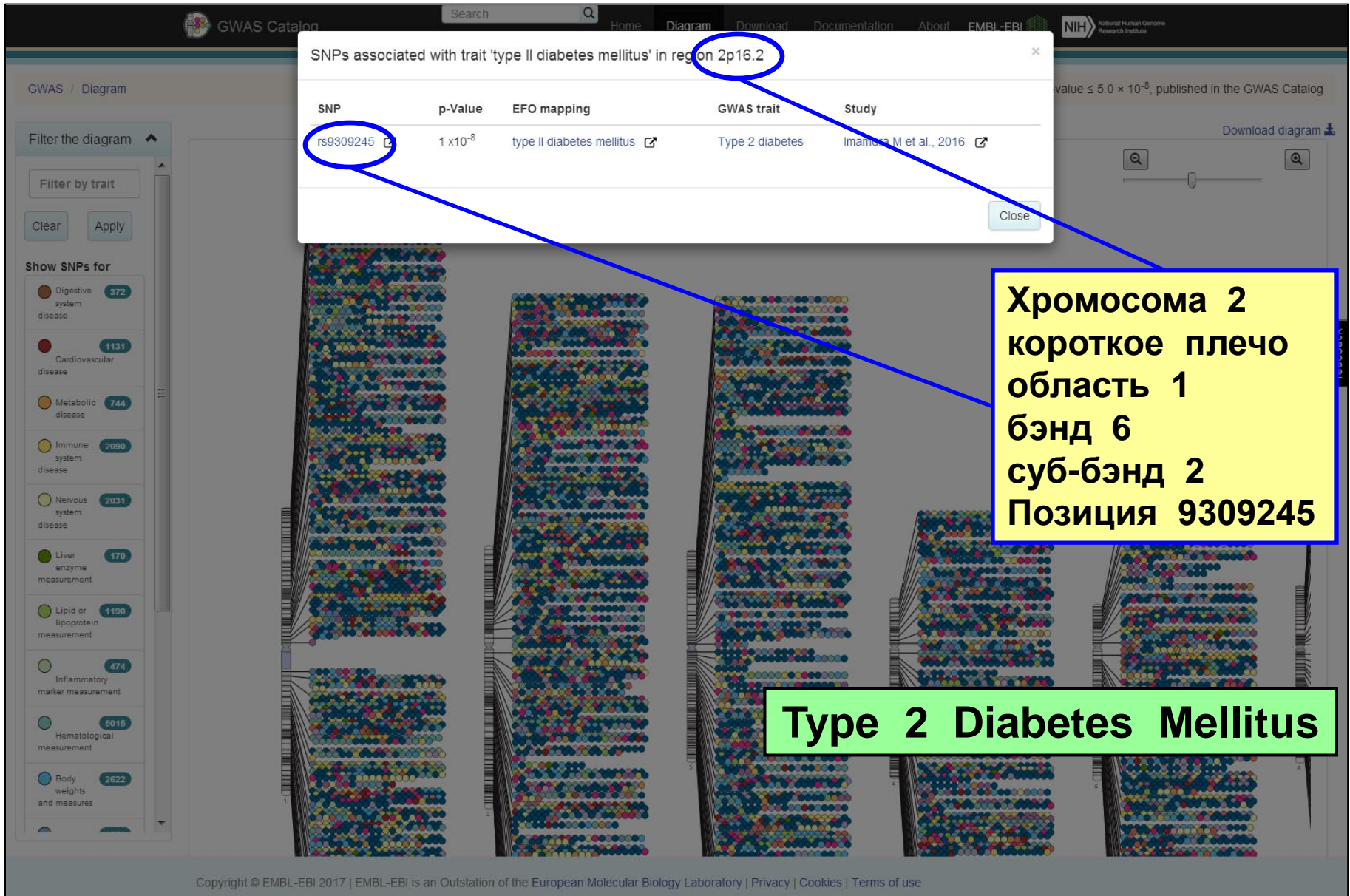
Download diagram



feedback



# GWAS Diagram



# GWAS

**Table 1** GWA studies showing SNPs association with cancer.

Disease	Chromosomal location	Gene	Strongest SNPs-risk allele	P-value	Reference
Breast cancer	10q26.13	<i>FGFR2</i>	rs2981582-G	$2 \times 10^{-76}$	[11,36-38]
	16q12.1	<i>TNCR9/LOC643714</i>	rs3803662-C	$1 \times 10^{-36}$	
	5q11.2	<i>MAP3K1</i>	rs889312-A	$7 \times 10^{-20}$	
	11p15.5	<i>LSP1</i>	rs3817198-T	$3 \times 10^{-9}$	
	8q24.21	Intergenic	rs13281615-T	$5 \times 10^{-12}$	
	2q34	<i>ERBB4</i>	rs13393577-C/T	$8.8 \times 10^{-14}$	
	10q26.13	<i>FGFR2</i>	rs1219648-G	$1 \times 10^{-10}$	
	2q35	Intergenic	rs13387042-A	$1 \times 10^{-13}$	
Basal cell carcinoma	1q42.13	<i>RHOA</i>	rs801114-G	$6 \times 10^{-12}$	[39-44]
	1p36.13	<i>PADI4, PADI6, RCC2, ARHGEF10L</i>	rs7538876-A	$4 \times 10^{-12}$	
	12q12.13	<i>KRT5</i>	rs11170164-A	$2 \times 10^{-9}$	
	9p21	<i>CDKN2A/B</i>	rs2151280-C	$7 \times 10^{-9}$	
	8q24.21	Intergenic	rs16901979-A	$1 \times 10^{-12}$	
	8q24.21	Intergenic	rs6983267-G	$9 \times 10^{-13}$	
	17q12	<i>TCF2</i>	rs4430796-A	$1 \times 10^{-11}$	
	11p15	<i>IGF2, IGF2A, INS, TH</i>	rs7127900-A	$3 \times 10^{-33}$	
Colorectal cancer	8q24.21	<i>ORF, DQ515897</i>	rs10505477-A	$3 \times 10^{-11}$	[45,46]
	18q21.1	<i>SMAD7</i>	rs4939827-T	$1 \times 10^{-12}$	
Lung cancer	15q25.1	<i>CHRNA3, CHRNA5, CHRNB4, PSMA4, LOC123688</i>	rs8034191-C	$5 \times 10^{-20}$	[47]
	6p22.1	<i>TRNA-UGC</i>	rs4324798-A	$2 \times 10^{-8}$	[48]
Melanoma	20q11.22	<i>CDC91LI</i>	rs910873-T	$1 \times 10^{-15}$	[49,50]
	22q13.1	Intergenic	rs2284063-G	$2 \times 10^{-9}$	
Neuroblastoma	6p22.3	<i>FLJ22536, FLJ44180</i>	rs6939340-G	$9 \times 10^{-15}$	[51,52]
	2q35	<i>BARD1</i>	rs6435862-G	$9 \times 10^{-18}$	
Ovarian cancer	9p22.2	Intergenic	rs3814113-T	$5 \times 10^{-19}$	[53]
Thyroid cancer	9q22.23	<i>F</i>	rs513-A	$2 \times 10^{-27}$	[15]
	14q13.3	<i>N</i>	rs289-T	$2 \times 10^{-9}$	

**Опухолевые заболевания**

# GWAS

**Table 2** SNPs significantly associated with infectious disease phenotypes in genome-wide studies.

Disease	SNP location	Strongest SNPs-risk allele	<i>P</i> -value	Reference
Creutzfeldt–Jakob disease	PRNP	rs1799990	$2.0 \times 10^{-27}$	[54]
Dengue shock Syndrome	MICB	rs3132468	$4.4 \times 10^{-11}$	[55]
Hepatitis B	HLA-DPA1	rs3077	$2.3 \times 10^{-38}$	[56]
	HLA-DPB1	rs9277535	$6.3 \times 10^{-39}$	
Hepatitis C	IL28B	rs8099917	$6.1 \times 10^{-9}$	[57]
HIV-1 and AIDS	HLA-C	rs9264942	$5.9 \times 10^{-32}$	[58–64]
	HLA-B, HCP5	rs2395029	$4.5 \times 10^{-35}$	
	HLA-B	rs2523608	$5.6 \times 10^{-10}$	
	HLA-C	rs9264942	$2.8 \times 10^{-35}$	
	MICA	rs4418214	$1.4 \times 10^{-34}$	
	HLA-B, HCP5	rs2395029	$9.7 \times 10^{-26}$	
	PSORS1C3	rs3131018	$4.2 \times 10^{-16}$	
	HLA-B	rs2523608	$8.9 \times 10^{-20}$	
	Intergenic	rs2255221	$3.5 \times 10^{-14}$	
	HLA-B	rs2523590	$1.7 \times 10^{-13}$	
	Intergenic	rs9262632	$1.0 \times 10^{-8}$	
	ZNRD1, RNF39	rs9261174	$1.8 \times 10^{-8}$	
	PARD3B	rs11884476	$3.4 \times 10^{-9}$	
	HLA-B, HCP5	rs2395029	$6.8 \times 10^{-10}$	
	CXCR6	rs2234358	$9.7 \times 10^{-10}$	
Leprosy	LACC1	rs3764147	$3.7 \times 10^{-54}$	[65]
	NOD2	rs9302752	$3.8 \times 10^{-40}$	
	RIPK2	rs42490	$1.4 \times 10^{-16}$	
	CCDC122	rs3088362	$1.4 \times 10^{-31}$	
	TNFSF15	rs6478108	$3.4 \times 10^{-21}$	
Meningococcal disease	CFH	rs1065489	$2.2 \times 10^{-11}$	[66]
Severe malaria			$3.7 \times 10^{-11}$	[67]
Tuberculosis			$6.8 \times 10^{-9}$	[68]

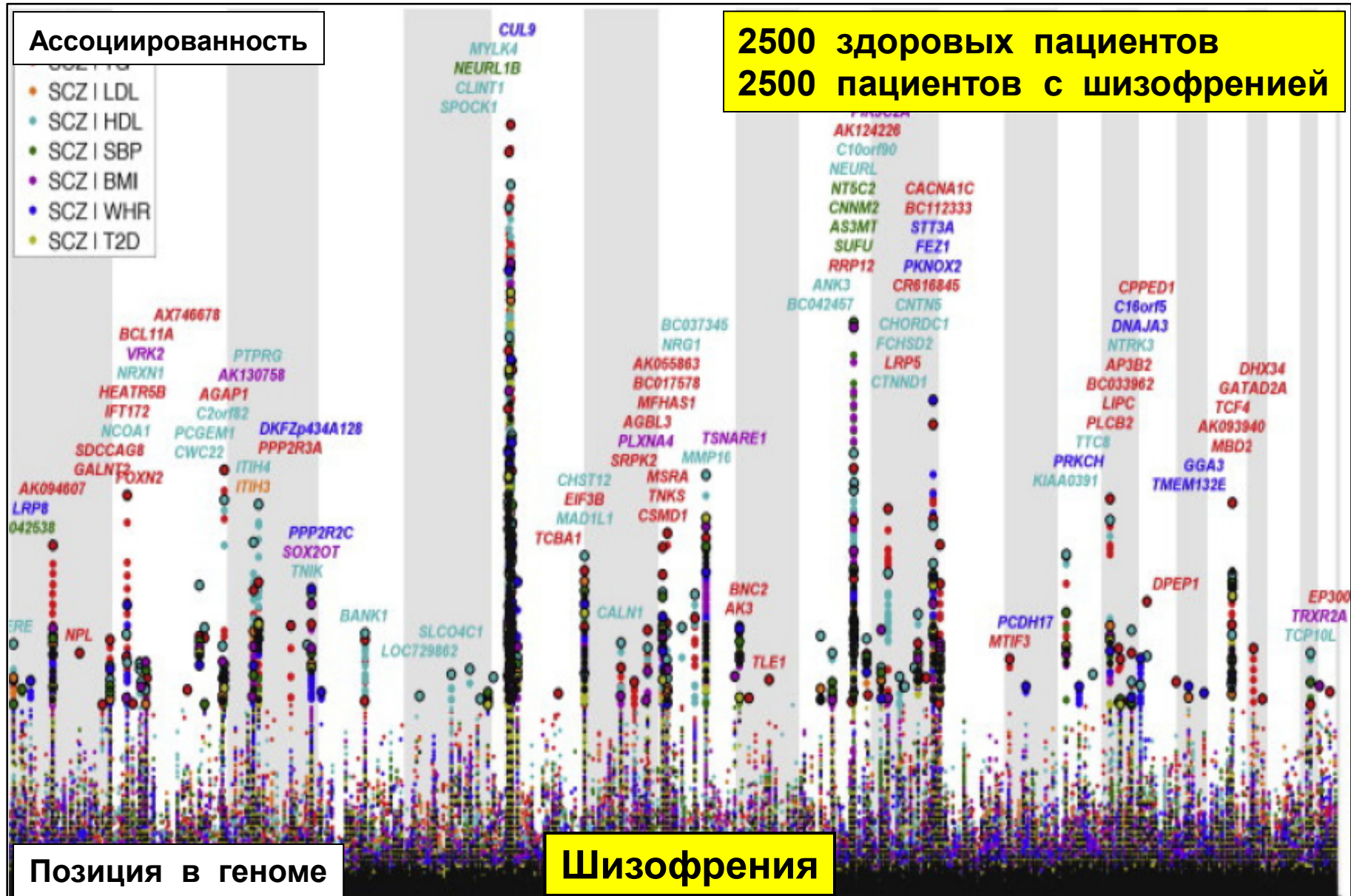
**Предрасположенность к инфекциям**

# GWAS

Ассоциированность

- SCZ | LDL
- SCZ | HDL
- SCZ | SBP
- SCZ | BMI
- SCZ | WHR
- SCZ | T2D

2500 здоровых пациентов  
2500 пациентов с шизофренией



Позиция в геноме

Шизофрения

**To be continued ...**

