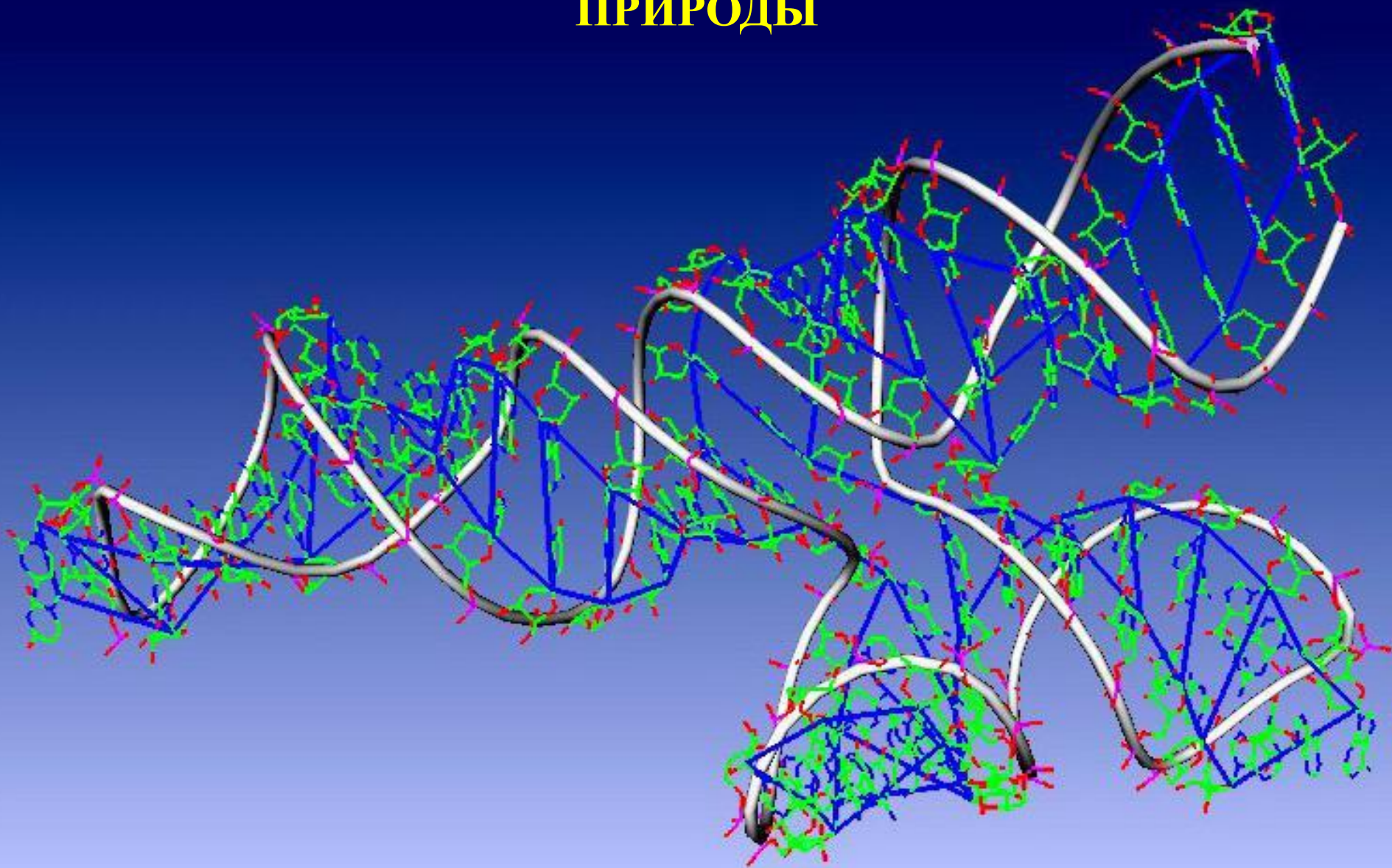
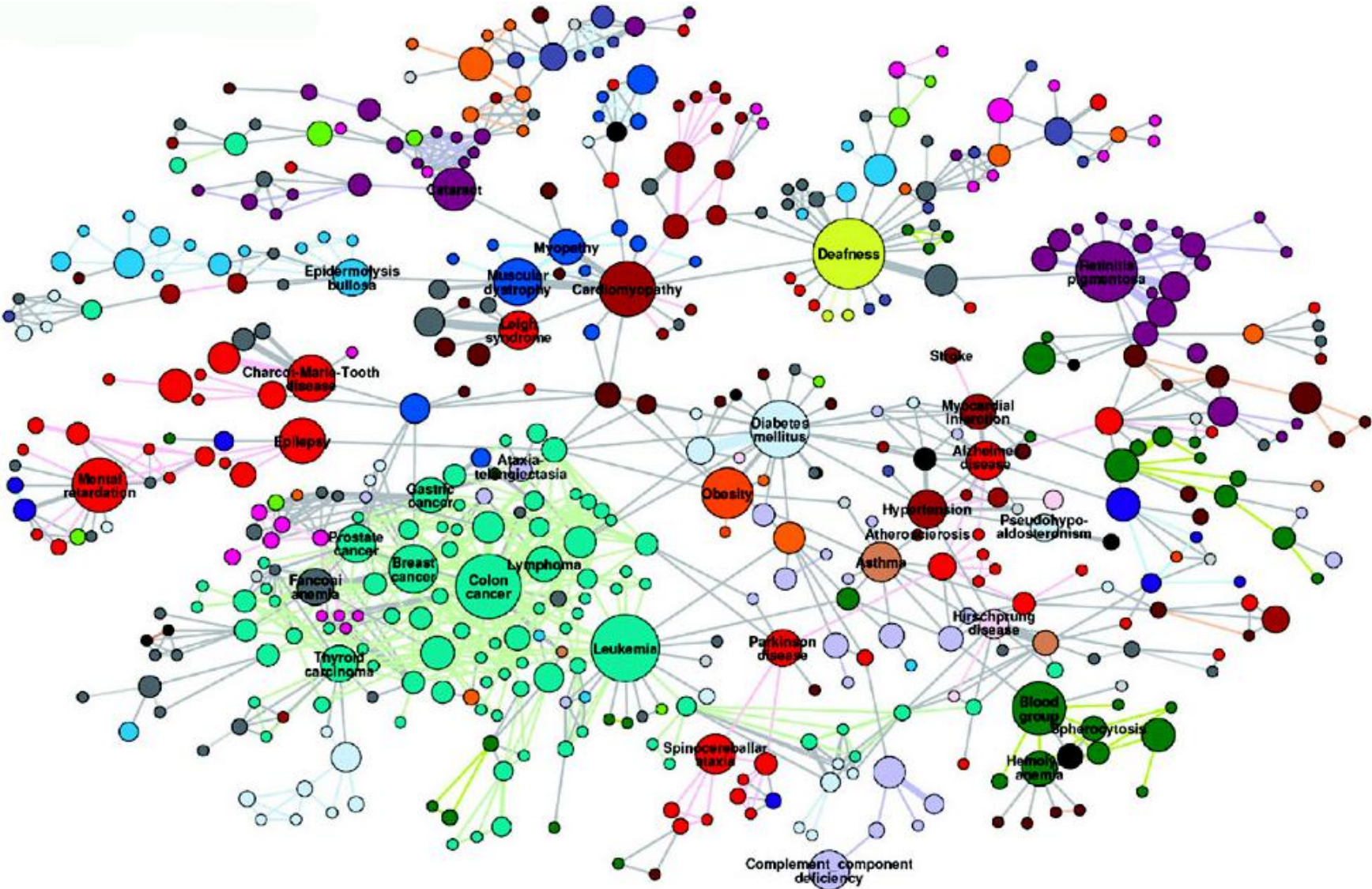


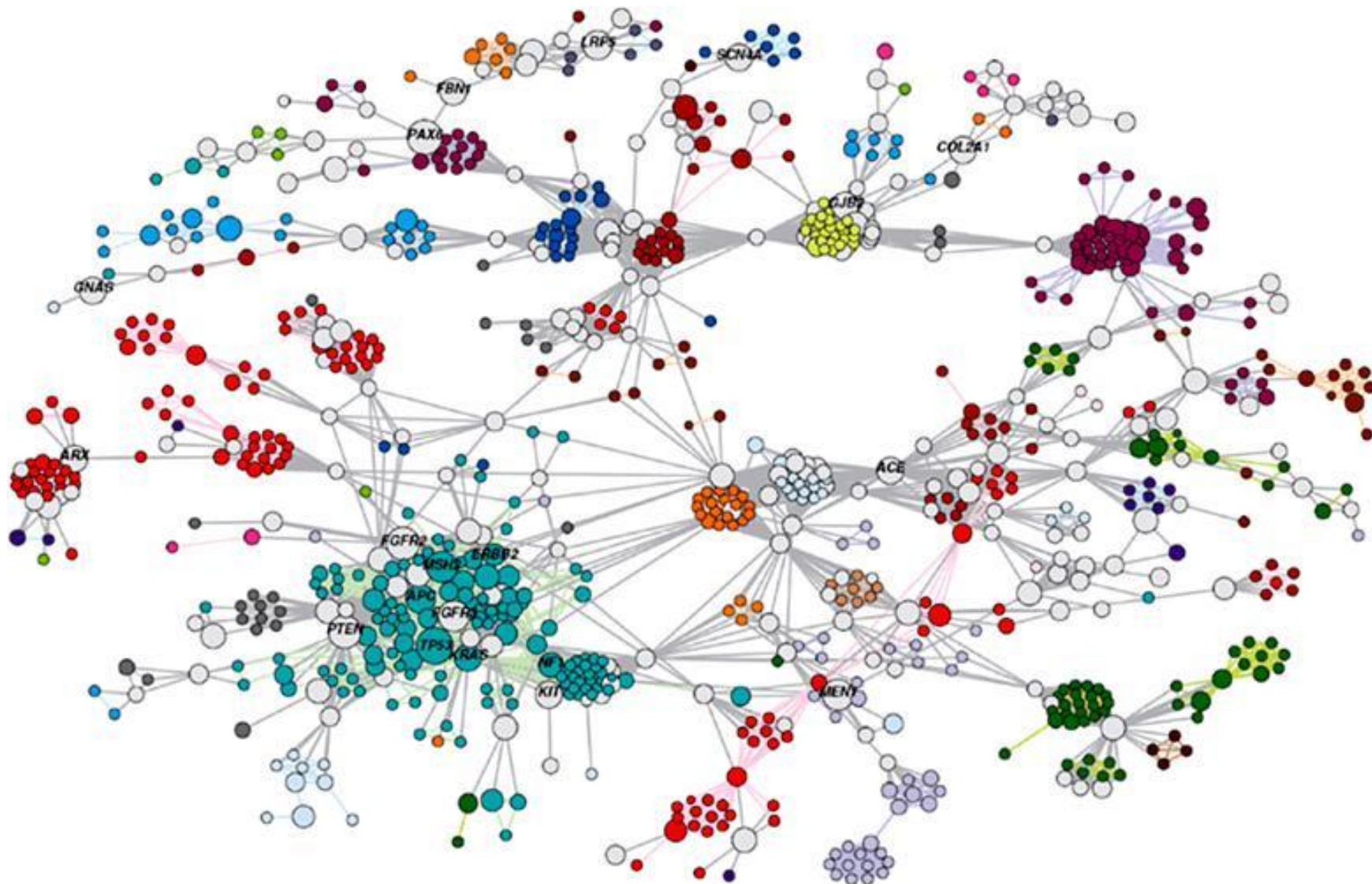
# ИДЕНТИФИКАЦИЯ ГЕНОВ, ОТВЕТСТВЕННЫХ ЗА ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



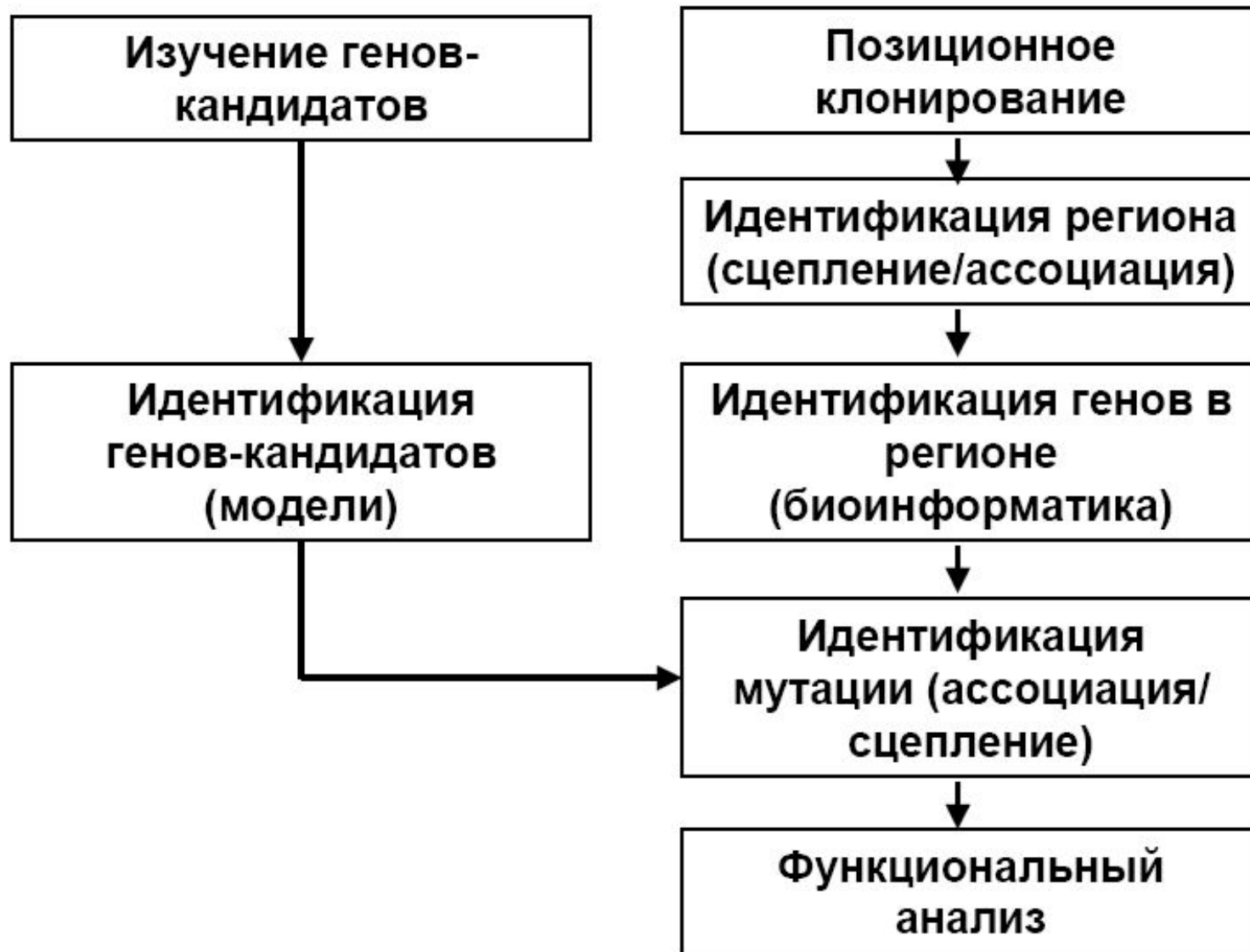
# СЕТЬ ЗАБОЛЕВАНИЙ ЧЕЛОВЕКА



# СЕТЬ ГЕНОВ, ОТВЕТСТВЕННЫХ ЗА ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ

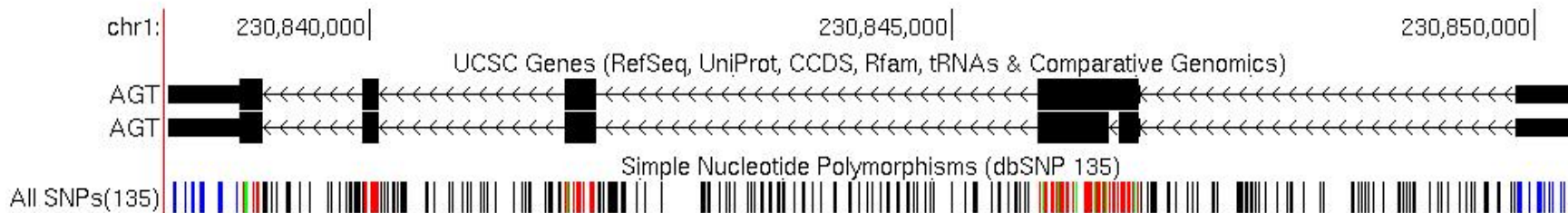


# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



Основные стратегии генетического картирования

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



**Ген ангиотензиногена**

# БАЗА ДАННЫХ OMIM

OMIM - HEMOCHROMATOSIS; HFE - Microsoft Internet Explorer

File Edit View Favorites Tools Help

Address <http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=235200> Go Links

NCBI

OMIM  
Online Mendelian Inheritance in Man  
Johns Hopkins University

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM

Search OMIM for Go Clear

Limits Preview/Index History Clipboard Details

Display Detailed Show: 20 Send to File

**\*235200** [Links](#)

**HEMOCHROMATOSIS; HFE**

*Alternative titles; symbols*

**HLAH**  
**HEMOCHROMATOSIS, HEREDITARY; HH**  
**HEMOCHROMATOSIS GENE, INCLUDED; HFE GENE, INCLUDED**

Gene map locus [6p21.3](#)

**TEXT**

**DESCRIPTION**

The features of hemochromatosis include cirrhosis of the liver, diabetes, hypermelanotic pigmentation of the skin, and heart failure. Primary hepatocellular carcinoma (HCC; [114550](#)), complicating cirrhosis, is responsible for about one-third of deaths in affected homozygotes. Since hemochromatosis is a relatively easily treated disorder if diagnosed, this is a form of preventable cancer. 🧠

Internet

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

# БАЗА ДАННЫХ ОМІМ

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

**Статистика по базе данных ОМІМ  
на 17 октября 2014 года**

# БАЗА ДАННЫХ GeneCards

The screenshot shows the top section of the GeneCards website. On the left is the GeneCards logo with the tagline 'The Human Gene Compendium'. In the center is the Weizmann Institute of Science logo with Hebrew text 'מכון ויצמן למדע' and the English name 'WEIZMANN INSTITUTE OF SCIENCE'. On the right is the XENVEIX logo, a division of LifeMap Sciences. Below the logos is a navigation bar with links: Home, GeneCards Guide, Suite, Terms and Conditions, About Us, User Feedback, and Mirror sites. Below the navigation bar are links for 'Set Analyses: Export List', 'GeneALaCart', and 'GeneDecks'. A search bar contains the text 'keyword(s)' and a 'Search' button. To the right of the search bar is a link for 'Advanced Search'.

## 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

 **NCBI**

Human genome overview page (Build 37.3)  
Human genome overview page (Build 36.3)  
[Map Viewer Home](#)

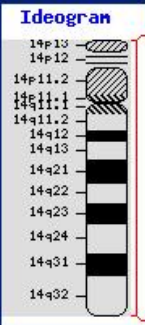
Map Viewer Help  
Human Maps Help  
FTP  
Data As Table View  
**Maps & Options**

Region Shown:



You are here:

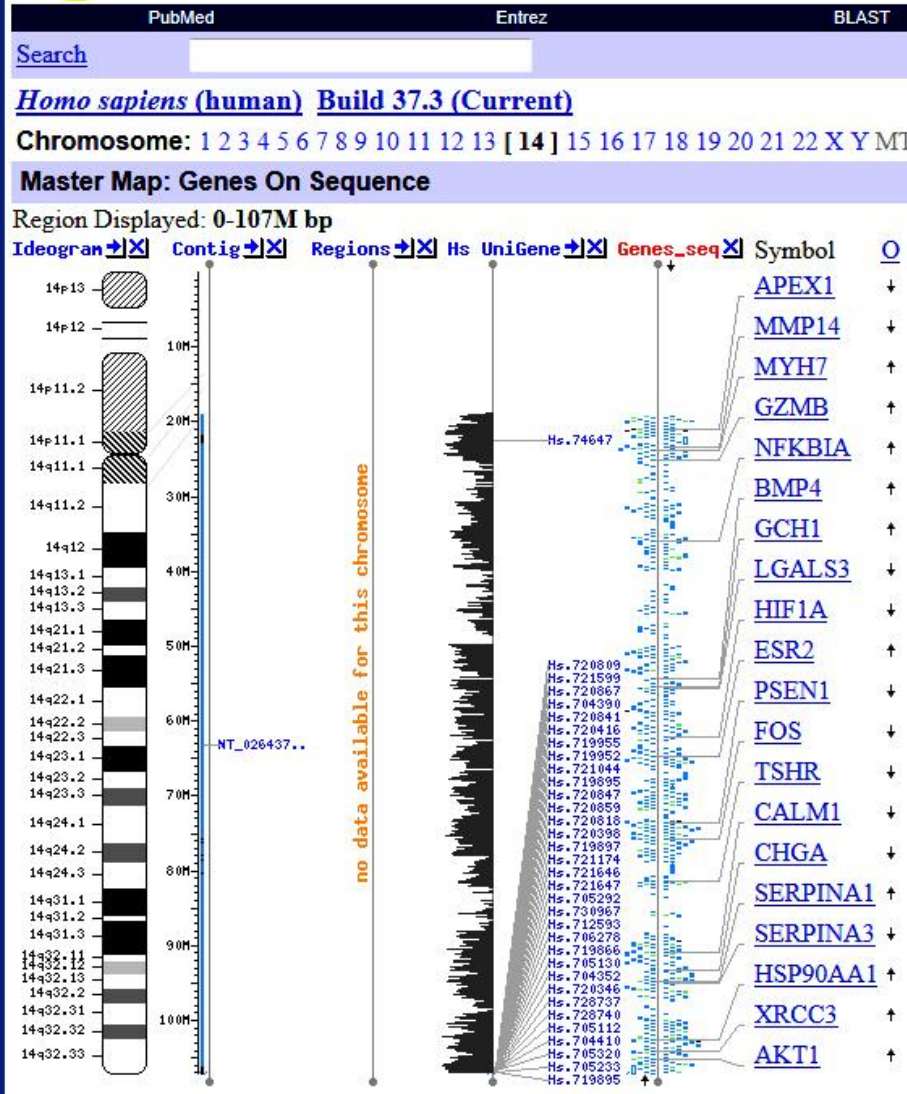
**Ideogram**



default  
 master

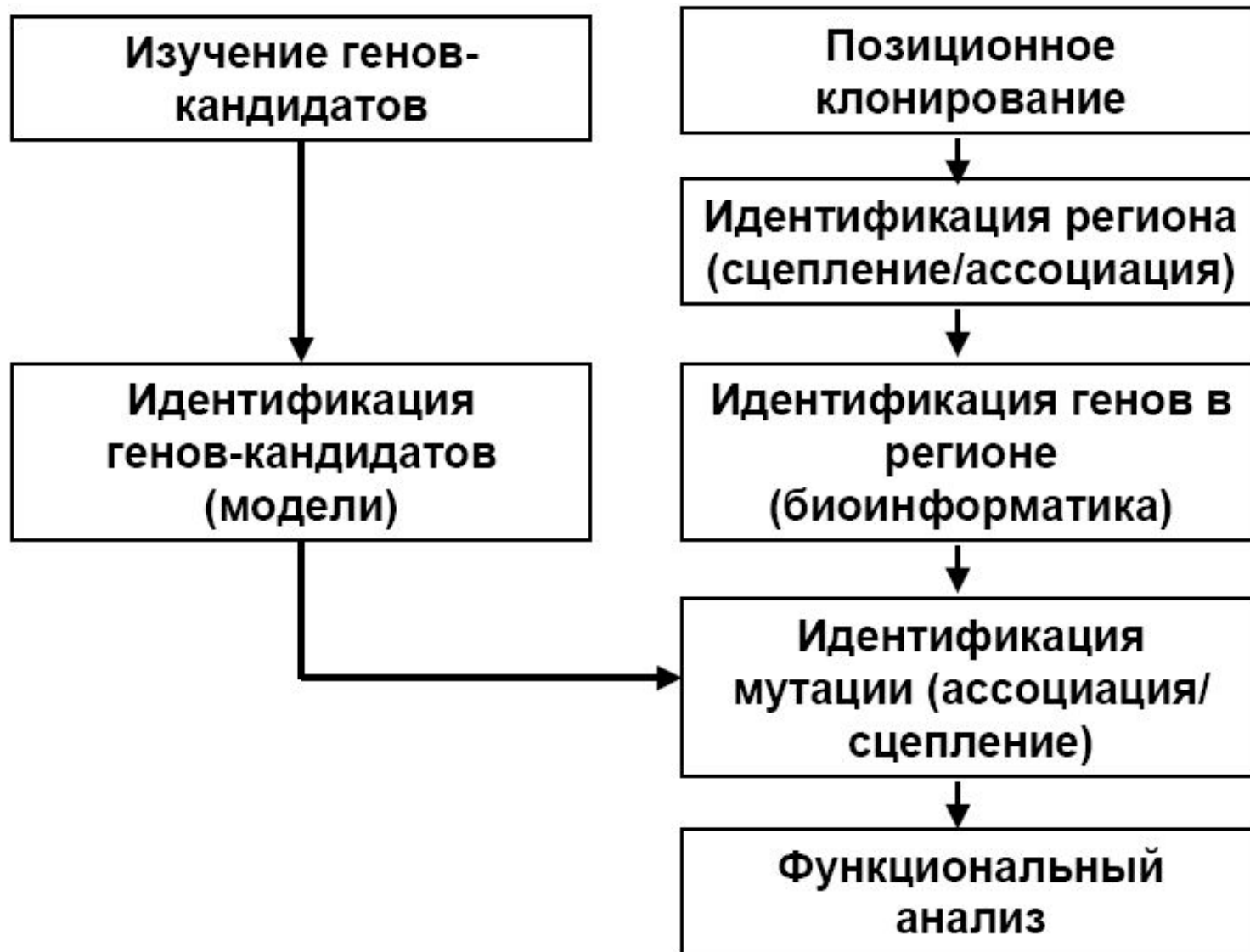


NCBI Map Viewer



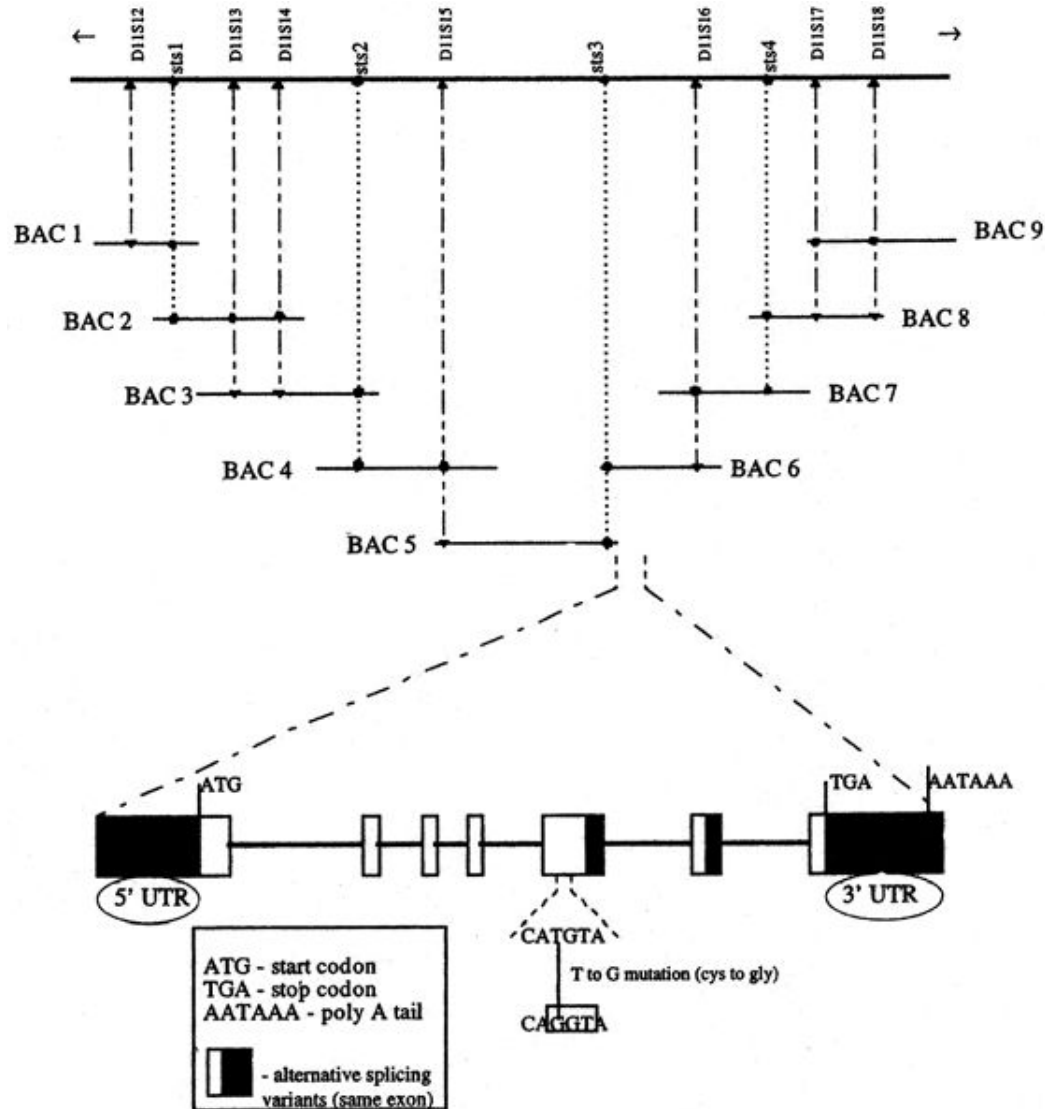
<http://www.ncbi.nlm.nih.gov/projects/mapview/maps.cgi?taxid=9606>

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



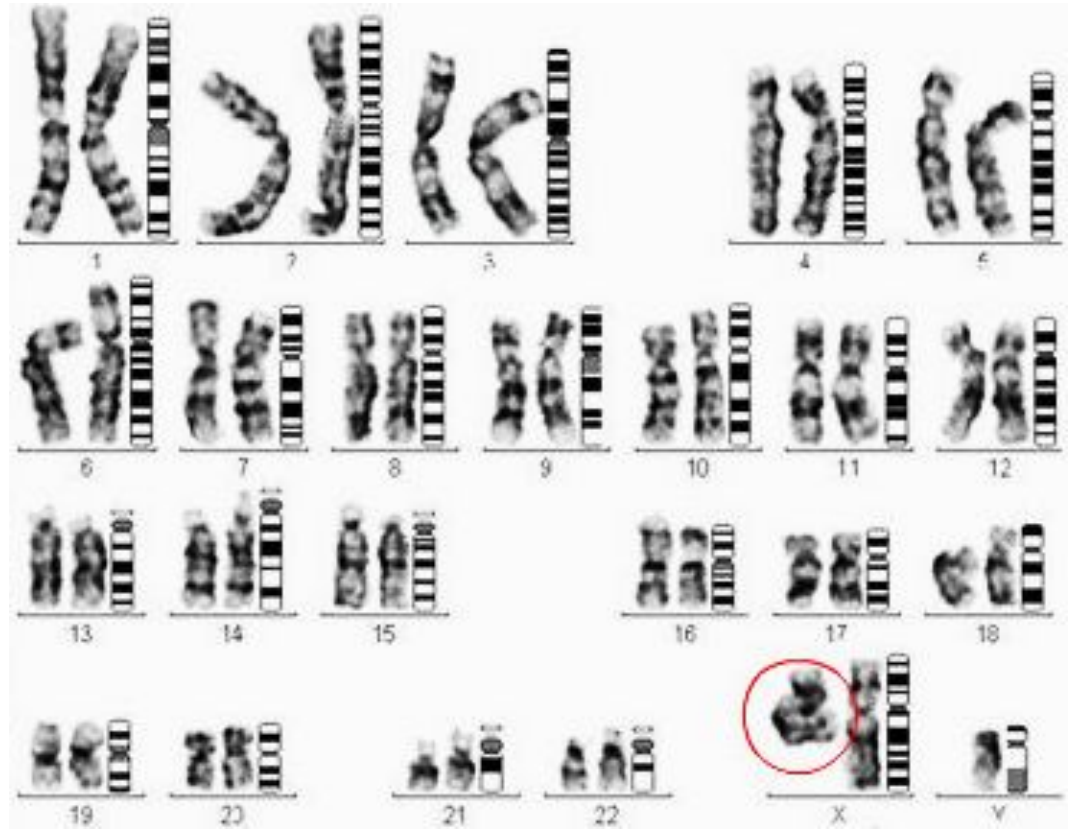
Основные стратегии генетического картирования

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



Основные этапы позиционного клонирования

# ЦИТОГЕНЕТИЧЕСКИЙ АНАЛИЗ



**Внешний вид пациента и кариотип при синдроме Клайнфельтера**

# FISH (ФЛУОРЕСЦЕНТНАЯ ГИБРИДИЗАЦИЯ IN SITU)

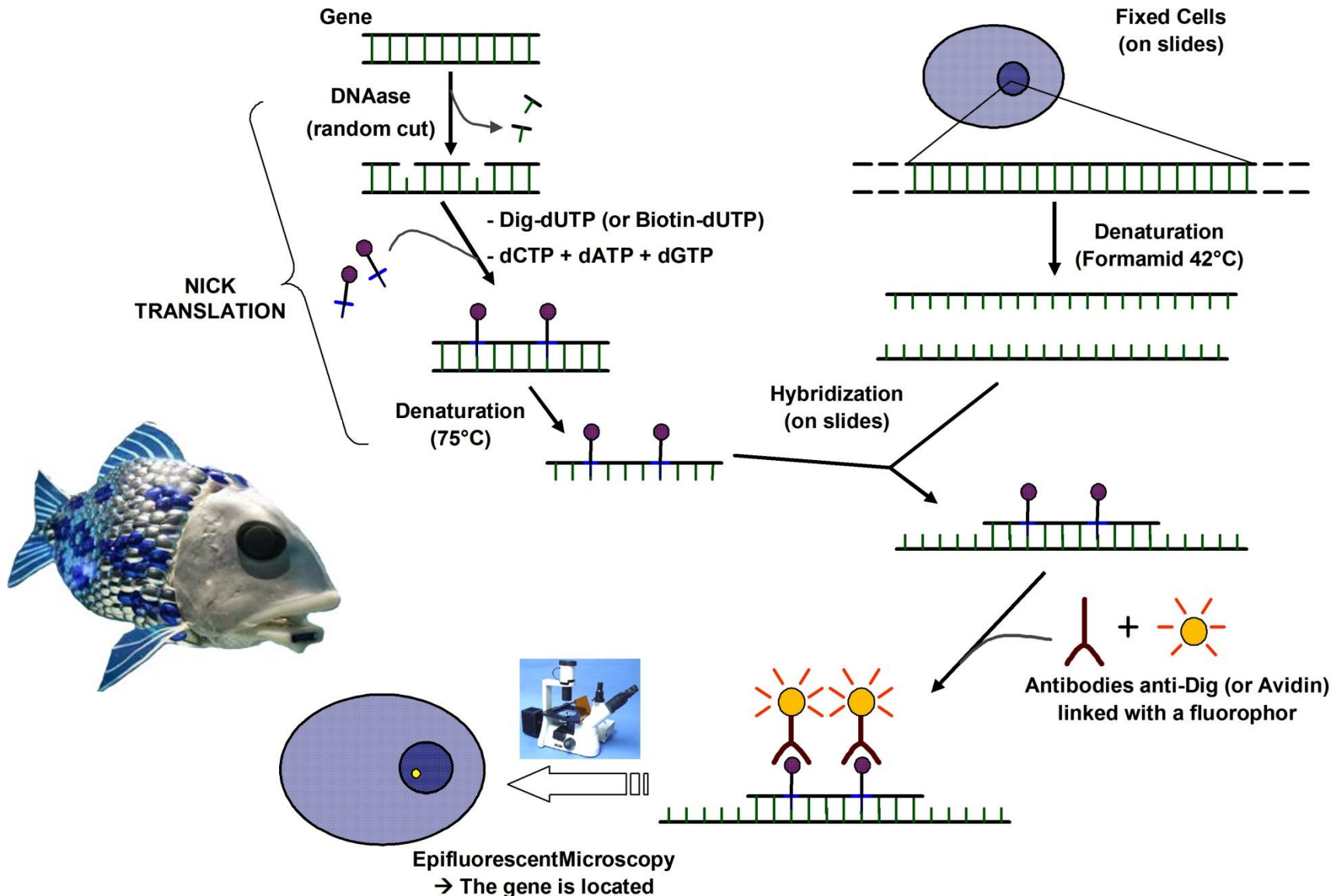
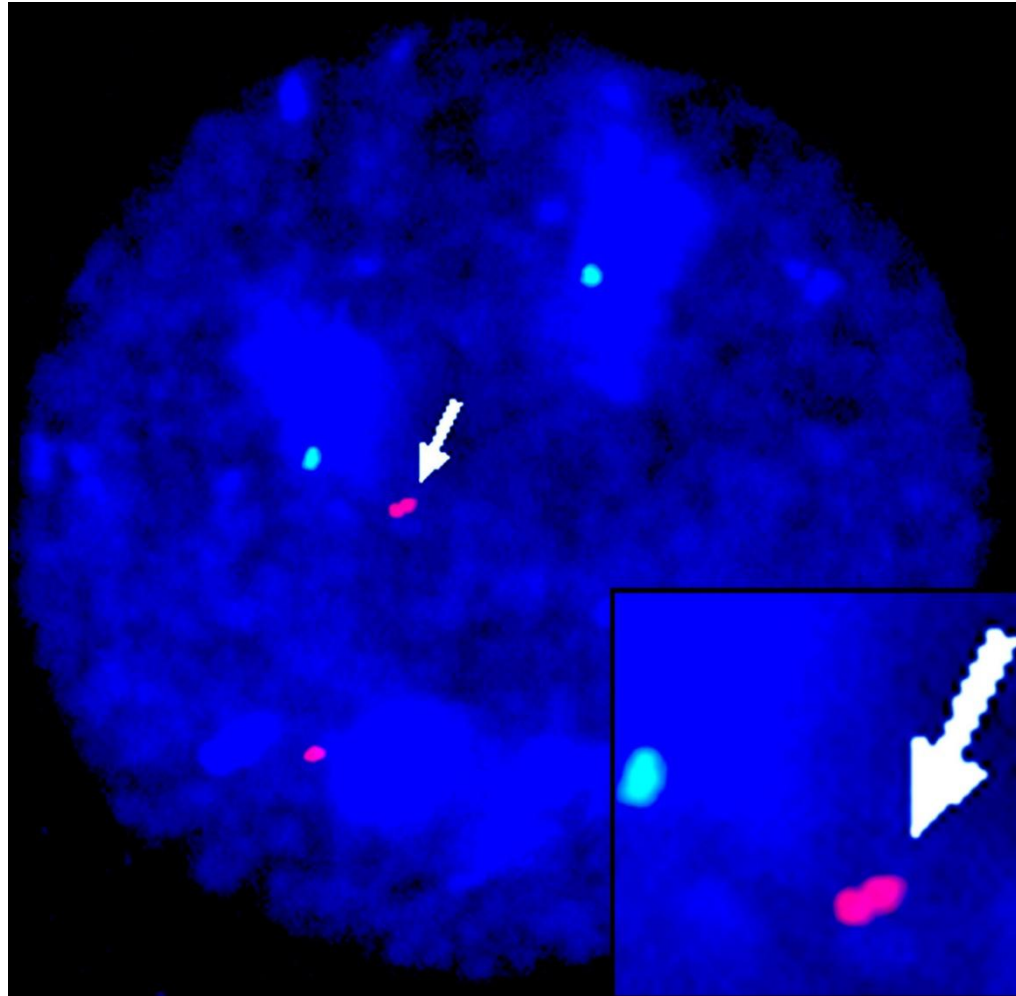


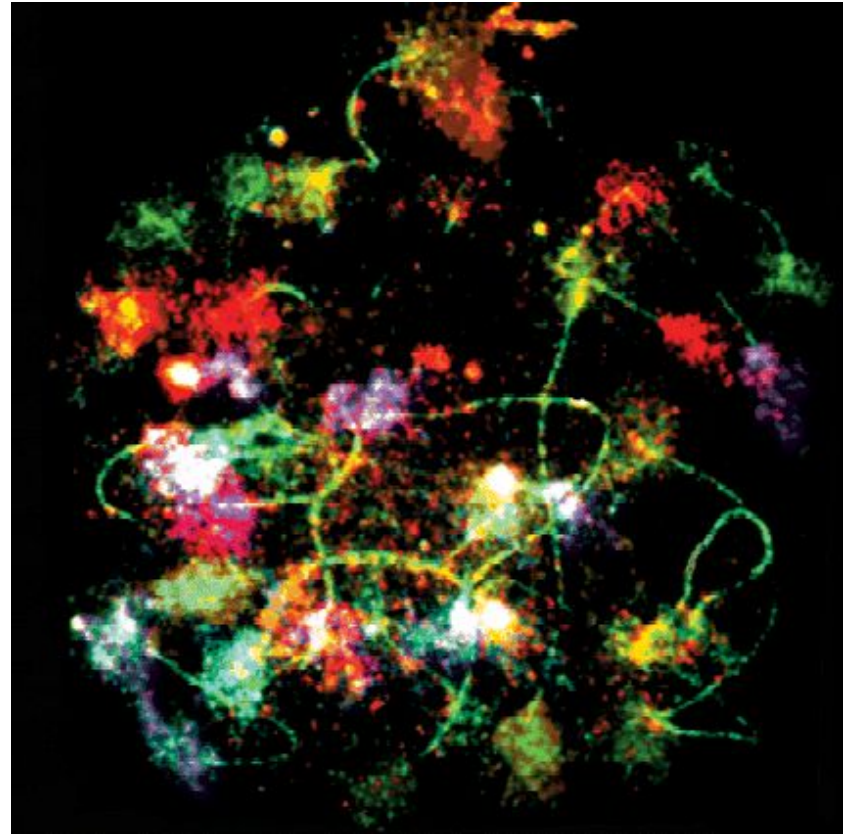
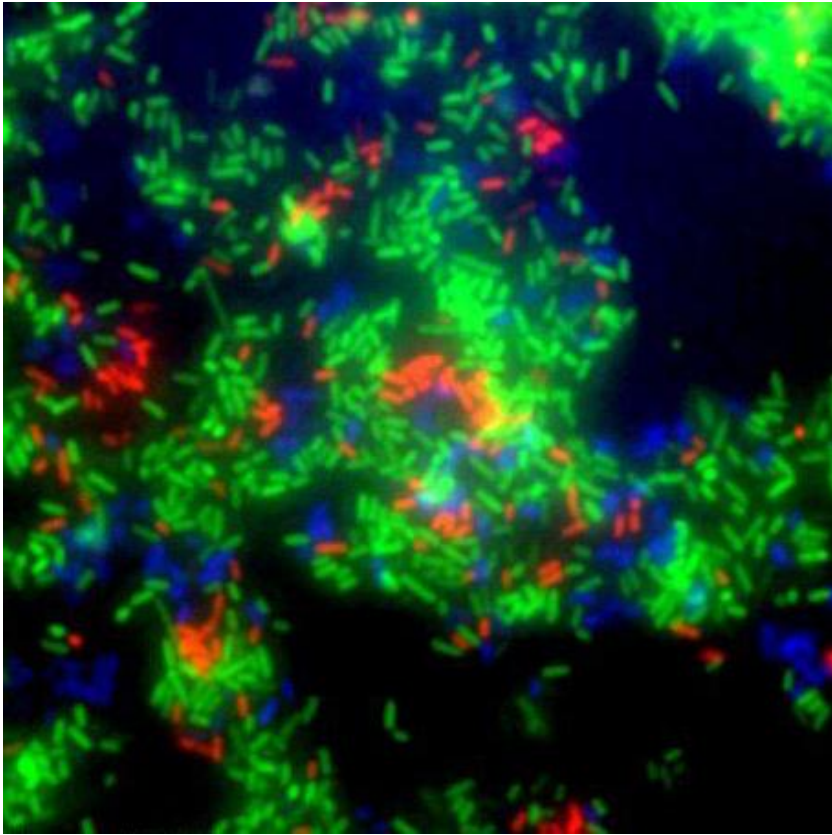
Схема постановки анализа FISH

# FISH (ФЛУОРЕСЦЕНТНАЯ ГИБРИДИЗАЦИЯ IN SITU)



Детекция амплификации гена  
с помощью двухцветного FISH

# FISH (ФЛУОРЕСЦЕНТНАЯ ГИБРИДИЗАЦИЯ IN SITU)



Трехцветный и многоцветный FISH

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



## International HapMap Project

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

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

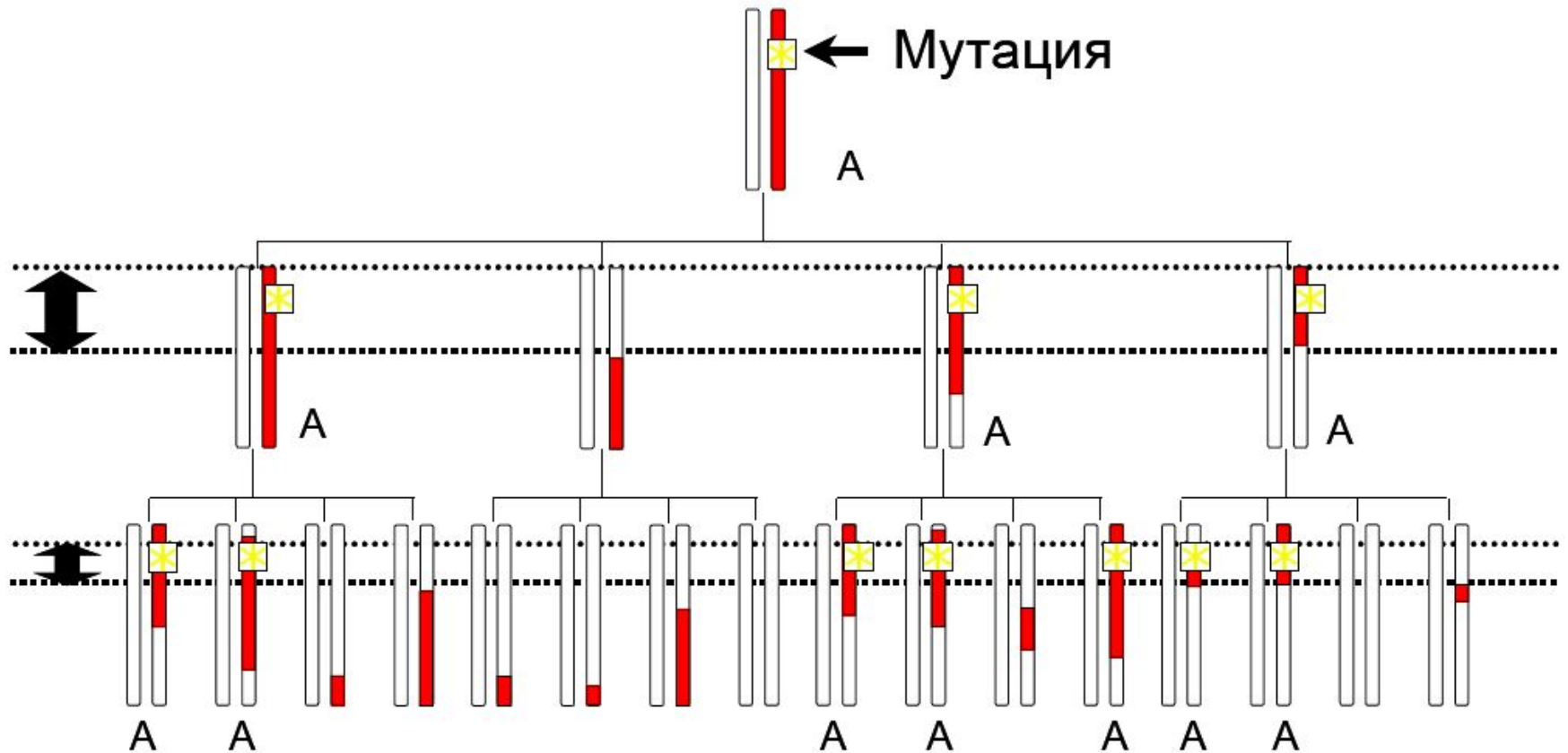
The International HapMap Project is a partnership of scientists and funding agencies from Canada, China, Japan, Nigeria, the United Kingdom and the United States to develop a public resource that will help researchers find genes associated with human disease and response to pharmaceuticals. See "[About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

Project Information	News
<p><b>About the Project</b></p> <ul style="list-style-type: none"><li><a href="#">HapMap Publications</a></li><li><a href="#">HapMap Tutorial</a></li><li><a href="#">HapMap Mailing List</a></li><li><a href="#">HapMap Project Participants</a></li></ul>	<ul style="list-style-type: none"><li>• 2011-06-13: <a href="#">HapMap help desk announcement</a> 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.</li><li>• 2011-04-20: <a href="#">Hapmap help desk service interruption notice</a> There will be no help desk support from 05/03/2011 to 05/23/2011. Sorry for the inconvenience.</li><li>• 2011-02-02: <a href="#">Haploview issues with rel 28 data</a> 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.</li><li>• 2011-01-19: <a href="#">HapMap phase II recombination rate on GRCh37</a> The liftover of the HapMap II genetic map from human genome build b35 to GRCh37 is available. Data is <a href="#">available for bulk download</a>.</li><li>• 2010-08-18: <a href="#">HapMap Public Release #28</a> 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 <a href="#">available for bulk download</a> and also <a href="#">available for browsing</a>. Click here to read the latest <a href="#">release notes</a>.</li><li>• 2010-05-28: <a href="#">HapMap3 Public Release #3</a> 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 <a href="#">available for bulk download</a> and also <a href="#">available for browsing</a>. Click here to read the latest <a href="#">release notes</a>.</li><li>• 2010-05-28: <a href="#">HapMap3 CNV Genotypes</a> Copy Number Variation genotypes for HapMap phase samples are <a href="#">available for bulk download</a>.</li><li>• 2009-12-10: <a href="#">Corrected HapMap3 phased haplotypes available for chromosome X</a></li></ul>
<p><b>Project Data</b></p> <ul style="list-style-type: none"><li><a href="#">HapMap Genome Browser release #28 ( Phases 1, 2 &amp; 3 - merged genotypes &amp; frequencies )</a></li><li><a href="#">HapMap3 Genome Browser release #3 ( Phase 3 - genotypes &amp; frequencies )</a></li><li><a href="#">HapMap Genome Browser release #27 ( Phase 1, 2 &amp; 3 - merged genotypes &amp; frequencies )</a></li><li><a href="#">HapMap3 Genome Browser release #2 ( Phase 3 - genotypes, frequencies &amp; LD )</a></li><li><a href="#">HapMap Genome Browser release#24 ( Phase 1 &amp; 2 - full dataset )</a></li><li><a href="#">GWAs Karyogram</a></li><li><a href="#">HapMart</a></li><li><a href="#">HapMap FTP</a></li><li><a href="#">Bulk Data Download</a></li><li><a href="#">Data Freezes for Publication</a></li><li><a href="#">ENCODE Project</a></li><li><a href="#">Guidelines For Data Use</a></li></ul>	
<p><b>Useful Links</b></p> <ul style="list-style-type: none"><li><a href="#">TSC SNP Downloads</a></li><li><a href="#">HapMap Samples at Coriell Institute</a></li><li><a href="#">HapMap Project Press Release</a></li></ul>	

<http://hapmap.ncbi.nlm.nih.gov/index.html.en>

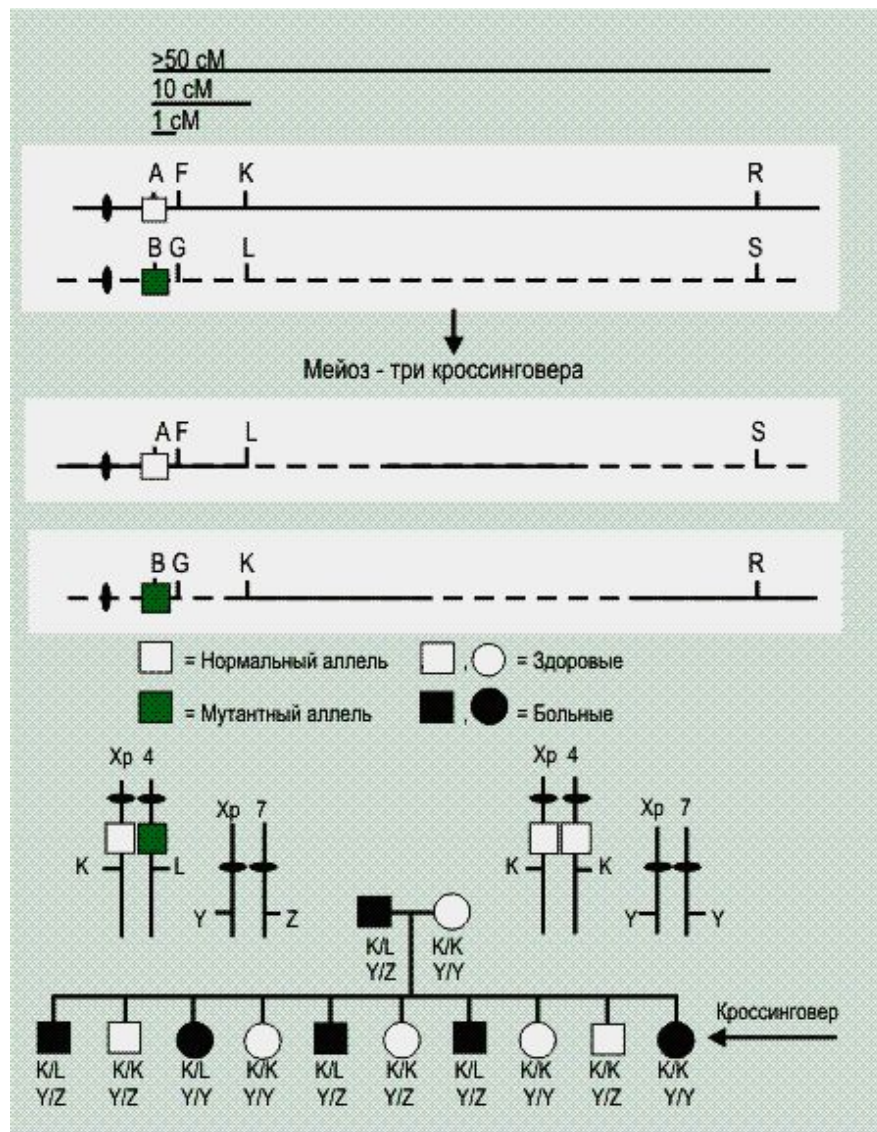


# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ




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

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



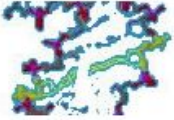
Анализ сцепления

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



## dbSNP

Short Genetic Variations



---


PubMed
Nucleotide
Protein
Genome
Structure
PopSet
Taxonomy
OMIM
Books
SNP

Search for SNP on NCBI Reference Assembly

Search Entrez  for

Have a question about dbSNP? Try searching the SNP FAQ Archive!

SNP linked to Gene SC35(geneID:836541) Via Contig Annotation

rs# on all gene models to Batch Query
  all rs# to file:  

Gene Model (mRNA alignment) information from genome sequence

Total gene model (contig mRNA transcript):		2				
mRNA	transcript	protein	mRNA orientation	Contig	Contig Label	List SNP
NM_180930.3	plus strand		forward	NC_003076.8	TAIR10	<- currently shown
NM_125816.4	plus strand		forward	NC_003076.8	TAIR10	<a href="#">View snp on GeneModel</a>

in gene region
  cSNP
  has frequency
  double hit

gene model	Contig Label	Contig	mRNA	protein	mRNA orientation	transcript	snp count
(contig mRNA transcript):	TAIR10	NC_003076.8	NM_180930.3		forward	plus strand	0, coding

No snp in above gene model(gene\_id=836541) when searching by:snp in coding region.

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# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ

## SHOX Variation Viewer [Download report](#) (136354 bytes)

Gene: SHOX; short stature homeobox

Description: growth control factor, X-linked | pseudoautosomal homeobox-containing osteogenic protein | short stature homeobox protein | short stature homeobox-containing protein  
Also known as: GCFX, PHOG, SHOXY, SS

Species: [Homo sapiens](#)

Cyto: Xp22.33;Yp11.3

Gene Reference Sequences: [NG\\_009385.1](#) genomic, [NM\\_006883.2](#) transcript, [NP\\_006874.1](#) protein  
*variation locations are based on these accessions*

Links: [HGMD](#), [Panther](#), [Gene](#), [OMIM](#)

Observed Variation | Page 1 of 2 | Displaying results 1 - 20 of 26

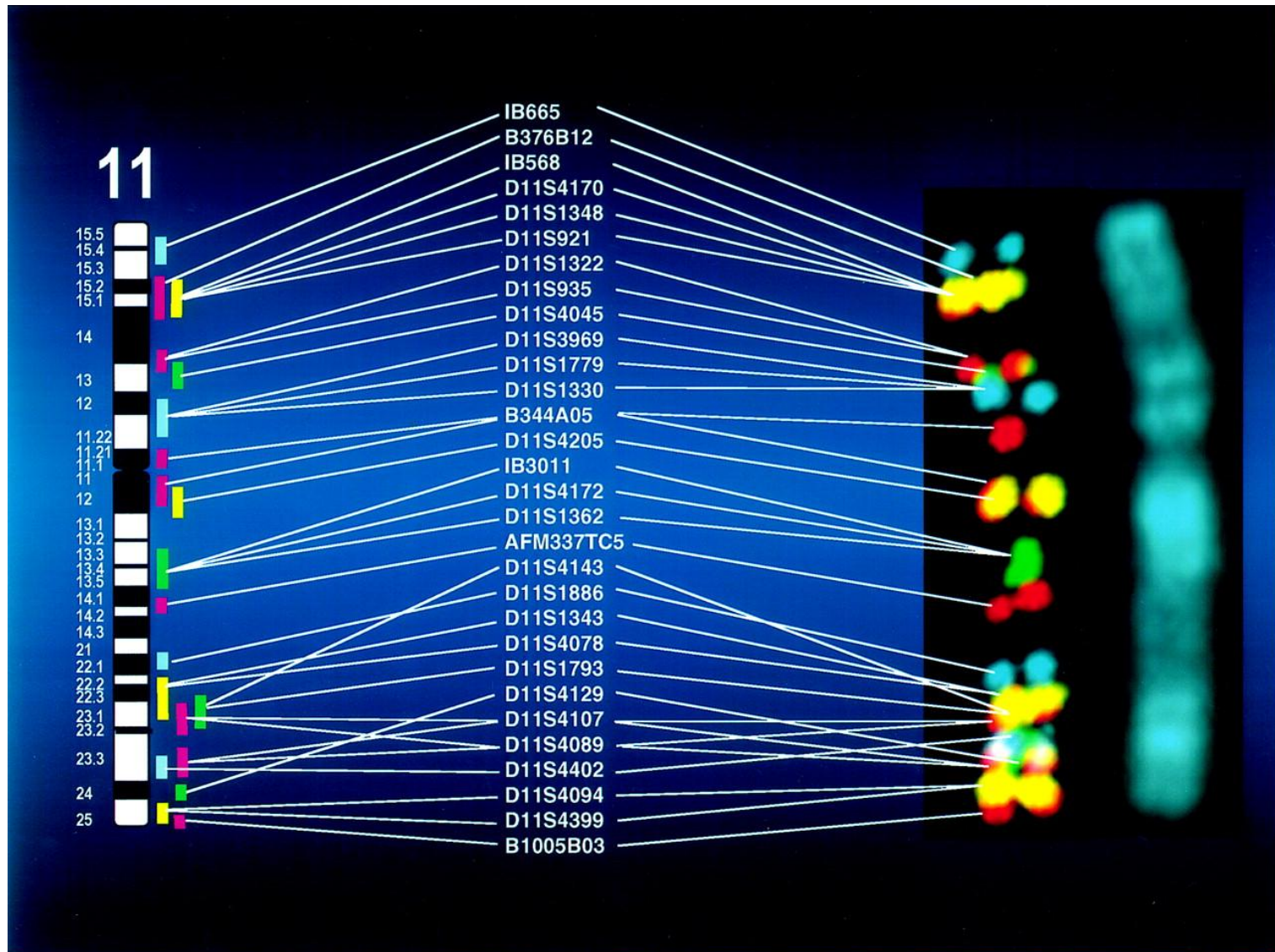
Var Class	Genomic	Transcript	Protein	Clinical interpretation	Test status	D...	O...	Freq	Pub.	MIM AI Var	Origin	MAF	Suspect	rs id
SNC	g.11617C>T	c.63C>T	p.Gly21=	probable-non-pathogenic		2	4		1		Germline	1.4..		<a href="#">rs142306835</a>
SNC	g.11671G>A	c.117G>A	p.Leu39=	other		1	1				Somatic			<a href="#">rs267606477</a>
SNC	g.15276G>T	c.279G>T	p.Gly93=	probable-non-pathogenic		1	2				Germline			<a href="#">rs193922465</a>
SNC	g.15301G>T	c.304G>T	p.Glu102Ter	pathogenic; pathogenic		1	1			<a href="#">312865.0011</a> ...	Germline			<a href="#">rs137852555</a>
SNC	g.15344A>G	c.347A>G	p.Lys116Arg	probable-pathogenic		1	1				Germline			<a href="#">rs193922466</a>
SNC	g.15391C>G	c.394C>G	p.Leu132Val	pathogenic; pathogenic		1	1			<a href="#">312865.0004</a> ...	Germline			<a href="#">rs137852555</a>
SNC	g.15397C>A	c.400C>A	p.Arg134=	probable-non-pathogenic		1	1				Germline			<a href="#">rs193922467</a>
SNC	g.15455G>T	c.458G>T	p.Arg153Leu	pathogenic; pathogenic		1	1			<a href="#">312865.0005</a> ...	Germline			<a href="#">rs137852555</a>
SNC	g.21493C>T	c.502C>T	p.Arg168Trp	pathogenic; pathogenic; pathog...		1	1			<a href="#">312865.0008</a> ...	Germline			<a href="#">rs137852557</a>
SNC	g.21508C>T	c.517C>T	p.Arg173Cys	pathogenic; pathogenic		1	1			<a href="#">312865.0007</a> ...	Germline			<a href="#">rs137852556</a>
SNC	g.21694C>T	c.583C>T	p.Arg195Ter	pathogenic; pathogenic; pathog...		1	3		6	<a href="#">312865.0001</a> ...	Germline			<a href="#">rs137852555</a>
SNC	g.21708C>G	c.597C>G	p.Tyr199Ter	pathogenic; pathogenic		1	1			<a href="#">312865.0002</a> ...	Germline			<a href="#">rs137852555</a>
SNC	g.25291T>C	c.633+3547T>C		pathogenic; pathogenic		1	1			<a href="#">312865.0012</a> ...	Germline			<a href="#">rs137852555</a>
SNC				pathogenic; pathogenic		1	1			<a href="#">312865.0002</a> ...	Germline			<a href="#">rs137852555</a>

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ

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

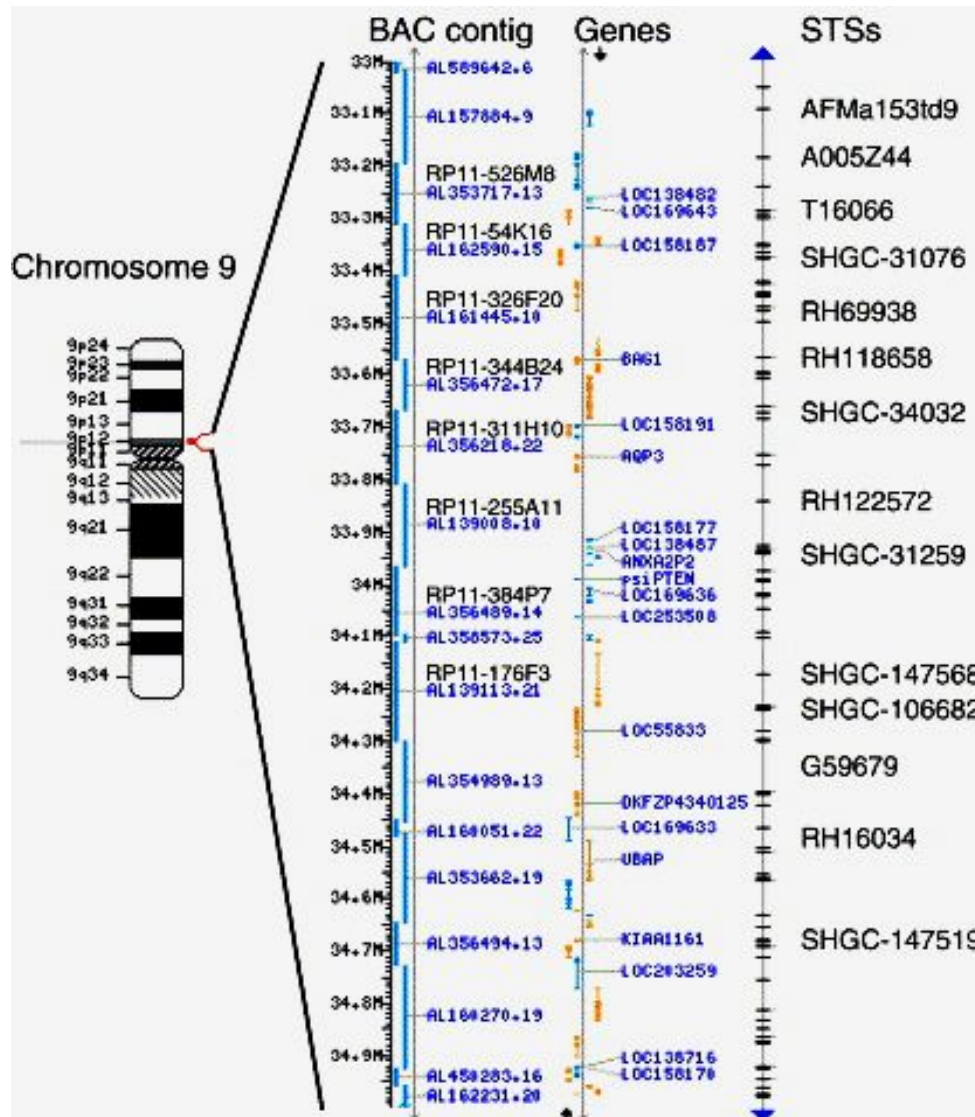
**Коммерчески доступные платформы генотипирования  
на микрочипах высокой плотности**

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



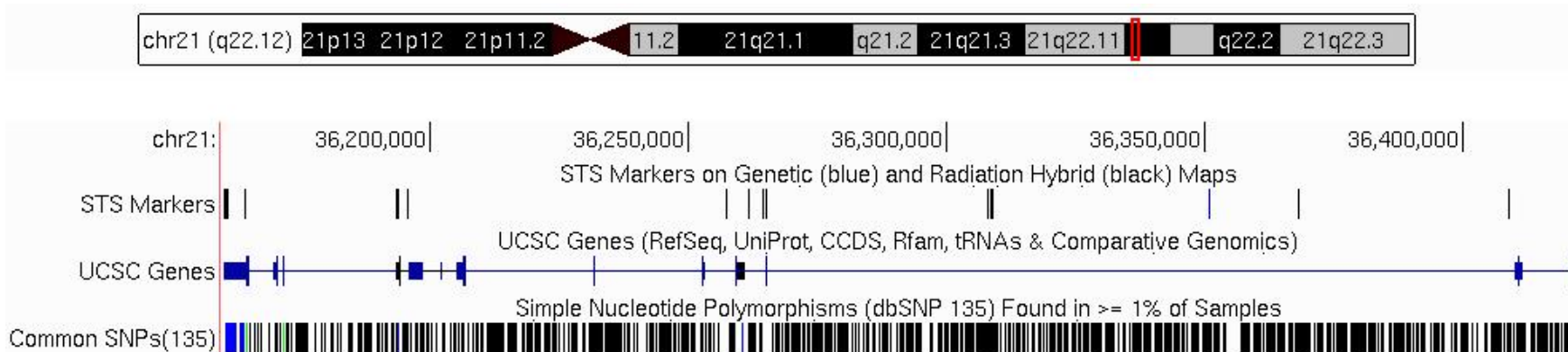
Группа STS-маркеров в 11-й хромосоме человека

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



Группа STS-маркеров в 9-й хромосоме человека

# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ



## STS-маркеры гена RUNX1 человека



# КАРТИРОВАНИЕ ГЕНОВ, КОНТРОЛИРУЮЩИХ ЗАБОЛЕВАНИЯ ЧЕЛОВЕКА ГЕНЕТИЧЕСКОЙ ПРИРОДЫ

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## STS Marker AFMB280XD9

**Chromosome:** chr21  
**Start:** 36350985  
**End:** 36351354  
**Band:** 21q22.12

**Other names:** D21S1895, SHGC-21164, RH31492, B280XD9, W6371, RH49565, RH72566, HSB280XD9, STSG17506

**UCSC STS id:** 4957  
**UniSTS id:** [16185](#)  
**Genbank:** [Z53307](#)  
**GDB:** GDB:610035  
**Organism:** Homo sapiens

**Left Primer:** AGTCCTACTGATAAACTGTGGGC  
**Right Primer:** CTGTCTCATAAGAACCTACCTGG  
**Distance:** 224-282 bps

### Genetic Map Positions

	Name	Chromosome	Position
<b>Genethon:</b>	AFMB280XD9	chr21	37.20
<b>Marshfield:</b>	AFMB280XD9	chr21	33.84

### RH Map Positions

	Name	Chromosome	Position (LOD)
<b>GM99 G3:</b>	RH31492	chr21	984.00 (3.50)
<b>Stanford TNG:</b>	SHGC-21164	chr21	15218.00

**STS-маркер AFMB280XD9**