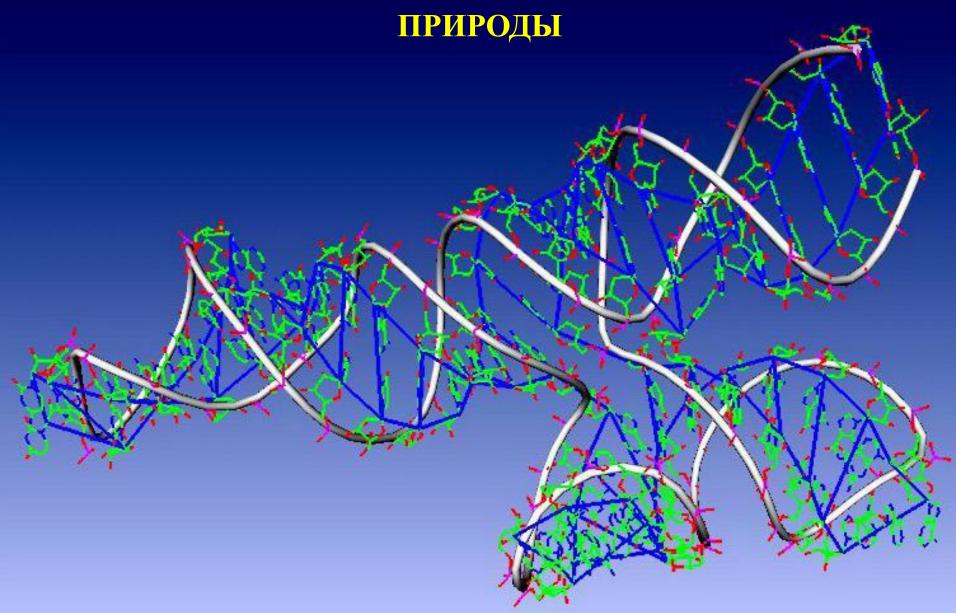
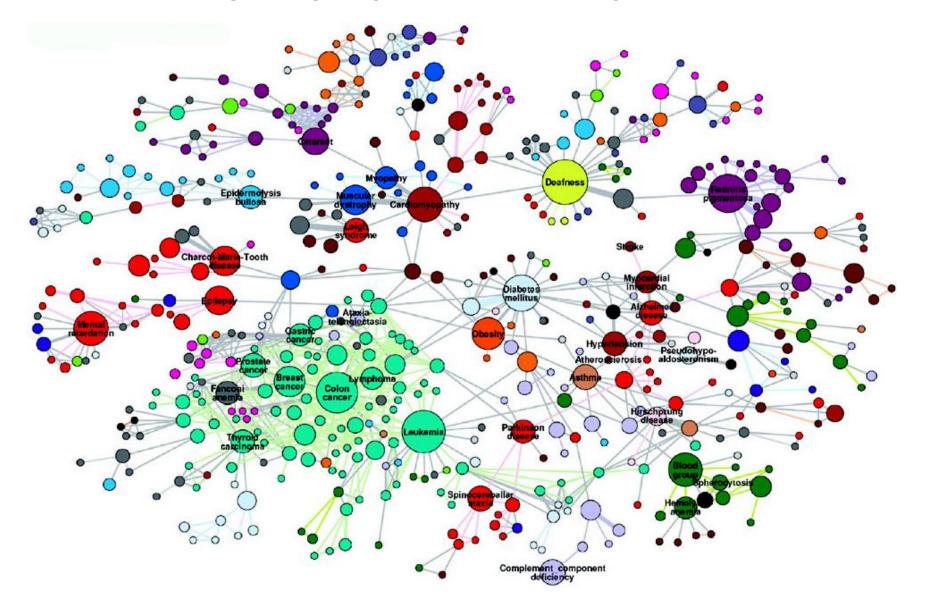
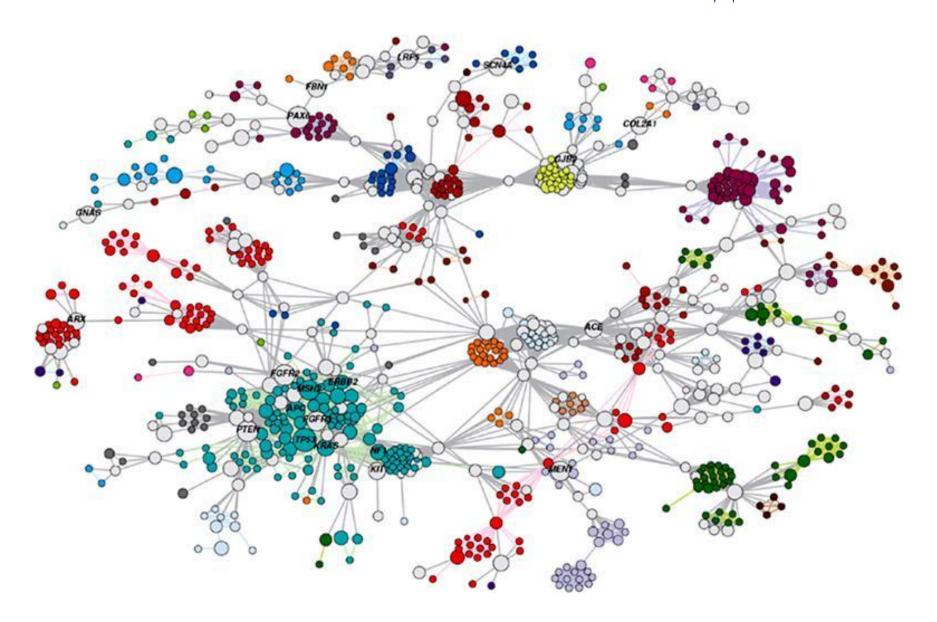
ИДЕНТИФИКАЦИЯ ГЕНОВ, ОТВЕТСТВЕННЫХ ЗА ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИМОНИИ

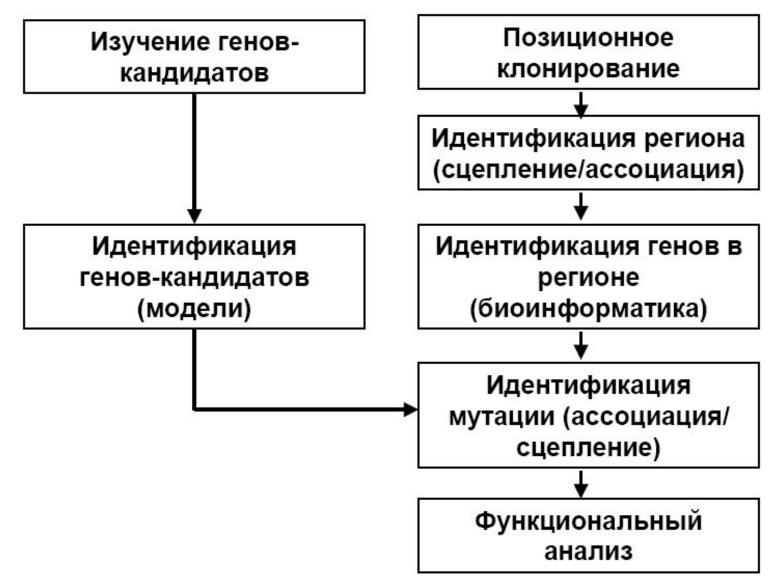


### СЕТЬ ЗАБОЛЕВАНИЙ ЧЕЛОВЕКА

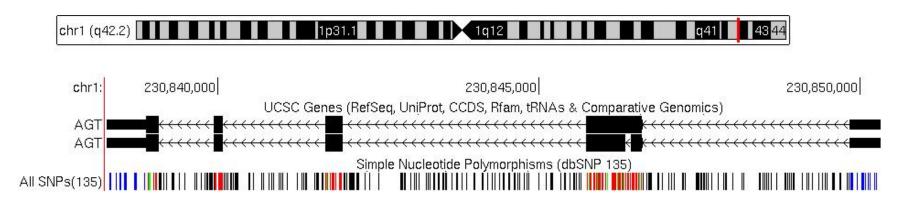


# СЕТЬ ГЕНОВ, ОТВЕТСТВЕННЫХ ЗА ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



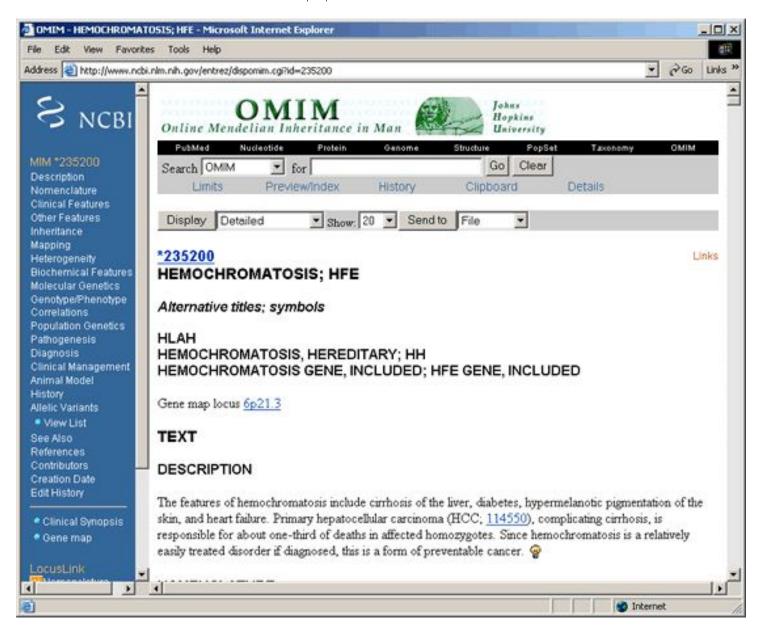


Основные стратегии генетического картирования



Ген ангиотензиногена

### БАЗА ДАННЫХ ОМІМ



http://www.ncbi.nlm.nih.gov/omim/

## БАЗА ДАННЫХ ОМІМ

	Аутосомные гены		Y- сцепленные	<b>Митохонд- риальные</b>	Всего
Количество описанных генов	13963	683	48	35	14729
Количество описанных признаков с установленной молекулярной природой	3950	287	4	28	4269
Количество описанных признаков с неустановленной молекулярной природой	1543	134	5	0	1682
Количество малоизученных признаков с предположительно менделевским типом наследования	1731	113	2	0	1846

Статистика по базе данных ОМІМ на 17 октября 2014 года

### БАЗА ДАННЫХ GeneCards



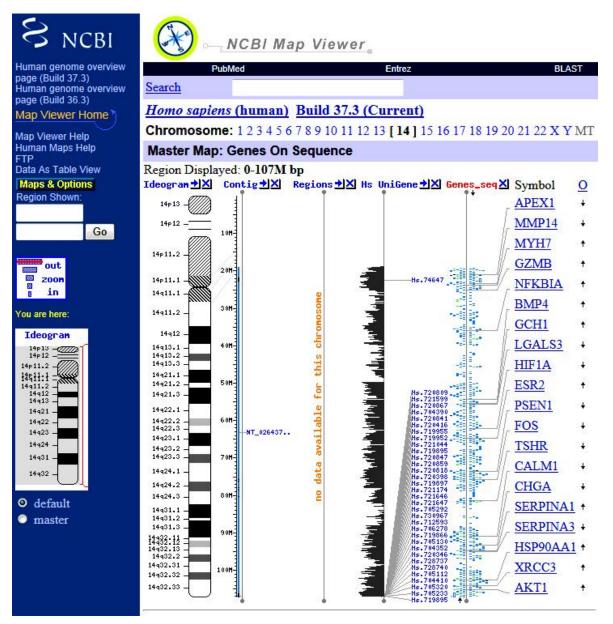
### Genes Associated with Diseases

Genetic diseases are caused by abnormalities in genes or chromosomes. Many genetic diseases are conditions present from before birth. Most genetic disorders are quite rare. A genetic disease may or may not be a heritable disorder. Some genetic diseases are passed down from the parents' genes, but others are frequently or always caused by new mutations or changes to the DNA. In other instances, the same disease, for example, some forms of cancer, may stem from an inherited genetic condition in some people, from new mutations in other people, and from non-genetic causes in still other people. There are more than 6,000 known single-gene (or monogenic) disorders, which occur in about 1 out of every 200 births. As their name suggests, these diseases are caused by a mutation in one gene. By contrast, polygenic disorders are caused by several genes, frequently in combination with environmental factors. Examples of genetic phenotypes include Alzheimer's disease, breast cancer, leukemia, Down syndrome, heart disease, and deafness.

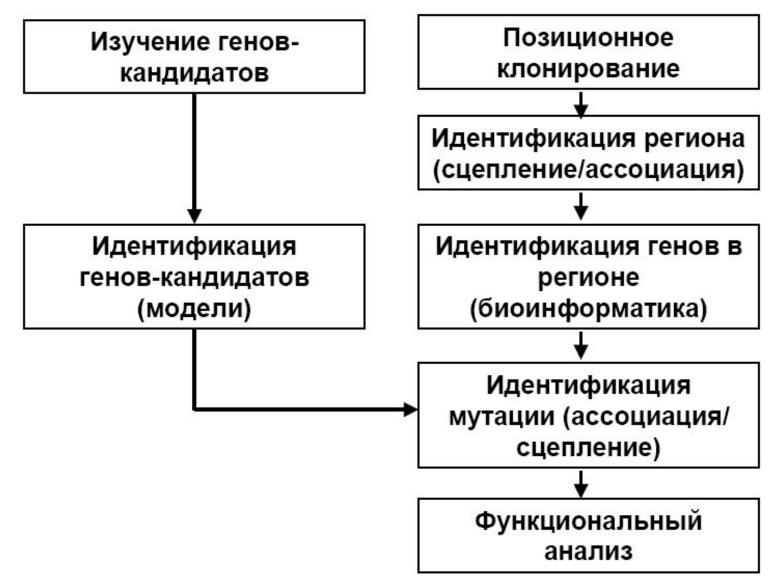
3931 "disease genes" are currently present in the GeneCards database

The genes listed here cause, predispose or protect from diseases (according to OMIM, SWISS-PROT, Genatlas, GeneTests, GAD, GDPInfo, bioalma, Leiden, Atlas, BCGD, TGDB and/or HGMD).

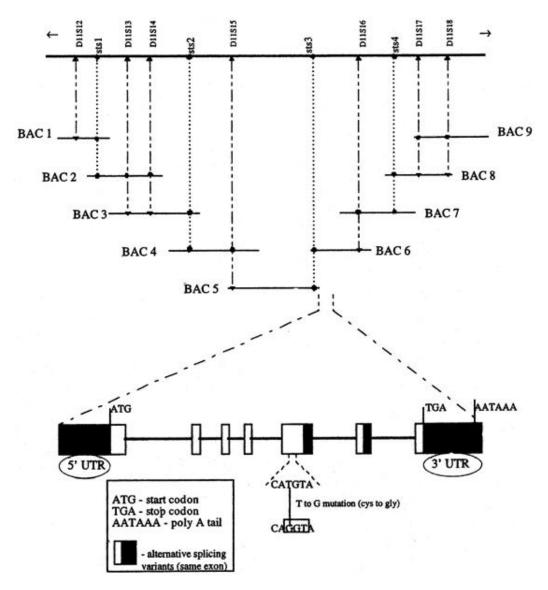
### **БАЗА ДАННЫХ NCBI**



http://www.ncbi.nlm.nih.gov/projects/mapview/maps.cgi?taxid=9606



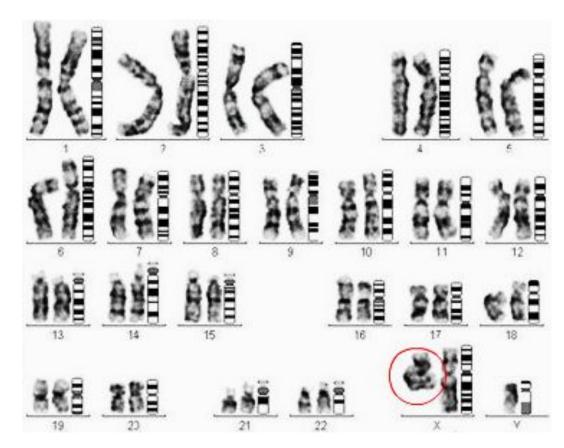
Основные стратегии генетического картирования



Основные этапы позиционного клонирования

# ЦИТОГЕНЕТИЧЕСКИЙ АНАЛИЗ





Внешний вид пациента и кариотип при синдроме Клайнфельтера

### FISH (ФЛУОРЕСЦЕНТНАЯ ГИБРИДИЗАЦИЯ IN SITU)

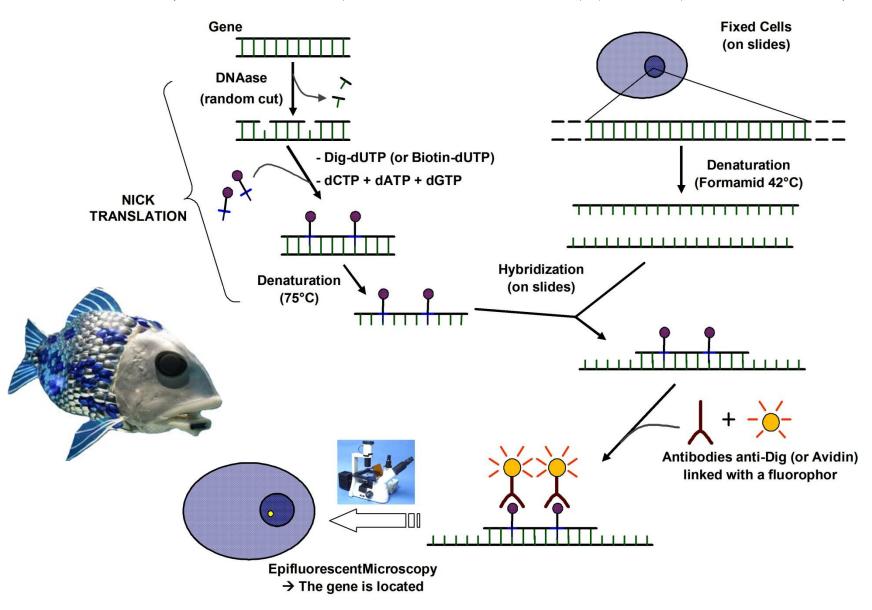
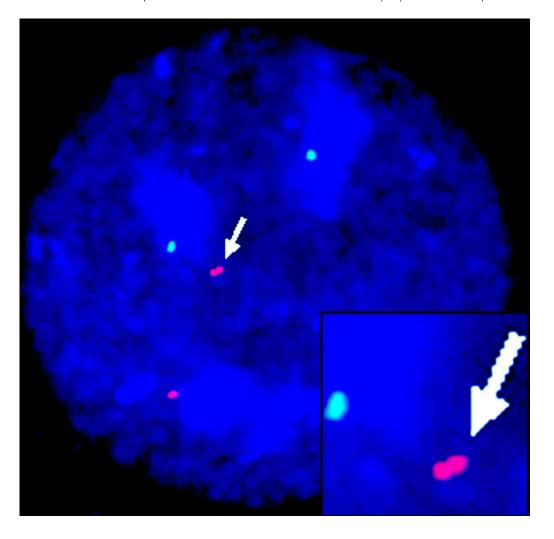


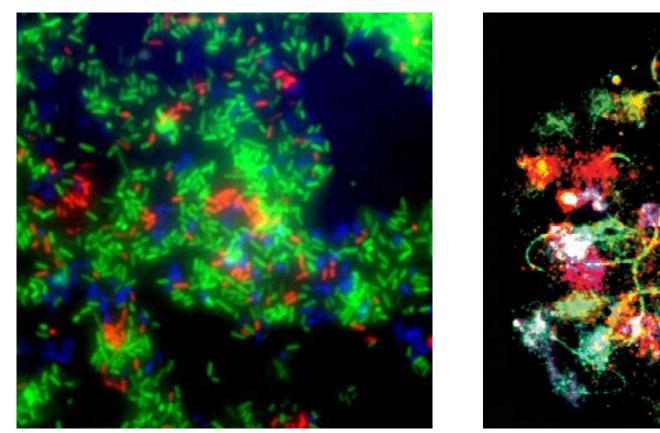
Схема постановки анализа FISH

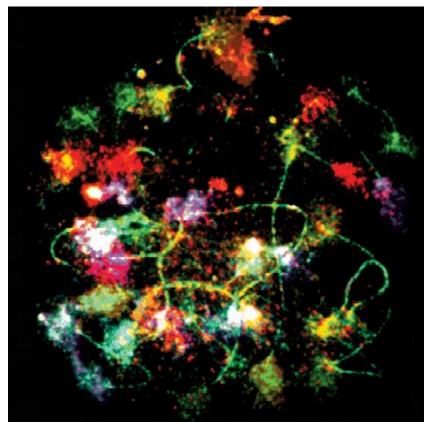
## FISH (ФЛУОРЕСЦЕНТНАЯ ГИБРИДИЗАЦИЯ IN SITU)



Детекция амплификации гена с помощью двухцветного FISH

# FISH (ФЛУОРЕСЦЕНТНАЯ ГИБРИДИЗАЦИЯ IN SITU)





Трехцветный и многоцветный FISH



### **International HapMap Project**

Home | About the Project | Data | Publications | Tutorial

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

#### Project Data

HapMap Genome Browser release #28 ( Phases 1, 2 & 3 - merged genotypes & frequencies )

HapMap3 Genome Browser release #3 ( Phase 3 - genotypes & frequencies )

HapMap Genome Browser release #27 ( Phase 1, 2 & 3 - merged genotypes & frequencies )

HapMap3 Genome Browser release #2 ( Phase 3 - genotypes, frequencies & LD )

HapMap Genome Browser release#24 ( Phase 1 & 2 - full dataset )

**GWAs Karyogram** 

HapMart

HapMap FTP

Bulk Data Download

Data Freezes for Publication

**ENCODE Project** 

Guidelines For Data Use

#### Useful Links

TSC SNP Downloads

HapMap Samples at Coriell Institute

HapMap Project Press Release

#### News

· 2011-06-13: HapMap help desk announcement

There was a problem with the HapMap help desk system. In the past several weeks, emails sent to hapmap-help@ncbi.nlm.nih.gov did not reach the help desk, and thus user requests were not addressed. Please resend your email request if you sent emails to the HapMap help desk in the past several weeks. Sorry for the inconvenience.

· 2011-04-20: Hapmap help desk service interruption notice

There will be no help desk support from 05/03/2011 to 05/23/2011. Sorry for the inconvenience.

· 2011-02-02: Haploview issues with rel 28 data

Recently, there are several questions about Haploview data format errors when users tried to analyze HapMap release 28 data. The current Haploview version (4.2) does not recognize the new individuals in release 28 and the software will generate an error similar to "Hapmap data format error: NA18876" when trying to open the data.

Haploview is developed and maintained by an organization different from HapMap. Please contact Haploview help desk (haploview@broadinstitute.org) for questions specific to this software.

2011-01-19: HapMap phase II recombination rate on GRCh37

The liftover of the HapMap II genetic map from human genome build b35 to GRCh37 is available. Data is available for bulk download.

2010-08-18: HapMap Public Release #28

Genotypes and frequency data in hapmap format are now available for data in merged HapMap phases I+II+III release #28 (NCBI build 36, dbSNP b126). Data is available for bulk download and also available for browsing. Click here to read the latest release notes.

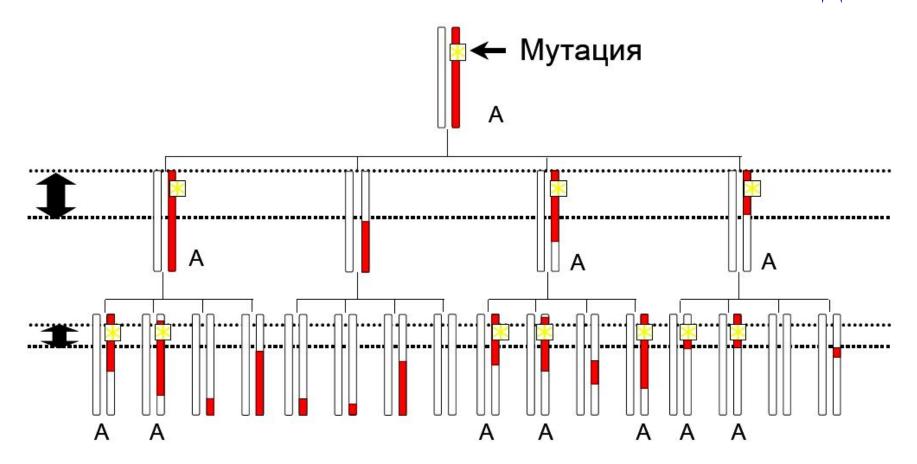
2010-05-28: HapMap3 Public Release #3

Genotypes and frequency data in hapmap format are now available for data in HapMap phase 3 release #3 (NCBI build 36, dbSNP b126). Data is available for bulk download and also available for browsing. Click here to read the latest release notes.

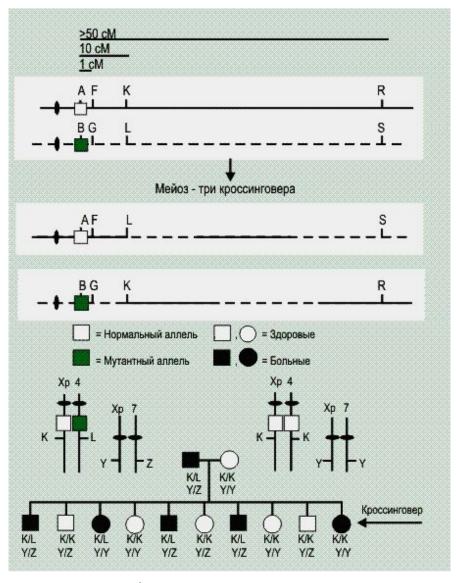
2010-05-28: HapMap3 CNV Genotypes

Copy Number Variation genotypes for HapMap phase samples are available for bulk download.

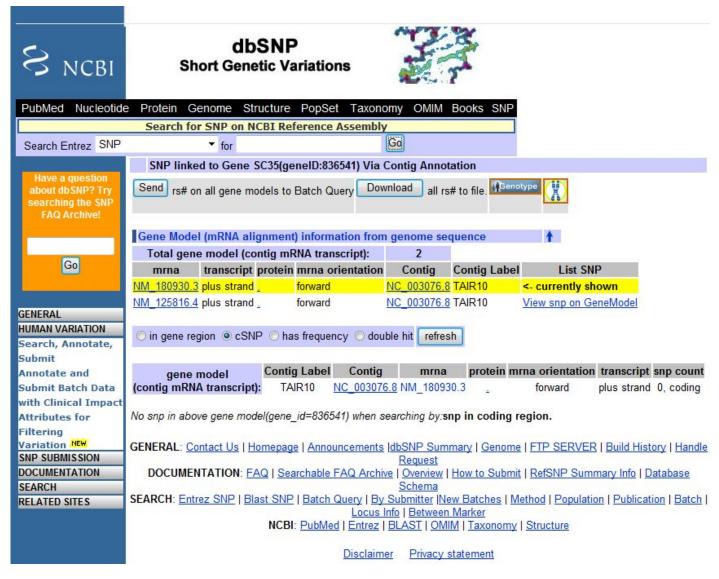
2009-12-10: Corrected HapMap3 phased haplotypes available for chromosome X



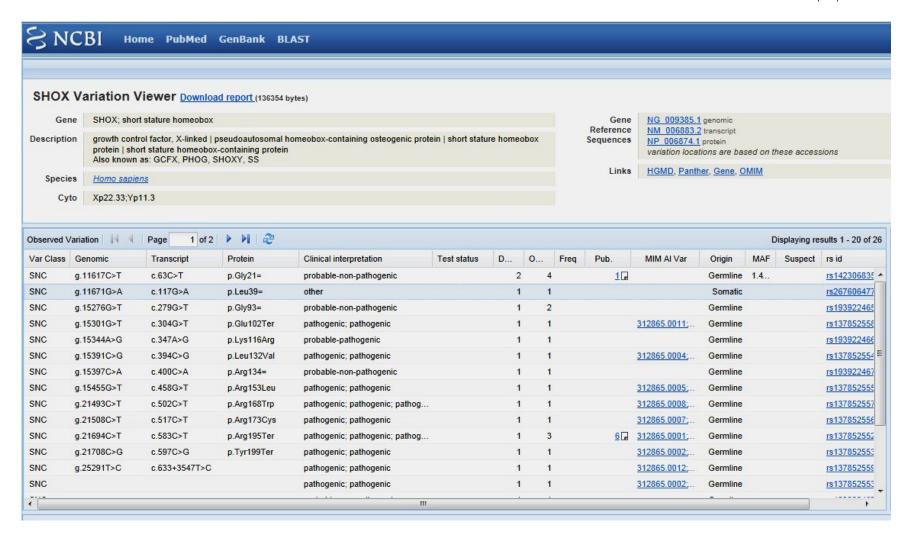
Совместная сегрегация мутации (отмечено квадратом) и болезни (А) в ряду поколений



Анализ сцепления

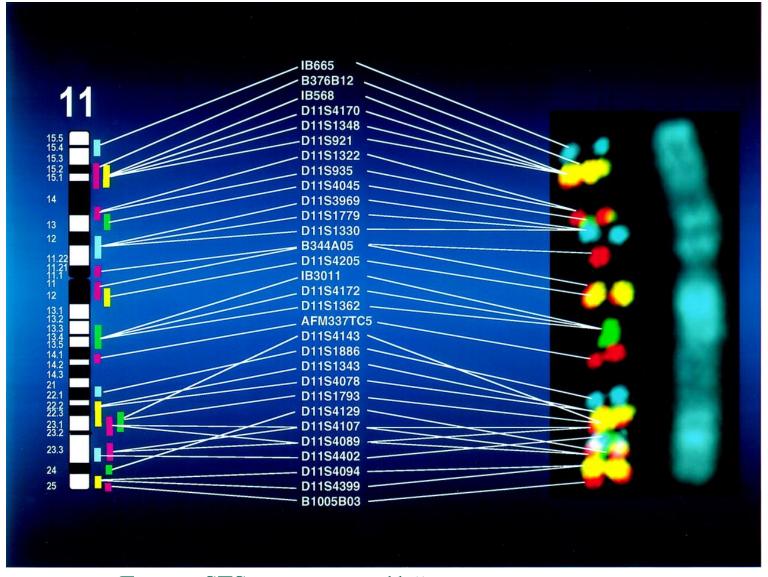


http://www.ncbi.nlm.nih.gov/snp

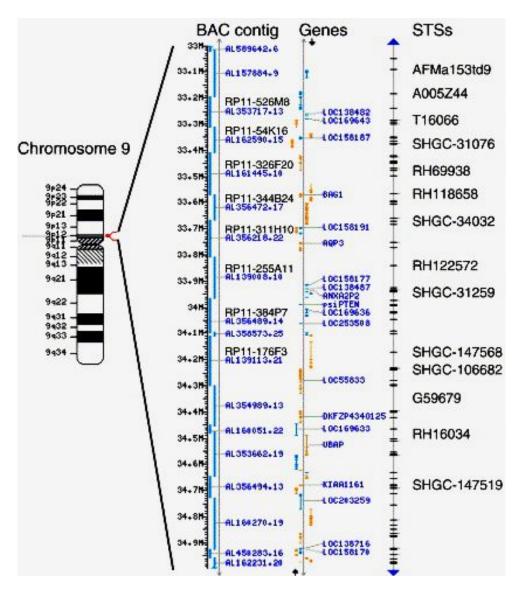


Платформа	Количество полиморфных маркеров	Охват, %
Illumina HumanHap300	317511	75
Affymetrix SNP Array 5.0	500568	65
Illumina HumanHap550	555352	87
Illumina Human610	620901	89
Illumina HumanHap650Y	660917	87
Affymetrix SNP Array 6.0	более 800000	83
Illumina Human1M	1199187	93

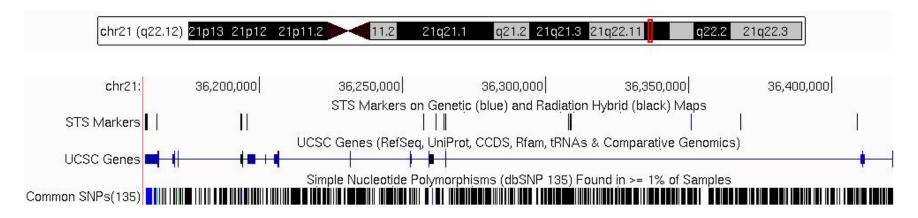
Коммерчески доступные платформы генотипирования на микрочипах высокой плотности



Группа STS-маркеров в 11-й хромосоме человека



Группа STS-маркеров в 9-й хромосоме человека



STS-маркеры гена RUNX1 человека

A	Geno	mes	Genome E	Browser 1	Tools	Mirrors	Downloads	My Data	About Us	Help
STS Mar	rker A	FMB2	80XD9							
Chromo	some	: chr21								
Start:		36350985								
End:		36351354								
Band:		21q22.12								
Other na	ames:	D21S1	895, SHG	C-21164, RH	131492,	B280XD9,	W6371, RH49	565, RH7256	6, HSB280XD	9, STSG17506
UCSC S	TS id:	4957								
UniSTS	id:	16185								
Genban	k:	Z53307	7							
GDB:		GDB:6	10035							
Organis	m:	Homo	sapiens							
Left Prin	ner:	AGTCC	TACTGA	TAAACTGT	GGGC					
Right Pr	imer:	CTGTC	TCATAAC	BAACCTAC	CTGG					
Distance	e:	224-282	2 bps							
Genetic	: Мар	Posit	ions							
	Na	ame		Chromos	ome	Positio	on			
Genethon: AFMB280XD9		XD9	chr21		37.20					
Marshfield: AFMB280XD9 chr21					33.84					
RH Map	Pos	itions								
		Name		Chron	nosome	Pos	ition (LOD)			
<b>GM99 G</b>	3:	RH314	192	chr21		984	.00 (3.50)			
			-21164	chr21		10000	18.00			

### STS-маркер AFMB280XD9