

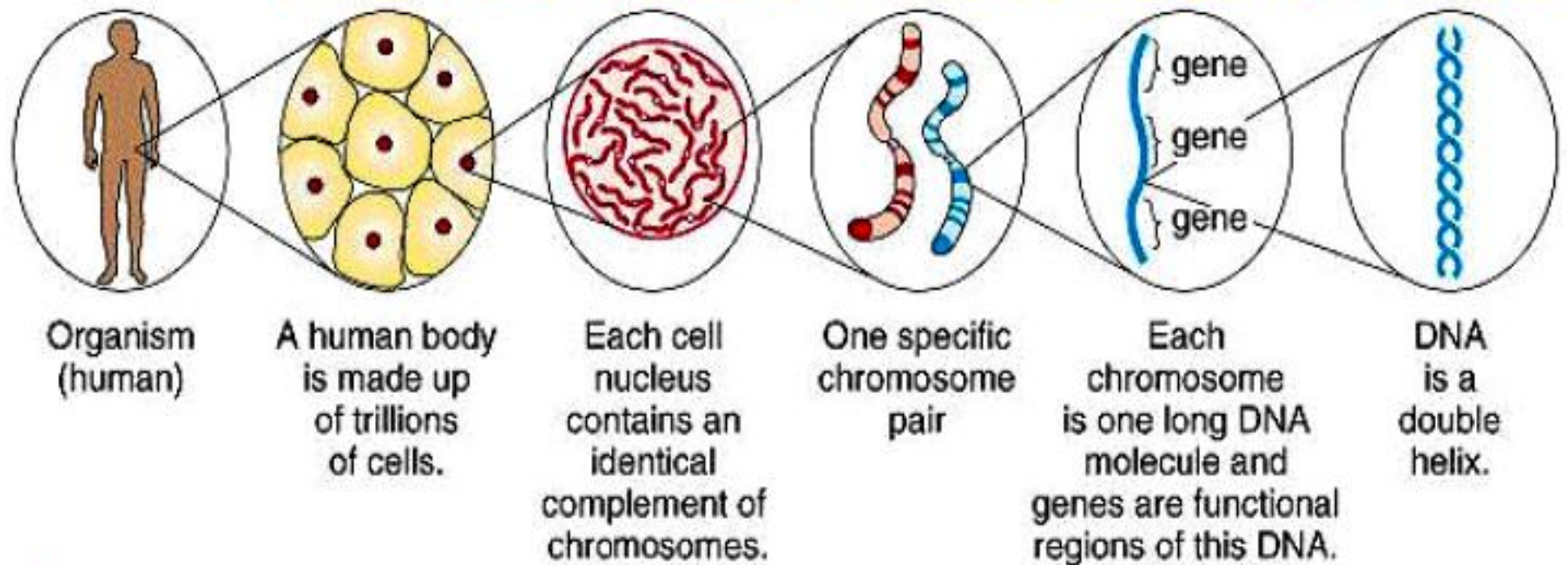
Online Genetic Variation Databases

Department of Biomathematics
University of Thessaly, Larissa
November 2011

Genetics

Scientific study of how physical, biochemical, and behavioral traits are transmitted from parents to their offspring. In essence, the science of heredity.

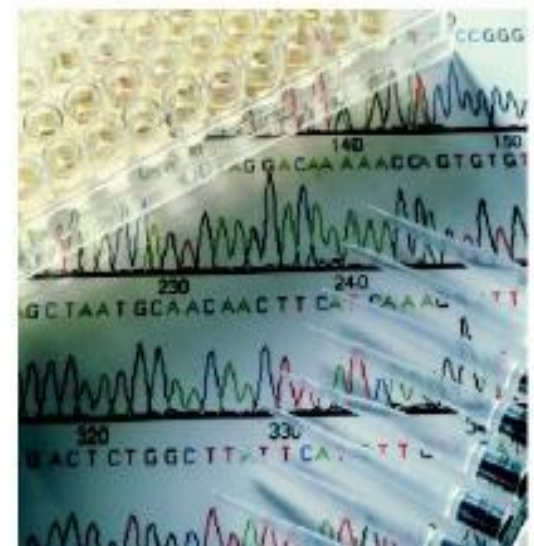


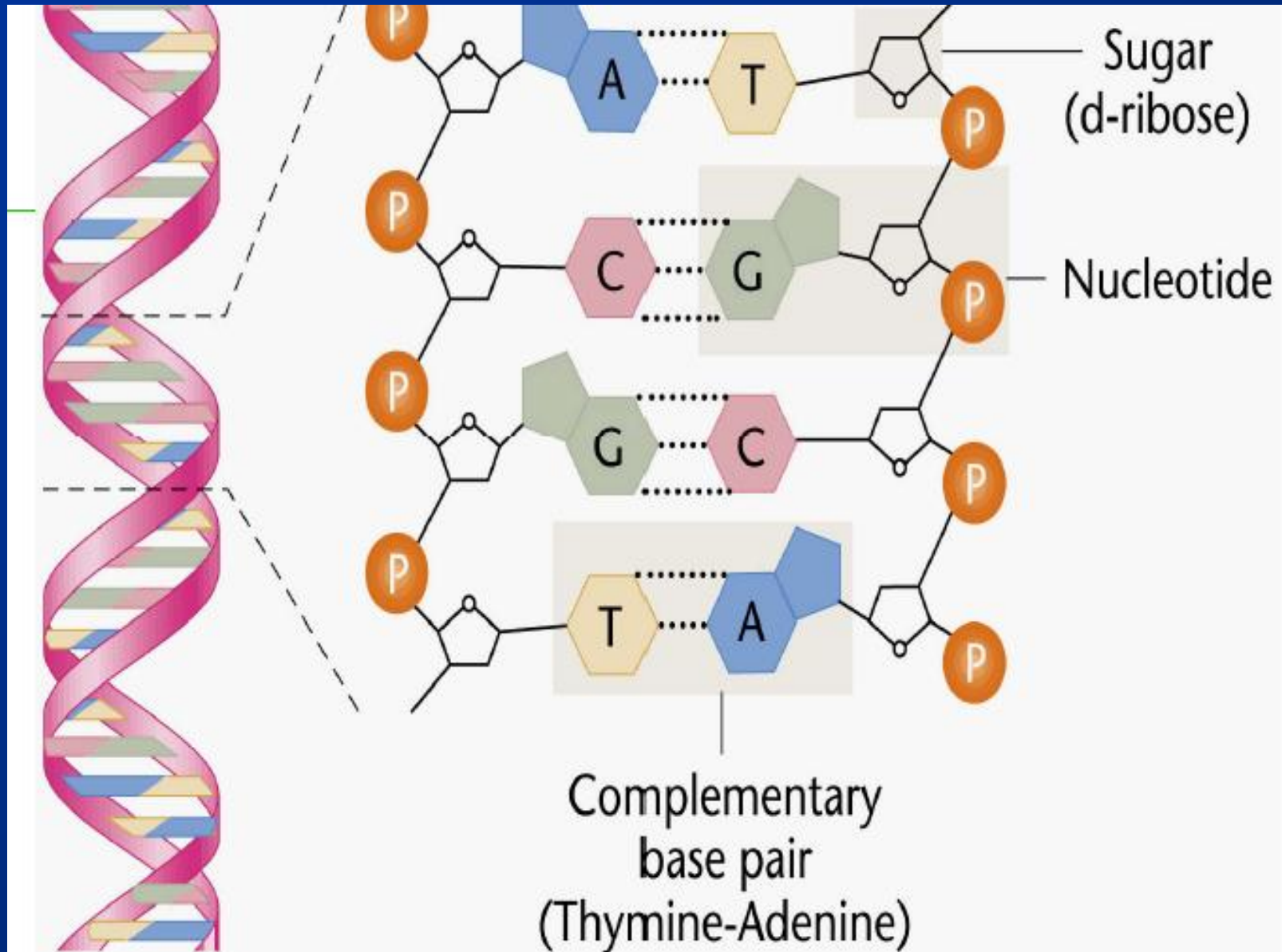


Genetic information contained in DNA

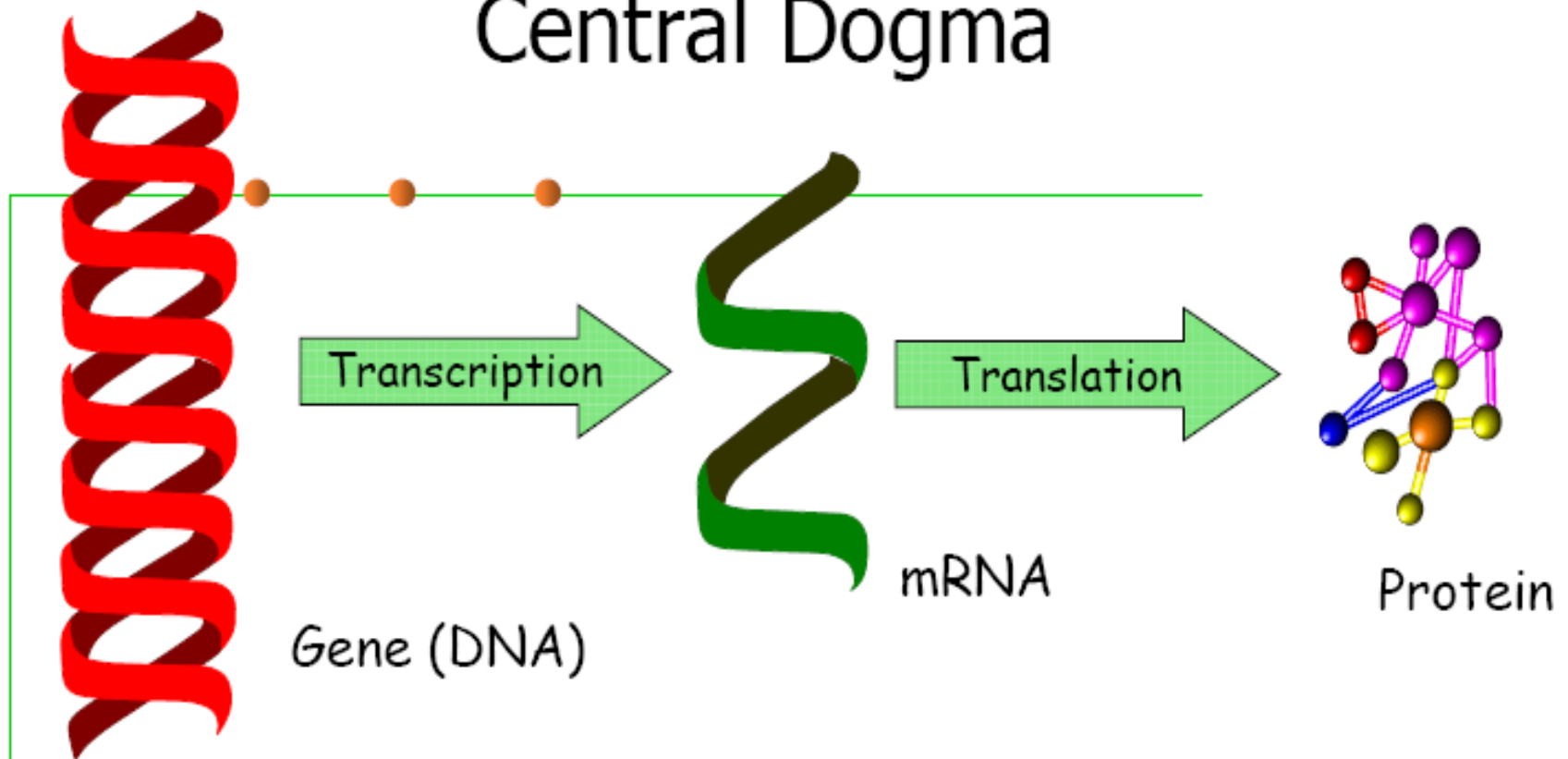
DNA is composed of nucleotides

A gene is a unique sequence of nucleotides

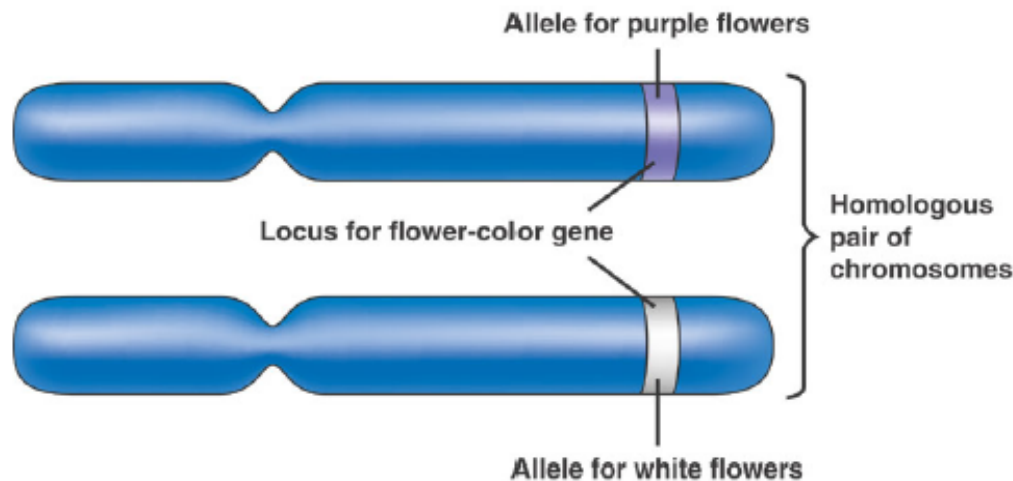




Central Dogma



Some Genetic Terms

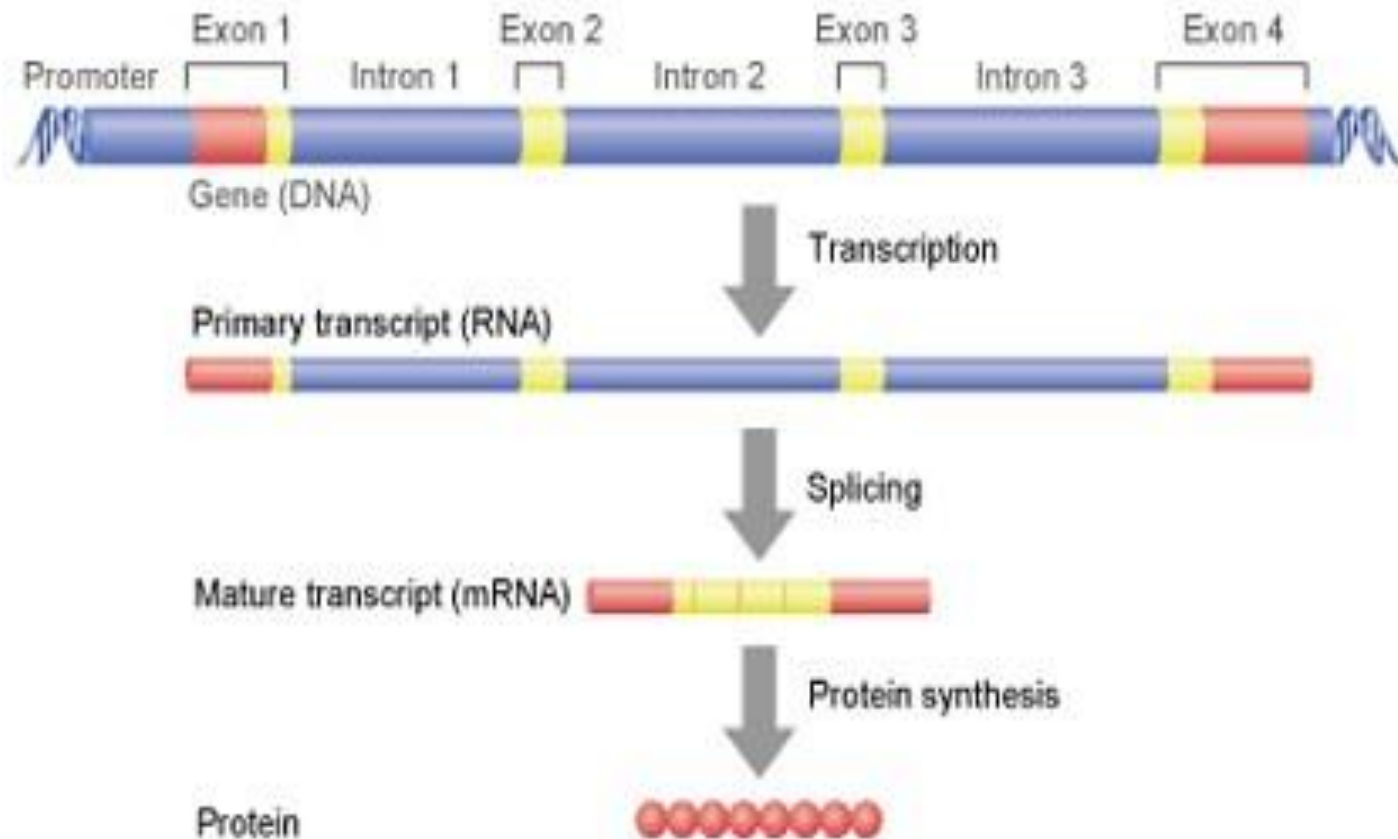


- **Genes** are instructions for producing a trait
- **Locus** is the spot each genes has on a chromosome
- Diploid cells have **two genes (a gene pair)** for each trait, each on a homologous chromosome
- **Alleles** are various molecular forms of a gene encoding for the same trait (i.e. flower color)

Τι είναι γονίδιο

- Βασική μονάδα κληρονομικότητας
- Ακολουθία βάσεων η οποία μεταφέρει την πληροφορία που απαιτείται για να δημιουργηθεί μια συγκεκριμένη πρωτεΐνη
- Ένα γονίδιο κωδικοποιεί μια πρωτεΐνη ή ένα μόριο RNA

Structure of a Gene



HUMAN INDIVIDUALITY





DNA sequence of all human beings is 99.9% identical

⇒ **Our DNAs differ by 0.1%.**

⇒ **Does it make a difference ?**

Yes

0.1% difference translates into 3 million separate “spelling” differences in a genome of 3 billion bases

**Small variations in genotype can make
big differences to phenotype (1.24%)**



Two unique
individuals

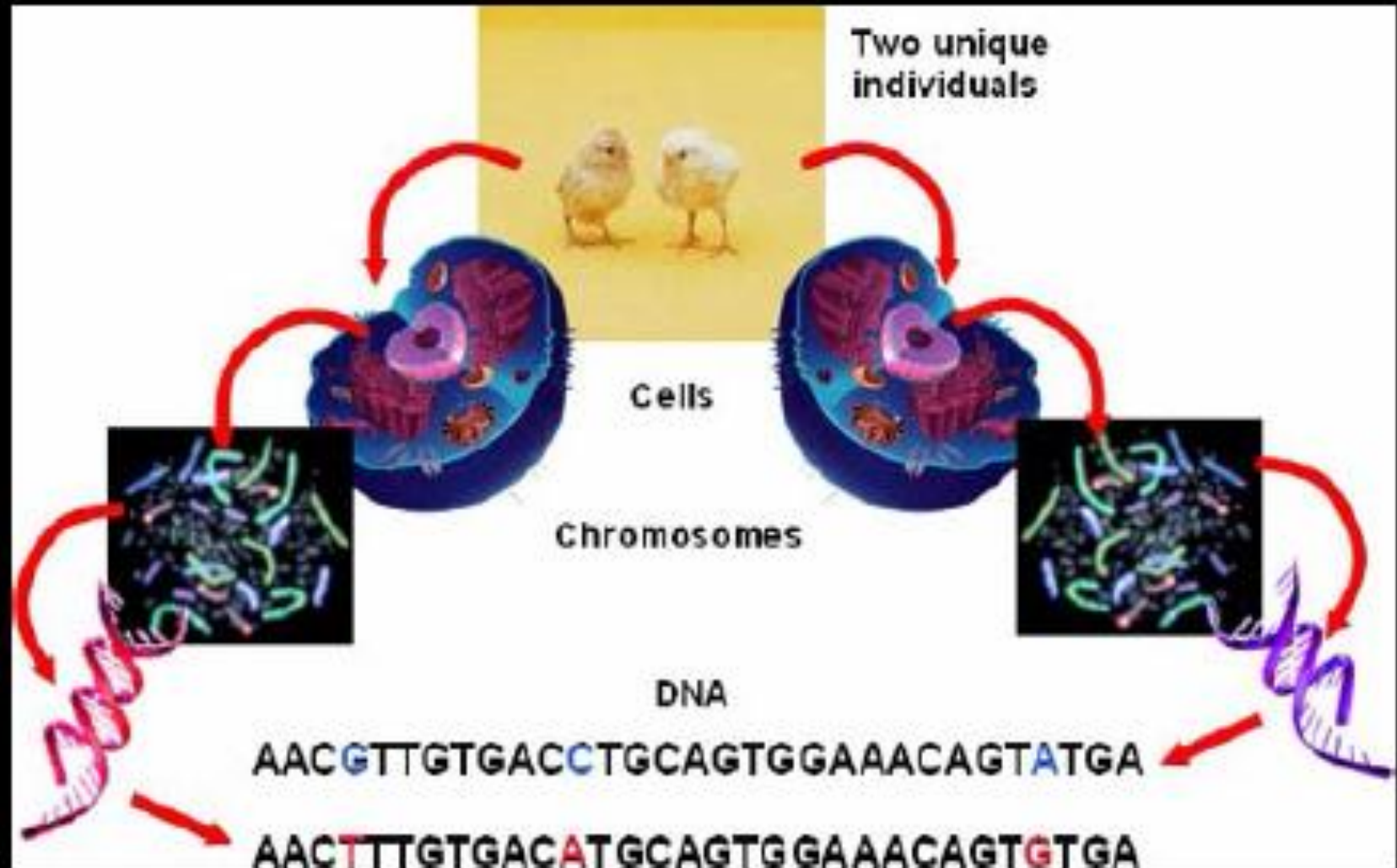
Cells

Chromosomes

DNA

AACGTTGTGACCTGCAGTGGAAACAGTATGA

AACTTTGTGACATGCAGTGGAAACAGTGTGA



definitions

- **Polymorphism** : a variation in nucleotide sequence at a given position in the genome, seen in the population at a frequency of 1% or higher
- **Mutation** : a change in nucleotide sequence which can occur in germ line cells or somatic cells. In germ line cells, mutations can be passed on to offspring who inherit the mutation. Mutations generally have a frequency of less than 1% in the population.
- **Allele** : the specific variant (nucleotide) of a polymorphism at a single locus on a chromosome; for example A, C, G or T

POLYMORPHISMS

SNP

- Missense
- Nonsense
- Silent
- Frameshift
- Splice site

INSERTIONS

- Missense
- Nonsense
- Frameshift

DELETIONS

- Missense
- Nonsense
- Frameshift

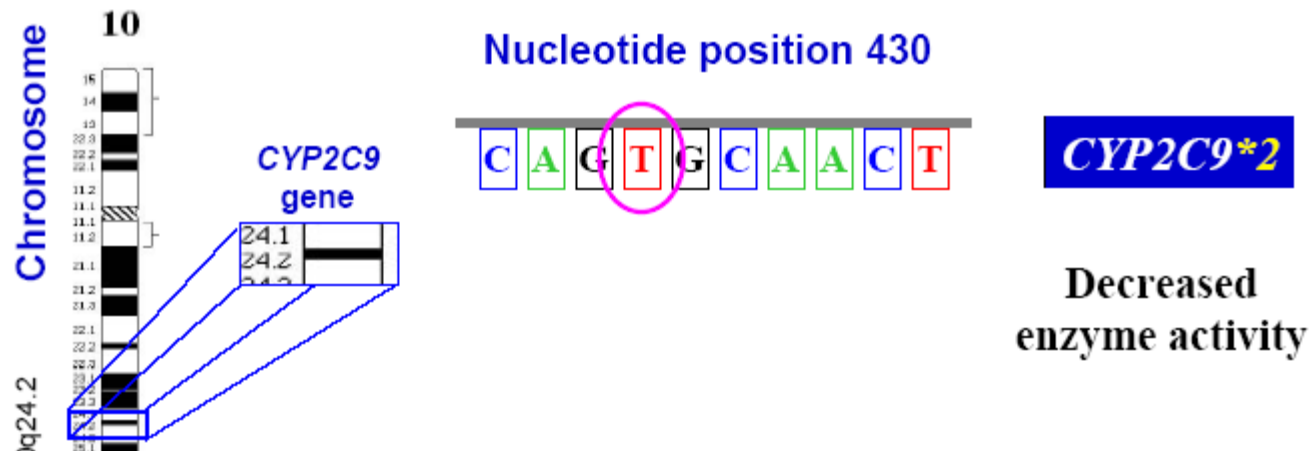
Single nucleotide polymorphisms (SNP)

.....G G **T** A A C T T G

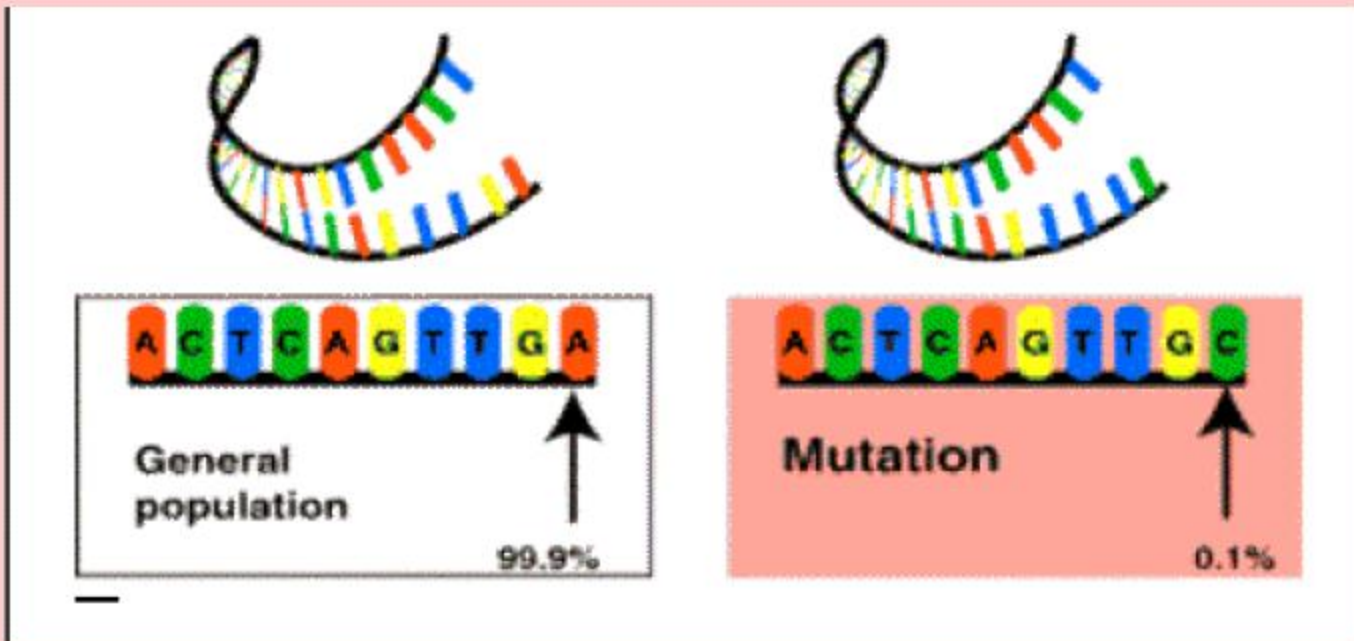
.....G G **C** A A C T T G

- Most common, Incidence 1 per 300 - 600bp

Example:



Mutation



SNPs

(pronounced snips)

Polymorphism

"Poly" *many* "morph" *form*



A C T C A G T T G A

General
population

94%

A C T C A G T T T A

Single nucleotide
polymorphism
(SNP)

6%

Mendelian Traits in Humans



Recessive phenotype: Albinism

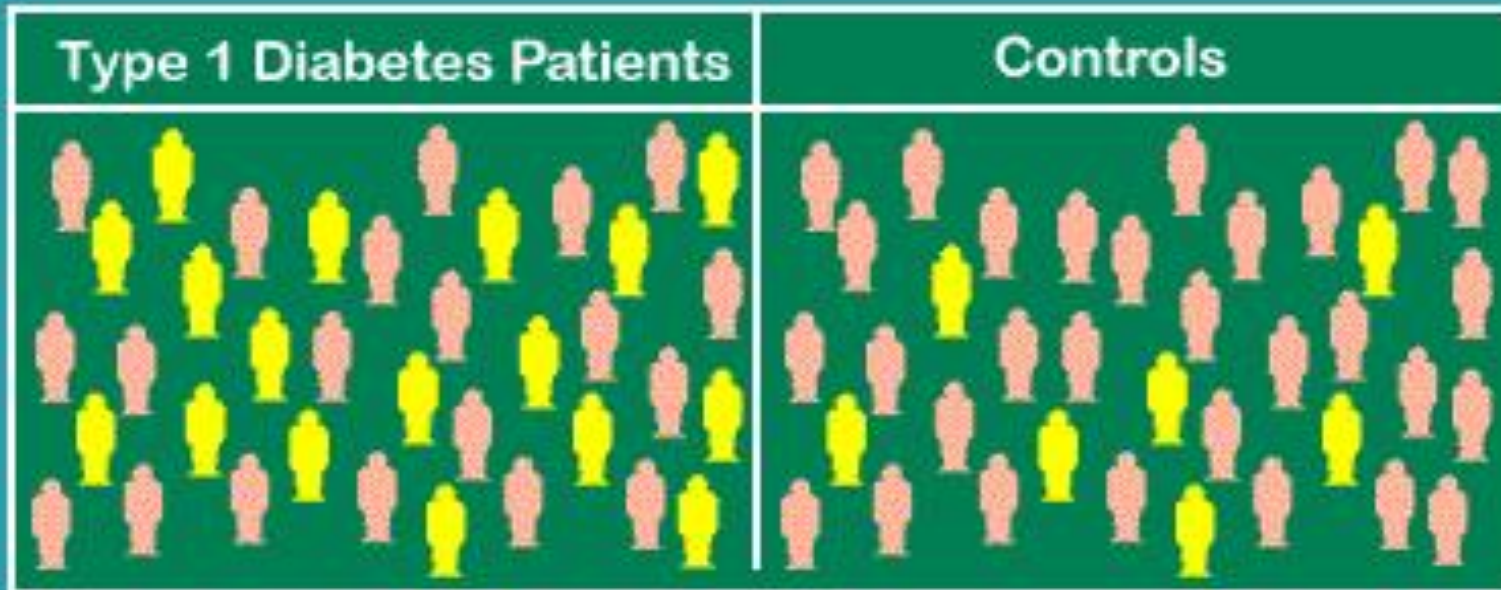


Dominant phenotype: Achondroplasia

- However, common Multifactorial disorders result from polymorphisms in multiple genes, often coupled with environmental causes. The complicated bases of these diseases make them difficult to study and to treat. Heart disorder, diabetes and cancer are examples of this type of disorder.

- SNPs may fall within coding sequences of genes, non-coding regions of genes, or in the intergenic regions between genes. SNPs within a coding sequence will not necessarily change the aminoacid sequence of the protein that is produced, due to degeneracy of the genetic code. A SNP in which both forms lead to the same polypeptide sequence is termed *synonymous* - if a different polypeptide sequence is produced they are *non-synonymous*.
- Variations in the DNA sequences of humans can affect how humans develop diseases and respond to pathogens, chemicals, drugs, vaccines, and other agents. However, their greatest importance in biomedical research is for comparing regions of the genome between cohorts (such as with matched cohorts with and without a disease).
- **Genetic association studies**

Association Studies



Genotype	Type 1	Controls	Total
HLA DR4	17	7	24
NON-HLA DR4	20	30	50
	37	37	

$$\chi^2_{.05} = 5.377$$

$$p < 0.025$$

Odds Ratio: 3.6
95% CI = 1.3 to 10.4

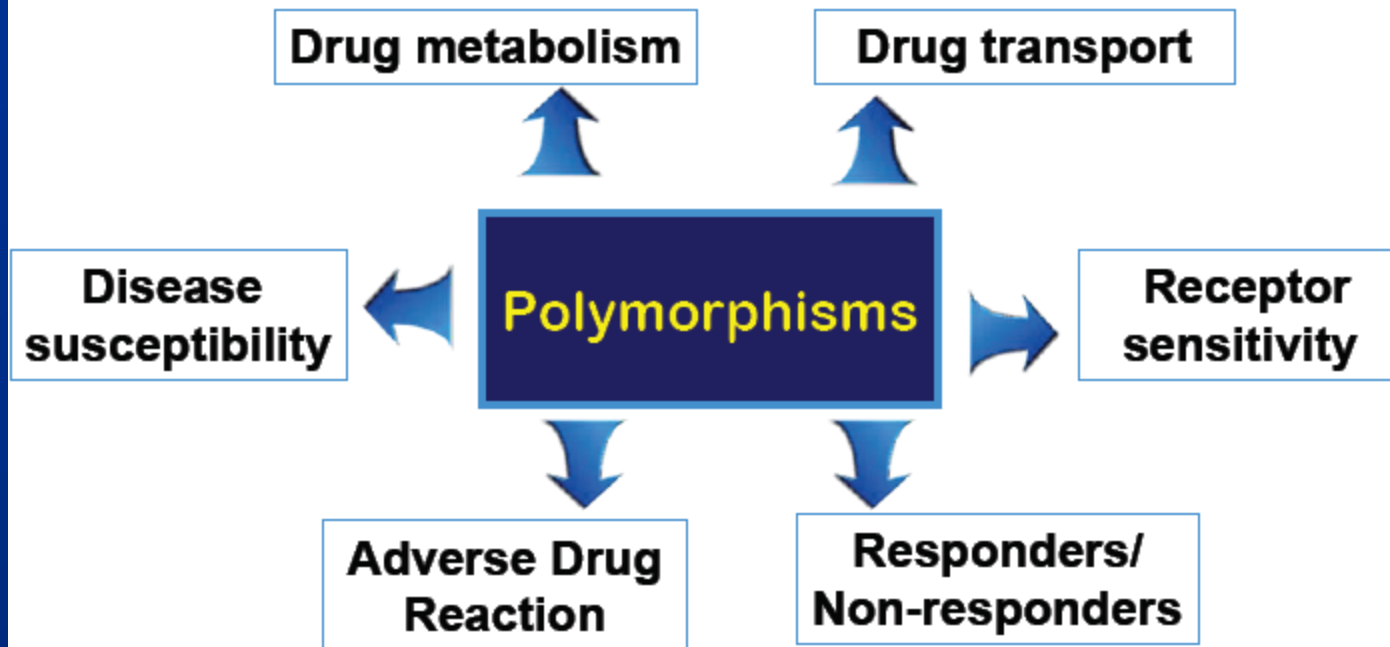


= HLA DR4

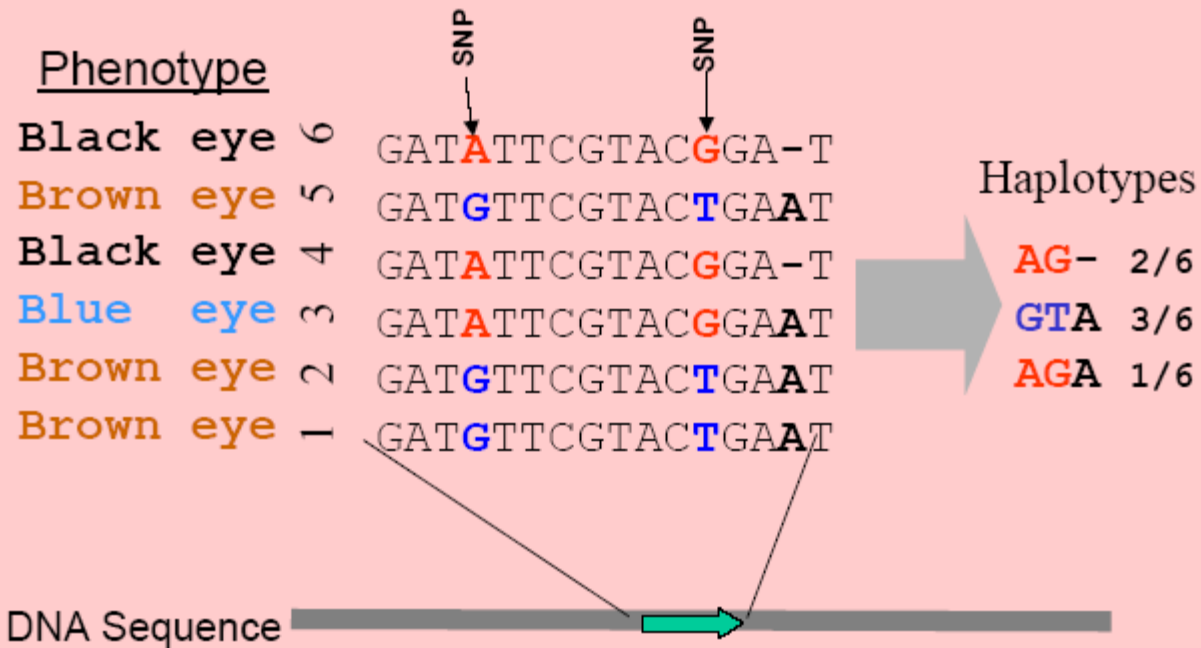


= non-HLA DR4

Consequences of polymorphisms



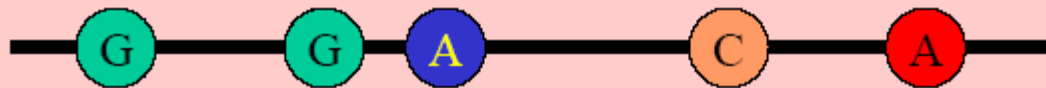
From SNP to Haplotype



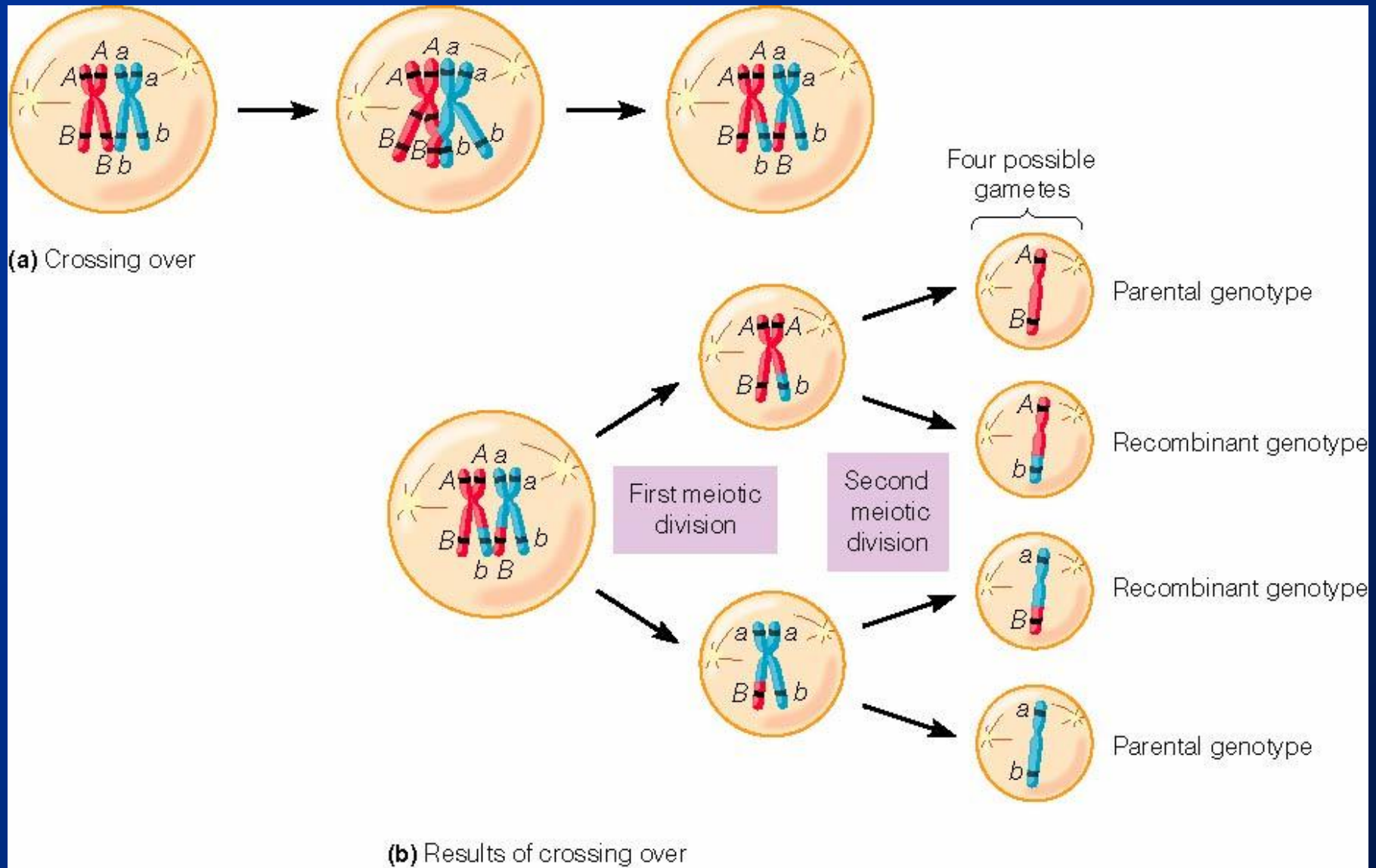
- Haplotype : the combination of two or more alleles at physically neighbouring positions within the genome

Haplotype

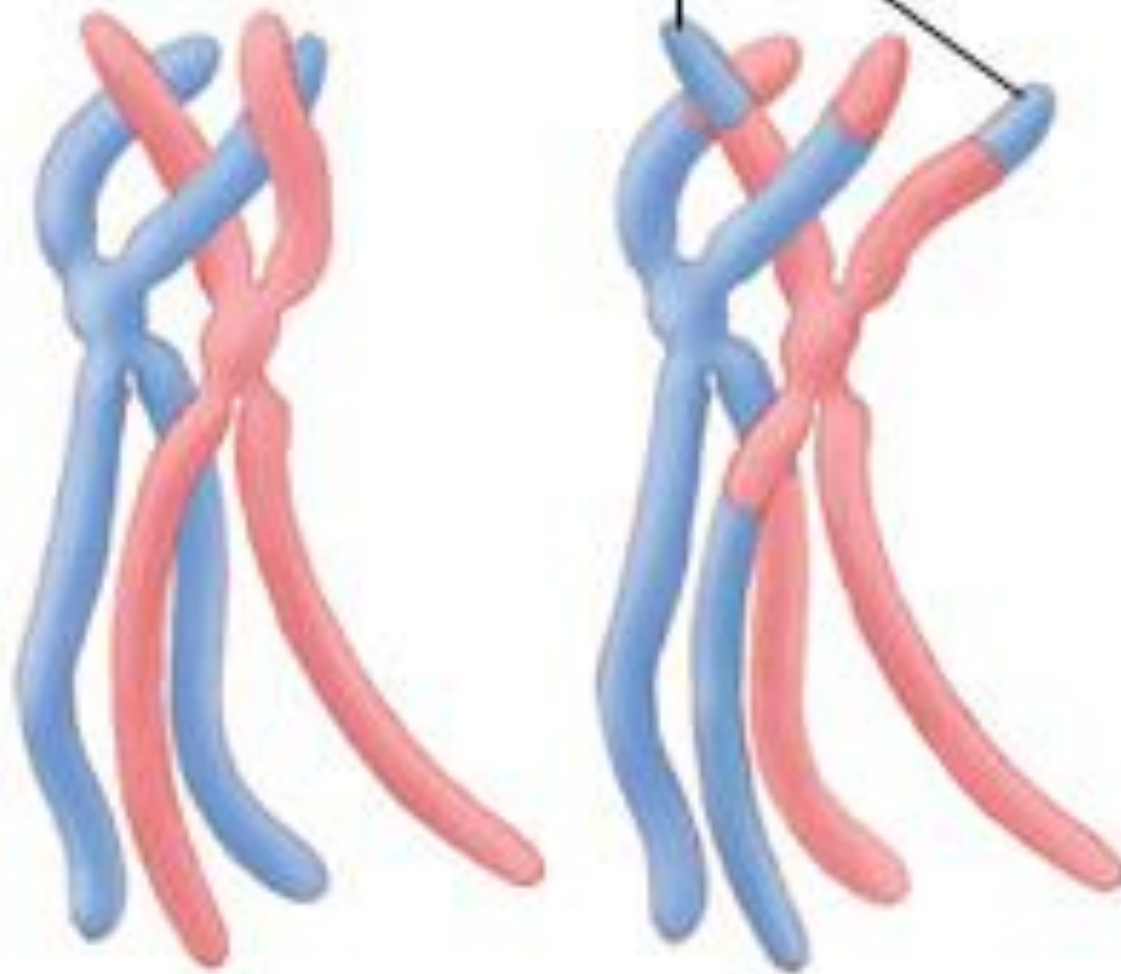
- Haplotype: A set of closely linked genetic markers present on one chromosome which tend to be inherited together (not easily separable by recombination).



Set of SNP polymorphisms: a SNP haplotype



Crossing Over Points



Linkage disequilibrium

- The occurrence together on the same chromosome of specific alleles at closely linked loci more frequently than would be expected by chance. Because linkage disequilibrium is a function of the distance between loci, it can be used to help infer the order of genes on chromosomes.
- LD metrics: D , r^2 , LOD

PERSPECTIVES

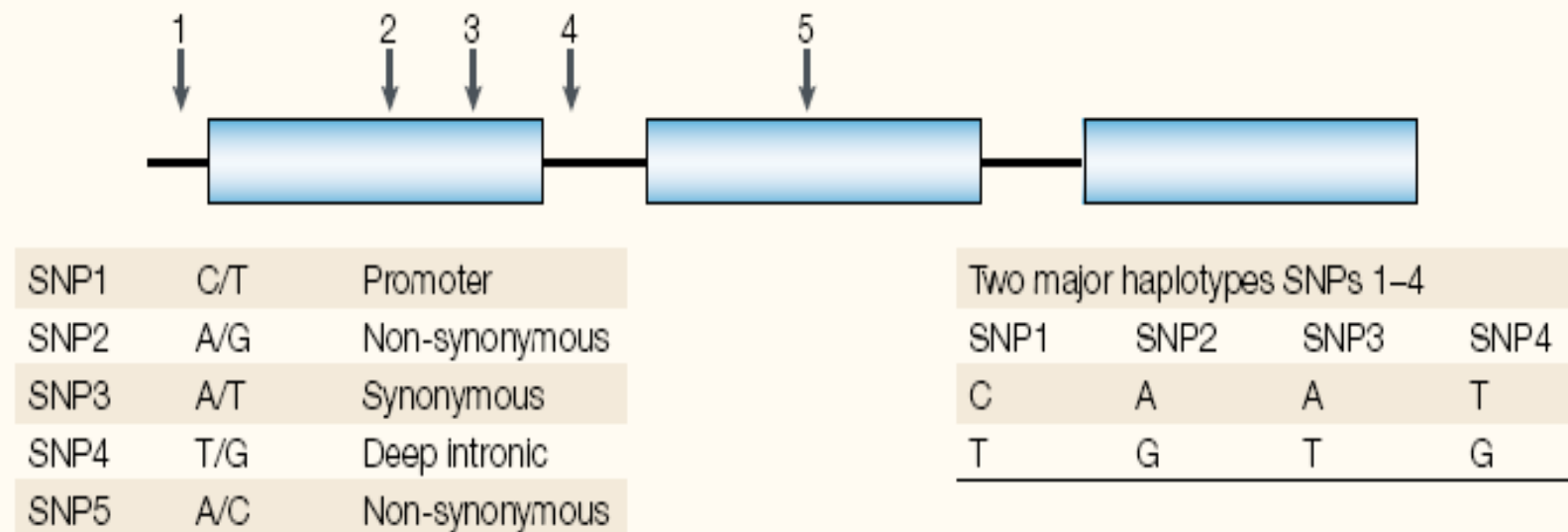
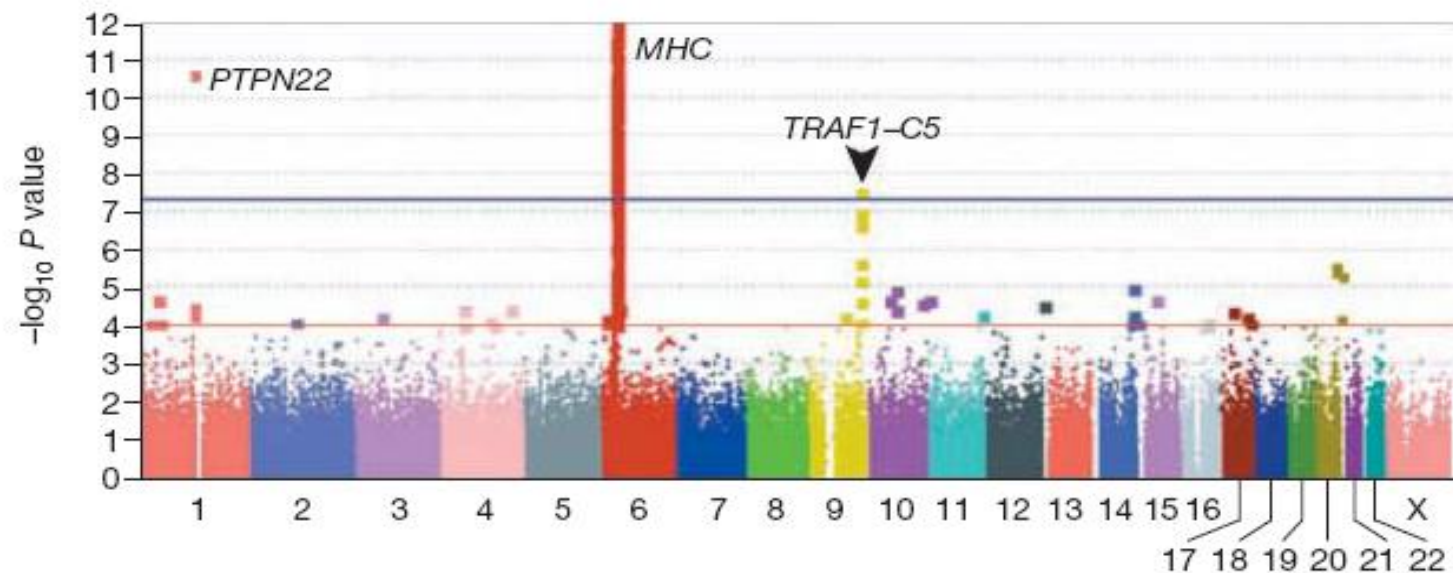


Figure 1 | **Haplotypes and linkage disequilibrium in single-nucleotide-polymorphism selection.** Single-nucleotide polymorphisms (SNPs) 1–4 are in linkage disequilibrium and form two common haplotypes, which can be characterized by any of the variants. Therefore, it is only necessary to determine the genotype of one of the SNPs to capture the information about all four SNPs. SNP5 is not in linkage disequilibrium with SNPs 1–4 and is not part of the haplotype, but it might still contribute independently to the risk of the phenotype.

Genome wide association studies

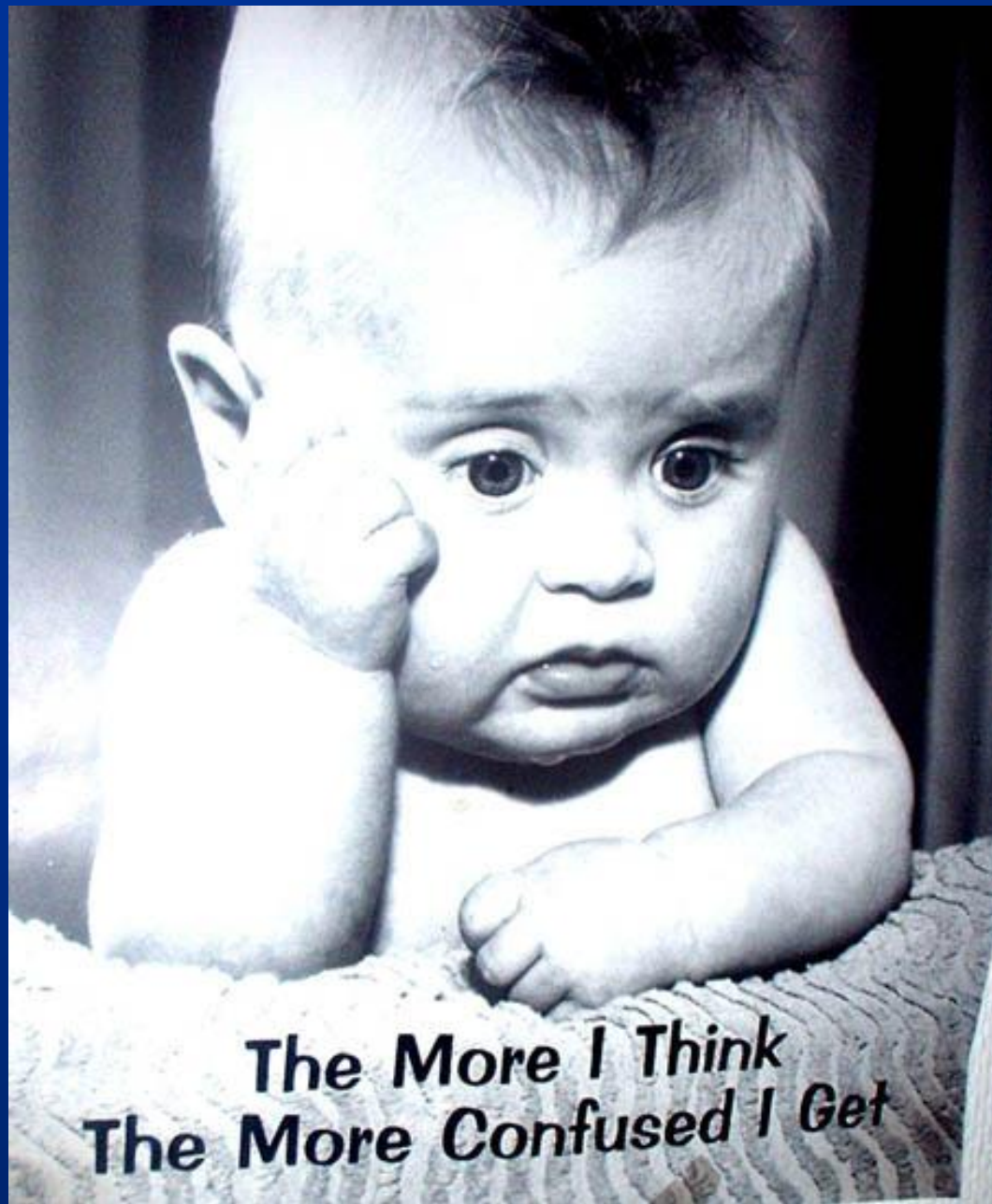
Figure 3. Genome-wide Association Findings in Rheumatoid Arthritis



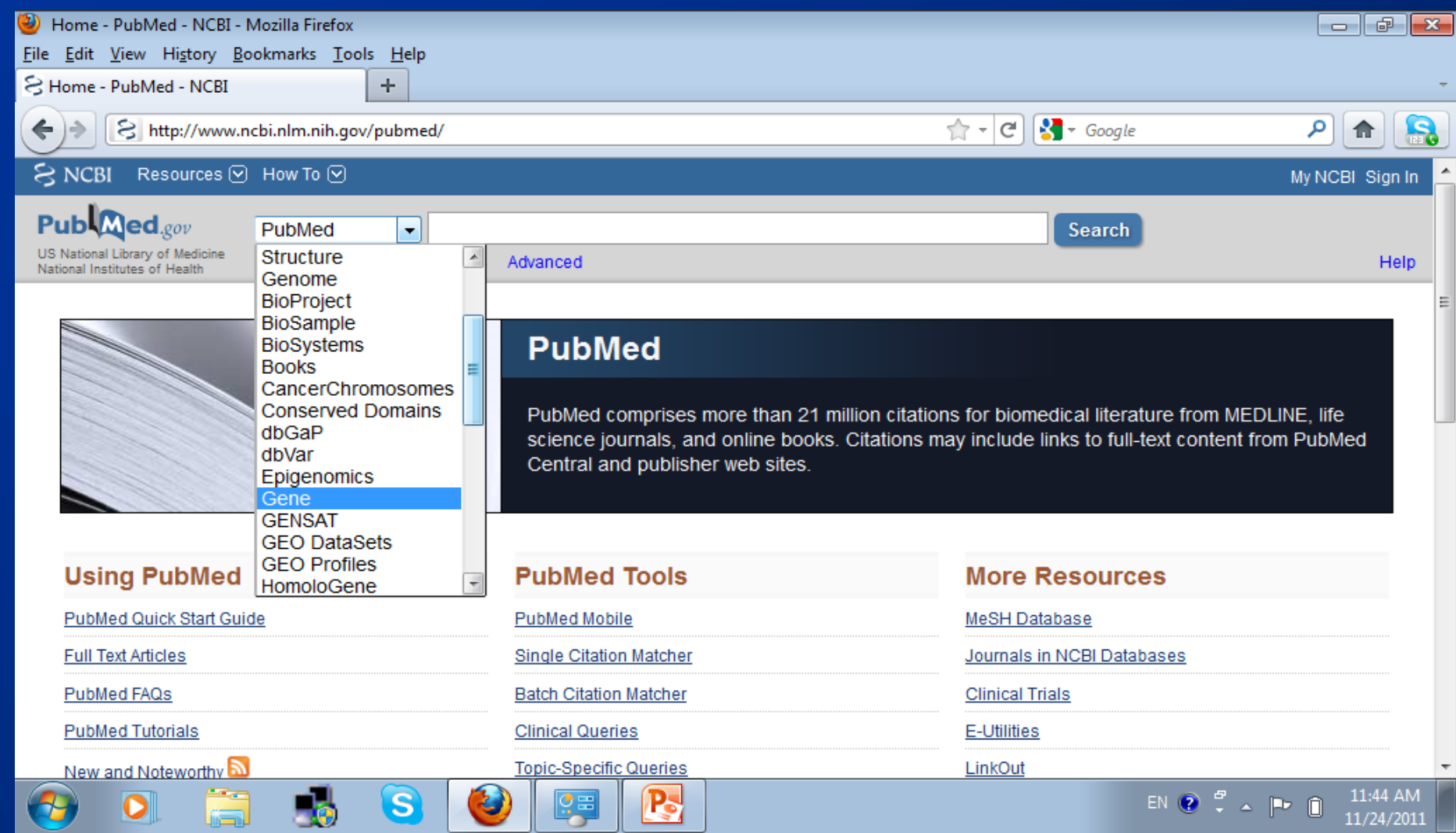
Genome-wide association studies assume a priori hypotheses about candidate genes or regions that might be associated with disease; rather, they test single-nucleotide polymorphisms (SNPs) throughout the genome for possible evidence of genetic susceptibility. Associations plotted as $-\log_{10} P$ values for a genome-wide association study in 1522 cases with rheumatoid arthritis and 1850 controls, showing single data points for SNPs with $P < 10^{-4}$ (lower horizontal red line) for 22 autosomes and the X chromosome. The predefined level of significance, at 5×10^{-8} is shown with a horizontal blue line. SNPs at *PTPN22* on chromosome 1, the major histocompatibility complex (MHC) on chromosome 6, and the *TRAF1-C5* locus on chromosome 9 exceed this threshold. Reproduced with permission from Plenge et al.⁴⁷

Overview

- Gene
- Polymorphism
- Mutation
- Genetic association studies
- Haplotype
- Linkage disequilibrium
- Genome-wide association studies



Example: MTHFR gene. Go to PubMed (www.ncbi.nlm.nih.gov/pubmed/) and type MTHFR and search GENE



Click MTHFR for Homo Sapiens

MTHFR - Gene - NCBI - Mozilla Firefox

File Edit View History Bookmarks Tools Help

MTHFR - Gene - NCBI

http://www.ncbi.nlm.nih.gov/gene?term=MTHFR

NCBI Resources How To My NCBI Sign In

Gene Gene MTHFR Search

Save search Limits Advanced Help

Display Settings: Summary, 20 per page, Sorted by Relevance Send to: Filter your results:

Results: 1 to 20 of 1750

<< First < Prev Page 1 of 88 Next > Last >>

☐ **MTHFR**

1. **Official Symbol:** MTHFR and **Name:** methylenetetrahydrofolate reductase (NAD(P)H) [*Homo sapiens*]
Other Aliases: RP11-56N19.4
Other Designations: 5,10-methylenetetrahydrofolate reductase (NADPH); methylenetetrahydrofolate reductase
Chromosome: 1; **Location:** 1p36.3
Annotation: Chromosome 1, NC_000001.10 (11845787..11866160, complement)
MIM: 607093
ID: 4524
[Order cDNA clone](#)

☐ **Mthfr**

2. **Official Symbol:** Mthfr and **Name:** 5,10-methylenetetrahydrofolate reductase [*Mus musculus*]
Other Aliases: RP23-139J21.2, AI323986
Other Designations: methylenetetrahydrofolate reductase

[Manage Filters](#)

▼ **Top Organisms** [Tree]

- Homo sapiens (70)
- Desulfatibacillum alkenivorans AK-01 (10)
- Desulfobacca acetoxidans DSM 11109 (10)
- Desulfobacterium autotrophicum HRM2 (9)
- Mus musculus (8)
- All other taxa (1643)
- More...

http://www.ncbi.nlm.nih.gov/pubmed/clinical cM

EN 11:44 AM 11/24/2011

Available information regarding :Summary, Genomic regions, transcripts, Genomic context, Bibliography, Interactions, General gene information General protein information, Reference Sequences, Related Sequences

MTHFR methylenetetrahydrofolate reductase (NAD(P)H) [Homo sapiens] - Gene - NCBI - Mozilla Firefox

File Edit View History Bookmarks Tools Help

Πανεπιστήμιο Θεσσαλίας Mail :: Inbox: [allstudents] Fwd: MTHFR methylenetetrahydrofolate re...

http://www.ncbi.nlm.nih.gov/gene/4524

NCBI Resources How To My NCBI Sign In

Gene Gene Search Limits Advanced Help

Display Settings: Full Report Send to:

MTHFR methylenetetrahydrofolate reductase (NAD(P)H) [Homo sapiens]
Gene ID: 4524, updated on 19-Nov-2011

Summary

Official Symbol MTHFR provided by HGNC
Official Full Name methylenetetrahydrofolate reductase (NAD(P)H) provided by HGNC
Primary source HGNC:7436
Locus tag RP11-56N19.4
See related Ensembl:ENSG00000177000; HPRD:06158; MIM:607093; Vega:OTTHUMG00000002277
Gene type protein coding
RefSeq status REVIEWED
Organism Homo sapiens
Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Summary The protein encoded by this gene catalyzes the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a co-substrate for homocysteine remethylation to methionine. Genetic

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- General gene info
- General protein info
- Reference sequences
- Related sequences
- Additional links

Links

Order cDNA clone

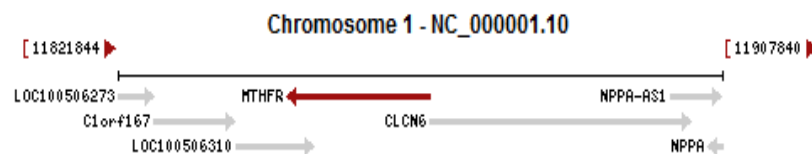
EN 10:54 PM 11/23/2011

Genomic context

Location: 1p36.3

See MTHFR in [Epigenomics](#), [MapViewer](#)

Sequence: Chromosome: 1; NC_000001.10 (11845787..11866160, complement)

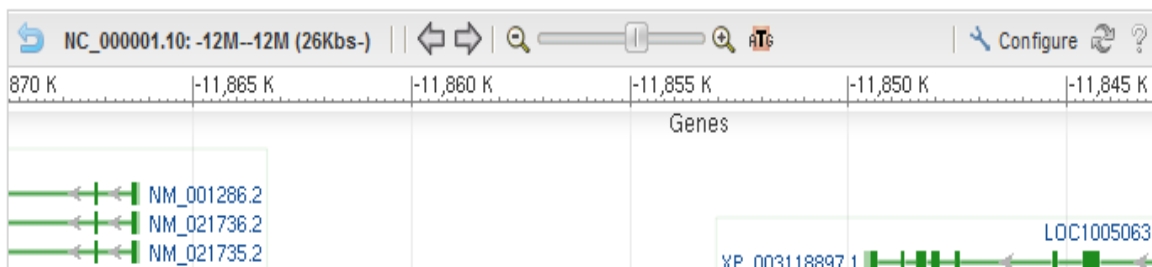


Genomic regions, transcripts, and products

Genomic Sequence NC_000001 chromosome 1 reference GRCh37.p5 Prim

Go to [reference sequence details](#)

Go to nucleotide [Graphics](#) [FASTA](#) [GenBank](#)



- Books
- CCDS
- Conserved Domains
- dbVar
- EST
- Full text in PMC
- Genome
- GEO Profiles
- HomoloGene
- Map Viewer
- Nucleotide
- OMIM
- Probe
- Protein
- PubChem Compound
- PubChem Substance
- PubMed
- PubMed (GeneRIF)
- PubMed (OMIM)
- RefSeq Proteins

Have a look to the references from PubMed...and how the gene and its SNPs are named in the literature...

MTHFR methylenetetrahydrofolate reductase (NAD(P)H) [Homo sapiens] - Gene - NCBI - Mozilla Firefox

File Edit View History Bookmarks Tools Help

MTHFR methylenetetrahydrofolate redu... +

http://www.ncbi.nlm.nih.gov/gene/4524

1 1 1

Bibliography

Related articles in PubMed

[Gene polymorphisms MTHFR C677T and MTR A2756G as predictive factors in adjuvant chemotherapy for stage III colorectal cancer](#). Tafin H, *et al.* Anticancer Res, 2011 Sep. PMID 21868559.

[\[The role of 1691G>A\(Leiden\) mutation in Factor V gene, 20210G>A in prothrombin gene and 677C>T in MTHFR gene in etiology of early pregnancy loss\]](#). Slezak R, *et al.* Ginekol Pol, 2011 Jun. PMID 21853935.

[Nutrients and genetic variation involved in one-carbon metabolism and Hodgkin lymphoma risk: a population-based case-control study](#). Kasperzyk JL, *et al.* Am J Epidemiol, 2011 Oct 1. PMID 21810727.

[\[Allelic polymorphism of MTHFR, MTR and MTRR genes in patients with cleft lip and/or palate and their mothers\]](#). Chorna LB, *et al.* Tsitol Genet, 2011 May-Jun. PMID 21774403.

[Methylenetetrahydrofolate reductase \(MTHFR\) genotype, smoking habit, metastasis and oral cancer in Taiwan](#). Tsai CW, *et al.* Anticancer Res, 2011 Jun. PMID 21737671.

[See all \(2264\) citations in PubMed](#)

[See citations in PubMed for homologs of this gene provided by HomoloGene](#)

Links to other resources

- AceView
- Ensembl
- Evidence Viewer
- GeneTests for MIM: 236250
- GeneTests for MIM: 607093
- HGNC
- HPRD
- HuGE Navigator
- KEGG
- MGC
- ModelMaker
- PharmGKB
- Reactome
- UCSC
- Vega

General information

About Gene

GeneRIFs: Gene References Into Functions [What's a GeneRIF?](#)

11:03 PM 11/23/2011

click on the link MIM 607093, hyperlink to OMIM database

MTHFR methylenetetrahydrofolate reductase (NAD(P)H) [Homo sapiens] - Gene - NCBI - Mozilla Firefox

File Edit View History Bookmarks Tools Help

Πανεπιστήμιο Θεσσαλίας x Mail :: Inbox: [allstudents] Fwd: x MTHFR methylenetetrahydrofolate re... x +

http://www.ncbi.nlm.nih.gov/gene/4524

NCBI Resources How To My NCBI Sign In

Gene Gene Search Limits Advanced Help

Display Settings: Full Report Send to:

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Gene ID: 4524, updated on 19-Nov-2011

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- General protein info
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- Additional links

Links

- Order cDNA clone

EN 10:54 PM 11/23/2011

High quality description of the gene in text format

OMIM Entry - *607093 - 5,10-METHYLENETETRAHYDROFOLATE REDUCTASE; MTHFR - Mozilla Firefox

File Edit View History Bookmarks Tools Help

OMIM Entry - *607093 - 5,10-METHYLEN... +

http://omim.org/entry/607093

*607093

5,10-METHYLENETETRAHYDROFOLATE REDUCTASE;
MTHFR

HGNC Approved Gene Symbol: [MTHFR](#)

Cytogenetic location: [1p36.22](#) *Genomic coordinates (GRCh37):* [1:11,845,786 - 11,866,159](#)
(from NCBI)

Gene Phenotype Relationships

Location	Phenotype	Phenotype MIM number
1p36.22	Homocystinuria due to MTHFR deficiency	236250
	{Neural tube defects, susceptibility to}	601634
	{Schizophrenia, susceptibility to}	181500
	{Thromboembolism, susceptibility to}	188050
	{Vascular disease, susceptibility to}	

Table of Contents - *607093

External Links:

- Genome
- DNA
- Protein
- Gene Info
- Clinical Resources
- Variation
- Animal Models
- Cellular Pathways

EN ? 10:56 PM 11/23/2011

Back to GENE, click on ENSEMBL link

MTHFR methylenetetrahydrofolate reductase (NAD(P)H) [Homo sapiens] - Gene - NCBI - Mozilla Firefox

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Πανεπιστήμιο Θεσσαλίας Mail :: Inbox: [allstudents] Fwd: MTHFR methylenetetrahydrofolate re...

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Order cDNA clone

EN 10:54 PM 11/23/2011

Detailed description of the gene with many bioinformatic tools. Click on the location link to view its location...

Ensembl genome browser 64: Homo sapiens - Gene summary - Gene: MTHFR (ENSG00000177000) - Mozilla Firefox

File Edit View History Bookmarks Tools Help

Ensembl genome browser 64: Homo sap... +

http://www.ensembl.org/Homo_sapiens/Gene/Summary?g=ENSG00000177000;r=1:11845780-11866977

Ensembl BLAST/BLAT | BioMart | Tools | Downloads | More

Human (GRCh37) Location: 1:11,845,780-11,866,977 Gene: MTHFR

Gene-based displays

- Gene summary
- Splice variants (9)
- Supporting evidence
- Sequence
- External references
- Regulation
- Comparative Genomics
 - Genomic alignments
- Gene Tree (image)
 - Gene Tree (text)
 - Gene Tree (alignment)
- Orthologues (53)
- Paralogues
- Protein families (3)
- Phenotype
- Genetic Variation
 - Variation Table
 - Variation Image
 - Structural Variation
- External Data
 - Personal annotation

Gene: MTHFR (ENSG00000177000)

Description methylenetetrahydrofolate reductase (NAD(P)H) [Source:HGNC Symbol;Acc:7436]

Location [Chromosome 1: 11,845,780-11,866,977](#) reverse strand.

Transcripts This gene has 9 transcripts

Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
MTHFR-001	ENST00000376592	7057	ENSP00000365777	656	Protein coding	CCDS137
MTHFR-002	ENST00000376590	6232	ENSP00000365775	656	Protein coding	CCDS137
MTHFR-003	ENST00000376486	1135	ENSP00000365669	30	Protein coding	-
MTHFR-004	ENST00000418034	1034	ENSP00000405082	143	Protein coding	-
MTHFR-005	ENST00000423400	1539	ENSP00000398908	53	Protein coding	-
MTHFR-006	ENST00000431243	870	ENSP00000400460	30	Protein coding	-
MTHFR-007	ENST00000376585	5532	ENSP00000365770	697	Protein coding	-
MTHFR-008	ENST00000413656	398	ENSP00000408307	30	Protein coding	-

EN 10:59 PM 11/23/2011

Ensembl genome browser 64: Homo sapiens - Region in detail - Chromosome 1: 11,845,780-11,866,977 - Mozilla Firefox

File Edit View History Bookmarks Tools Help

Ensembl genome browser 64: Homo sap... +

http://www.ensembl.org/Homo_sapiens/Location/View?db=core;g=ENSG00000177000;r=1:11845780-11866977 Google

Login Register

e!Ensembl BLAST/BLAT | BioMart | Tools | Downloads | More ▾

Human (GRCh37) ▾ Location: 1:11,845,780-11,866,977 Gene: MTHFR

Location-based displays

- Whole genome
- Chromosome summary
- Region overview
- Region in detail**
- Comparative Genomics
 - Alignments (image) (56)
 - Alignments (text) (56)
 - Multi-species view (52)
 - Synteny (15)
- Genetic Variation
 - Resequencing (20)
 - Linkage Data
- Markers
- Other genome browsers
 - UCSC
 - NCBI
 - Vega

Configure this page

Manage your data

Chromosome 1: 11,845,780-11,866,977

Assembly exception chromosome 1

Assembly exception HG989_PATCH

HSCHR1_1_CT G31 HSCHR1_3_CT G31
HSCHR1_2_CT G31

Export Image

Region in detail [help](#)

Chromosome bands

Contigs

Ensembl/Havana...

1.00 Mb

Forward strand

11.40 Mb 11.50 Mb 11.60 Mb 11.70 Mb 11.80 Mb 11.90 Mb 12.00 Mb 12.10 Mb 12.20 Mb 12.30 Mb

p35.22

< AL031735.9 > AL357835.11 >

PTCHD2 > FBXO2 < CLorf167 > KIAA2013 < TNFRSF8 > VPS13D < FBXO44 > MTHFR < PLOD1 > TNFRSF1B > FBXO6 < CLCN6 > MFN2 > RP11-426M1.2 < MAD2L2 > NPPA < MIIP > RP5-1 < CLorf187 > NPPB < AGTRAP > RP5-934G17.6 >

EN ?

11:01 PM 11/23/2011

Back to GENE, click on SNP geneview, to search for MTHFR SNPs

The screenshot displays the NCBI Gene browser interface for the MTHFR gene (Gene ID: 4524). The browser window title is "MTHFR methylenetetrahydrofolate reductase (NAD(P)H) [Homo sapiens] - Gene - NCBI - Mozilla Firefox". The address bar shows the URL "http://www.ncbi.nlm.nih.gov/gene/4524".

The main content area shows the genomic sequence for NC_000001 (chromosome 1) with a scale from 870 K to -11,845 K. The MTHFR gene structure is highlighted, showing exons as green boxes and introns as lines with arrows indicating the direction of transcription. The gene is located on the negative strand. The MTHFR gene structure is shown with exons as green boxes and introns as lines with arrows indicating the direction of transcription. The gene is located on the negative strand.

Below the gene structure, there are several tracks showing variants:

- SNP:** A track showing a specific SNP, 11845787..11866160, which is currently loading.
- LSDB or Clinically Associated Variants:** A track showing variants associated with Leiden Steroid Defect Database (LSDB) or clinical significance. Variants are represented by colored boxes (purple, green, blue).
- Cited Variants:** A track showing variants that have been cited in the literature. Variants are represented by colored boxes (light blue, green, orange).
- NHGRI GWAS Catalog:** A track showing variants from the National Human Genome Research Institute (NHGRI) Genome-Wide Association Study (GWAS) Catalog. Variants are represented by colored boxes (red, orange).

On the right side of the browser, there is a sidebar with links to various resources:

- Probe
- Protein
- PubChem Compound
- PubChem Substance
- PubMed
- PubMed (GeneRIF)
- PubMed (OMIM)
- RefSeq Proteins
- RefSeq RNAs
- RefSeqGene
- SNP
- SNP: GeneView
- SNP: Genotype
- SNP: VarView
- Taxonomy
- UniGene
- UniSTS

At the bottom of the sidebar, there is a section titled "Links to other resources" with a link to AceView.

dbSNP for MTHFR gene: all coding SNPs are shown (default)

SNP linked to Gene MTHFR(geneID:4524) Via Contig Annotation - Mozilla Firefox

File Edit View History Bookmarks Tools Help

SNP linked to Gene MTHFR(geneID:4524)...

http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?locusId=4524

NCBI

dbSNP
Short Genetic Variations

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search for SNP on NCBI Reference Assembly

Search Entrez SNP for Go

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

Go

SNP linked to Gene MTHFR(geneID:4524) Via Contig Annotation

Send rs# on all gene models to Batch Query Download all rs# to file. Genotype VarView

Gene Model (mRNA alignment) information from genome sequence

Total gene model (contig mRNA transcript):				1		
mRNA	transcript	protein	mRNA orientation	Contig	Contig Label	List SNP
NM_005957.4	plus strand	NP_005948.3	forward	NT_021937.19	GRCh37.p5	<- currently shown

☐ Clinical Source ☐ in gene region ☒ cSNP ☐ has frequency ☐ double hit refresh

gene model (contig mRNA transcript):	Contig Label	Contig	mRNA	protein	mRNA orientation	transcript	snp count
	GRCh37.p5	NT_021937.19	NM_005957.4	NP_005948.3	forward	plus strand	81, coding

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HUMAN VARIATION

11:05 PM 11/23/2011

Scroll down...

SNP linked to Gene MTHFR(geneID:4524) Via Contig Annotation - Mozilla Firefox

File Edit View History Bookmarks Tools Help

SNP linked to Gene MTHFR(geneID:4524) +

http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?locusId=4524

Google

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DOCUMENTATION
SEARCH
RELATED SITES

Region	Chr. position	mRNA pos	dbSNP rs# cluster id	Hetero- zygosity	Validation	MAF	Allele origin	3D	Linkout	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos	PubM
	11850749	2188	rs45572531	0.006		0.0056				synonymous	A	Thr [T]	3	653	
										contig reference	G	Thr [T]	3	653	
	11850750	2187	rs35737219	0.035		0.0122				missense	T	Met [M]	2	653	
										contig reference	C	Thr [T]	2	653	
	11850759	2178	rs145544233	0.000						missense	T	Val [V]	2	650	
										contig reference	C	Ala [A]	2	650	
	11850767	2170	rs139586522	0.001						synonymous	A	Thr [T]	3	647	
										contig reference	C	Thr [T]	3	647	
	11850805	2132	rs190087897	N.D.						missense	T	Leu [L]	1	635	
										contig reference	G	Val [V]	1	635	
	11850842	2095	rs140496402	0.001						synonymous	A	Val [V]	3	622	
										contig reference	G	Val [V]	3	622	
	11850927	2010	rs2274976	0.099		0.0509				missense	A	Gln [Q]	2	594	
										missense	C	Pro [P]	2	594	

EN ?

11:06 PM
11/23/2011

From bibliography, we are interested for a SNP with a nucleotide change C677T resulting in aminoacid change Ala222Val. This SNP has been implicated with many disorders and we are interested in identifying its unique rs code. Tick include clinically sources and then click refresh

SNP linked to Gene MTHFR(geneID:4524) Via Contig Annotation - Mozilla Firefox

File Edit View History Bookmarks Tools Help

SNP linked to Gene MTHFR(geneID:4524)...

http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?locusId=4524

Short Genetic Variations

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search for SNP on NCBI Reference Assembly

Search Entrez SNP for Go

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

Go

SNP linked to Gene MTHFR(geneID:4524) Via Contig Annotation

Send rs# on all gene models to Batch Query Download all rs# to file Genotype VarView

Gene Model (mRNA alignment) information from genome sequence

Total gene model (contig mRNA transcript):				1		
mRNA	transcript	protein	mRNA orientation	Contig	Contig Label	List SNP
NM_005957.4	plus strand	NP_005948.3	forward	NT_021937.19	GRCh37.p5	<- currently shown

☒ Clinical Source ☐ in gene region ☒ cSNP ☐ has frequency ☐ double hit refresh

gene model (contig mRNA transcript):	Contig Label	Contig	mRNA	protein	mRNA orientation	transcript	snp count
GRCh37.p5	NT_021937.19	NM_005957.4	NP_005948.3		forward	plus strand	81, coding

Region	Chr. position	mRNA pos	dbSNP rs# cluster id	Heterozygosity	Validation	MAF	Allele origin	3D	Linkout	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos	PubMed

EN 11:09 PM 11/23/2011

In exon 5, it is rs1801133. Click...

SNP linked to Gene MTHFR(geneID:4524) Via Contig Annotation - Mozilla Firefox

File Edit View History Bookmarks Tools Help

SNP linked to Gene MTHFR(geneID:4524... +

http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?showRare=on&chooseRs=coding&locusId=4524&mrna=N

11856328	944	rs190090719	N.D.			Yes		missense	C	Leu [L]	1	2
								contig reference	T	Phe [F]	1	2
11856330	942	rs138760604	0.000			Yes		missense	A	His [H]	2	2
								contig reference	G	Arg [R]	2	2
11856335	937	rs34279942	0.004			Yes		synonymous	T	Phe [F]	3	2
								contig reference	C	Phe [F]	3	2
11856343	929	rs45589033	0.004			Yes		missense	A	Asn [N]	1	2
								contig reference	G	Asp [D]	1	2
11856376	896	rs150847674	0.001			Yes		missense	A	Asn [N]	1	2
								contig reference	G	Asp [D]	1	2
11856378	894	rs1801133	0.387			Yes		missense	T	Val [V]	2	2
								contig reference	C	Ala [A]	2	2
11856382	890	rs45438591	0.004			Yes		missense	A	Arg [R]	1	2
								contig reference	G	Gly [G]	1	2
11856383	889	rs141060174	0.000			Yes		synonymous	A	Ala [A]	3	2
								contig	G	Ala [A]	3	2

EN 11:12 PM 11/23/2011

dbSNP for rs1801133

Reference SNP(refSNP) Cluster Report: rs1801133 **Clinical Source** - Mozilla Firefox

File Edit View History Bookmarks Tools Help

Reference SNP(refSNP) Cluster Report: rs... +

http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?rs=1801133

NCBI dbSNP Short Genetic Variations

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search for SNP on NCBI Reference Assembly

Search Entrez SNP for Go

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SNP SUBMISSION

DOCUMENTATION

Reference SNP(refSNP) Cluster Report: rs1801133 **Clinical Source**

RefSNP	Allele	HGVS Names
Organism: human (Homo sapiens)	Variation Class : SNV: single nucleotide variation	NC_000001.10:g.11856378C>T
Molecule Type: Genomic	RefSNP Alleles: C/T	NG_013351.1:g.14783C>T
Created/Updated in build: 89/135	Allele Origin:	NM_005957.4:c.665C>T
Map to Genome Build: 37.3	Ancestral Allele: C	NP_005948.3:p.Ala222Val
Validation Status :	Clinical Source: VarView OMIM	
Citation: PubMed	Clinical Significance: NA	
Association: NHGRI GWAS PheGenI	MAF/MinorAlleleCount : A=0.325/711	
	MAF Source: 1000 Genomes	

SNP Details are organized in the following sections:

[GeneView](#) [Map](#) [Submission](#) [Fasta](#) [Resource](#) [Diversity](#) [Validation](#)

[Integrated Maps](#) (Hint: click on 'Chr Pos' or 'Contig Pos' column value to see variation in NCBI sequence viewer)

Integrated Maps

http://main.genome-browser.bx.psu.edu/cgi...&db=hg18&position=chr1 11778944 11778985

EN ? 11:13 PM 11/23/2011

dbSNP for rs1801133

Reference SNP(refSNP) Cluster Report: rs1801133 **Clinical Source** - Mozilla Firefox

File Edit View History Bookmarks Tools Help

Reference SNP(refSNP) Cluster Report: rs... +

http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?rs=1801133

Fasta sequence (Legend)

>gnl|dbSNP|rs1801133|allelePos=401|totalLen=801|taxid=9606|snpclass=1|alleles='C/T'|mol=Genomic|build=135

```
TCTTTTCTAT GGCCACCAAG TGCAGGCCTG ATTTGCTTGG CTGCTCAAGG CAGGACAGTG
TGGGAGTTTG GAGCAATCCA CCCCCACTCT TGGAAGTGGG CTCTGAGCCA CCTCCCCTGA
GAGTCATCTC TGGGGTCAGA AGCATATCAG TCATGAGCCC AGCCACTCAC TGTTTTAGTT
CAGGCTGTGC TGTGCTGTTG GAAGGTGCAA GATCAGAGCC CCCAAAGCAG AGGACTCTCT
CTGCCAGTC CCTGTGGTCT CTTTCATCCCT CGCCTTGAAC AGGTGGAGGC CAGCCTCTCC
TGACTGTCTC CCCTATTGGC AGGTTACCCC AAAGGCCACC CCGAAGCAGG GAGCTTTGAG
GCTGACCTGA AGCACTTGAA GGAGAAGGTG TCTGCGGGAG
Y
CGATTTCATC ATCAGCGAGC TTTTCTTTGA GGCTGACACA TTCTTCCGCT TTGTGAAGGC
ATGCACCGAC ATGGGCATCA CTTGCCCCAT CGTCCCCGGG ATCTTTCCCA TCCAGGTGAG
GGGCCAGGA GAGCCCATAA GCTCCCTCCA CCCCCTCTC ACCGCACCGT CCTCGCACAG
GCTGGGGGCT CTGGGTGGAG TGCTGAGTTC GCTGAGTTCT TCCCAGATCT CCTCTCAGGT
CCAGAACTTG CACAGCGTTG CTTGGCCACC CCATTTTGGT TACCTCTAAT TTTCCCCCA
AAACCCAGCA ACAGTGTCTG TTGAGGGGTT TGTTGTACTT TGGCCAACAA GCATCACCAG
AAGGGATTCT AATTCTCATT ACAAATCCTG CTTAAATCAG
```

NCBI Resource Links

Resource

Submitter-Referenced	dbSNP Blast Analysis	UniGene Cluster ID	3D structure mapping	OMIM
dbSTS GenBank		214142	NP_005948	607093.0003
80 G67549 NT_021937.17				

Population Diversity

http://main.genome-browser.bx.psu.edu/cgi...&db=hg18&position=chr1 11778944 11778985

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dbSNP for rs1801133

Reference SNP(refSNP) Cluster Report: rs1801133 **Clinical Source** - Mozilla Firefox

File Edit View History Bookmarks Tools Help

Reference SNP(refSNP) Cluster Report: rs... +

http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?rs=1801133

Population Diversity

ss#	Sample Ascertainment				Genotype Detail <small>NEW</small>						Alleles	
	Population	Individual Group	Chrom. Sample Cnt.	Source	C	C/C	C/T	T	T/T	HWP	C	T
ss105110012	PA155830959		2308	AF							0.684	0.316
ss161162859	ENSEMBL_celera		6	IG		0.667			0.333		0.667	0.333
ss166100327	PGP		2	IG			1.000				0.500	0.500
ss198024849	BUSHMAN_POP		2	IG			1.000				0.500	0.500
ss218238438	pilot_1_YRI_low_coverage_panel		118	AF							0.881	0.119
ss230427635	pilot_1_CEU_low_coverage_panel		120	AF							0.725	0.275
ss238142821	pilot_1_CHB+JPT_low_coverage_panel		120	AF							0.567	0.433
ss23844687	AFD_EUR_PANEL	European	48	IG		0.458	0.500		0.042	0.317	0.708	0.292
	AFD_AFR_PANEL	African American	46	IG		0.739	0.261			0.752	0.870	0.130
	AFD_CHN_PANEL	Asian	48	IG		0.500	0.333		0.167	0.251	0.667	0.333
ss2978066	NIHPDR	Global	74	IG		0.649	0.270		0.081	0.251	0.784	0.216
ss3210929	JBIC-allele		1484	AF							0.597	0.403
ss341933225	ESP_Cohort_Populations		4552	GF		0.558	0.364		0.078	0.020	0.740	0.260

http://main.genome-browser.bx.psu.edu/cgi...&db=hg18&position=chr1 11778944 11778985

EN ?

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We are now interested to see the haplotype structure of the gene. Is our SNP in LD with other SNPs? Type www.hapmap.org

HapMap Homepage - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Homepage

http://hapmap.ncbi.nlm.nih.gov/

International HapMap Project

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
About the Project HapMap Publications HapMap Tutorial HapMap Mailing List HapMap Project Participants	<ul style="list-style-type: none">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

Project Data

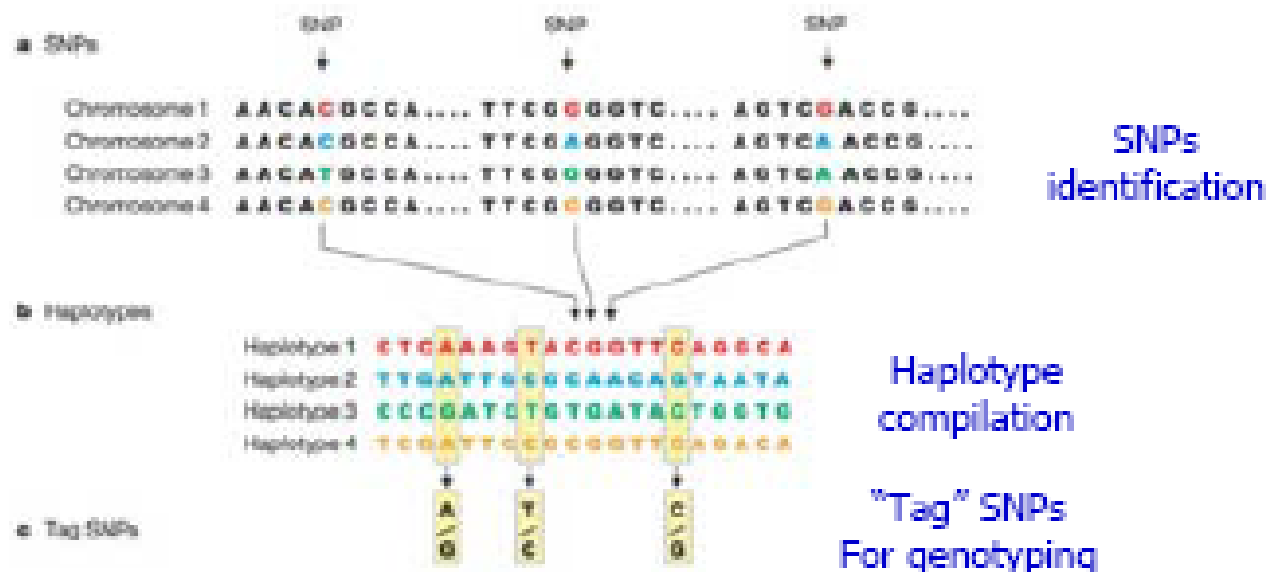
HapMap Genome Browser release #28 (Phases 1, 2 & 3 - merged genotypes & frequencies)

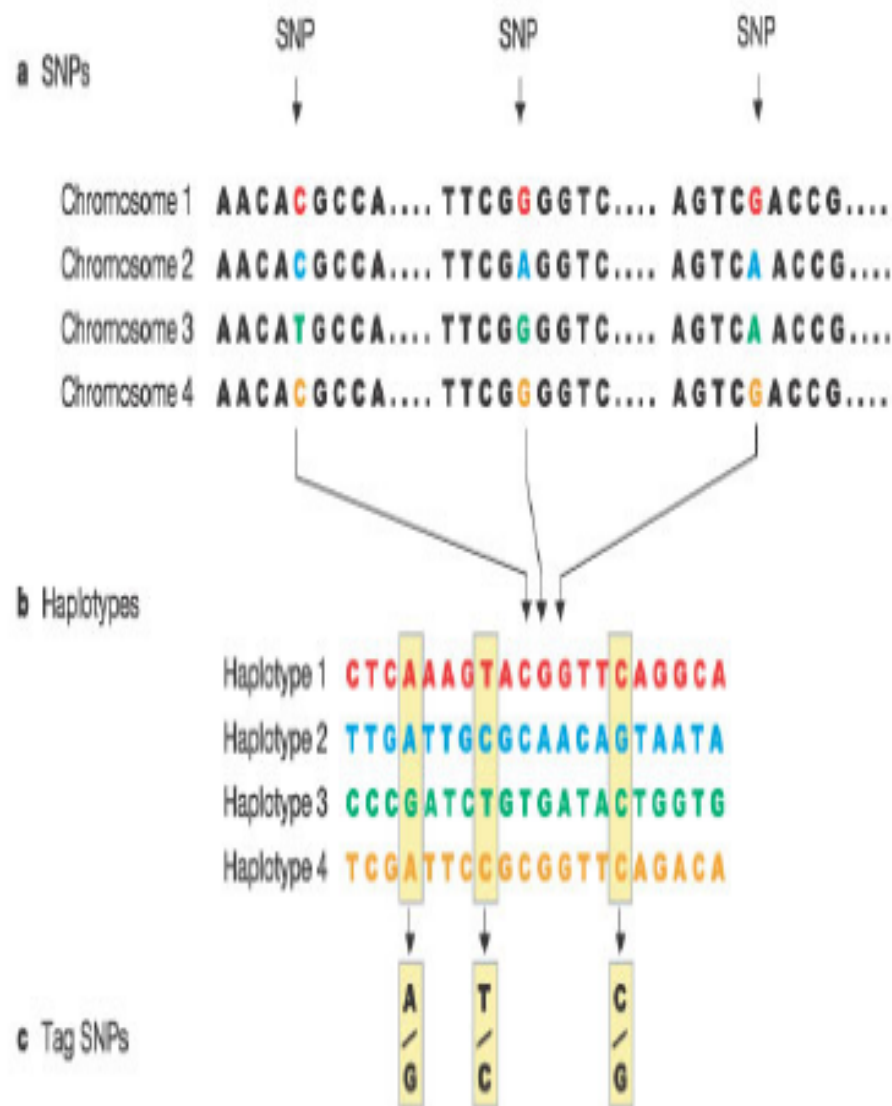
EN ?

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International HapMap Project

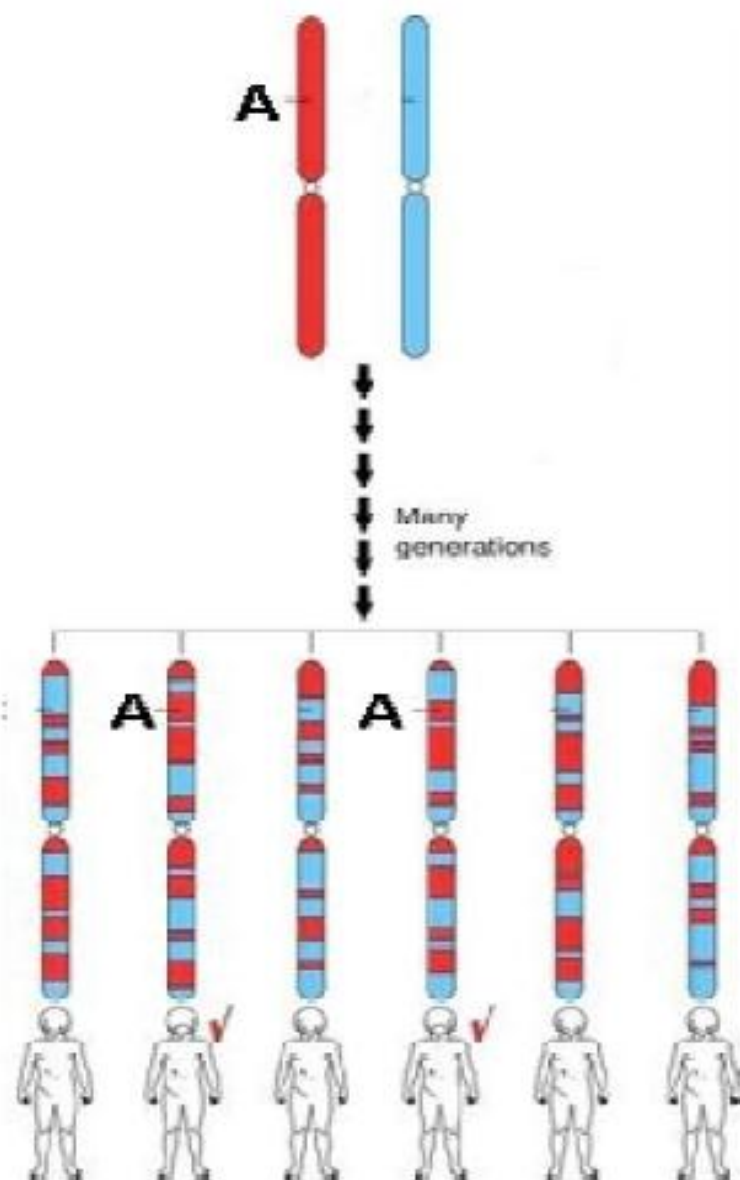
- A catalog of common genetic variants that occur in human beings.
 - what these variants are
 - where they occur in our DNA
 - how they are distributed among people within populations and among populations in different parts of the world





Εικόνα 2: Η κατασκευή του HapMap γίνεται σε 3 στάδια. (a)

Προσδιορίζονται SNPs σε δείγματα DNA από πολλά άτομα. (b) Γειτονικοί SNPs που κληρονομούνται μαζί κατατάσσονται σε απλοτύπους (c) Προσδιορίζονται "Tag" SNPs μέσα στους απλοτύπους τα οποία περιγράφουν πλήρως τους απλοτύπους. Με τη γονοτύπωση 3 tag SNPs που φαίνονται στην εικόνα, οι ερευνητές προσδιορίζουν ποιός από τους 4 απλοτύπους είναι παρόν σε κάθε άτομο..



Το διάγραμμα δείχνει 2 προγονικά χρωμοσώματα που αναμιγνύονται μέσω ανασυνδυασμού για πολλές γενιές ώστε να δώσουν διαφορετικά χρωμοσώματα απογόνων. Εάν μια γενετική παραλλαγή που σημειώνεται με το γράμμα A στο προγονικό χρωμόσωμα αυξάνει τον κίνδυνο για κάποιο νόσημα, οι 2 απόγονοι της σύγχρονης γενιάς που κληρονομούν το συγκεκριμένο τμήμα του χρωμοσώματος, θα έχουν αυξημένο κίνδυνο. Πλησίον της παραλλαγής A, υπάρχουν πολλοί SNPs που μπορούν να χρησιμοποιηθούν για τον εντοπισμό της A.

Click HapMap Genome Browser (#24 - full data set)

The screenshot shows the HapMap Genome Browser homepage in a Mozilla Firefox browser window. The address bar displays <http://hapmap.ncbi.nlm.nih.gov/>. The page layout includes a left sidebar with navigation links and a main content area with announcements.

Left Sidebar:

- 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

Main Content Area:

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](#).

The browser's status bar at the bottom shows the system clock as 11:23 PM on 11/23/2011.

Search for MTHFR

HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126 - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Data Rel 24/phaseII Nov08, on ...

http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/

Google

International HapMap Project

Home | About the Project | Data | Publications | Tutorial

Instructions

Searching: Search using a sequence name, gene name, locus, or other landmark. The wildcard character * is allowed.

Navigation: Click one of the rulers to center on a location, or click and drag to select a region. Use the Scroll/Zoom buttons to change magnification and position.

Examples : Chr20, Chr9:660,000..760,000, SNP:rs6870660, NM_153254, BRCA2, 5q31, ENm010, gwa*, PARK3.

[Help] [Reset]

Search

Help links:

- LD - - tagSNPs - - Phased Haplotype - - Genotype data - - Frequency data - - Symbols and colours used -

Landmark or Region : MTHFR Search

Reports & Analysis : Annotate LD Plot Configure... Go

Data Source HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126

Population descriptors: YRI: Yoruba in Ibadan, Nigeria, JPT: Japanese in Tokyo, Japan, CHB: Han Chinese in Beijing, China, CEU: CEPH (Utah residents with ancestry from northern and western Europe)

EN ? 11:26 PM 11/23/2011

HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126: chr1:11768374..11788702 - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Data Rel 24/phaseII Nov08, on ... +

http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/#search

Google

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Showing 20.33 kbp from chr1, positions 11,768,374 to 11,788,702

Instructions

Searching: Search using a sequence name, gene name, locus, or other landmark. The wildcard character * is allowed.

Navigation: Click one of the rulers to center on a location, or click and drag to select a region. Use the Scroll/Zoom buttons to change magnification and position.

Examples: Chr20, Chr9:660,000..760,000, SNP:rs6870660, NM_153254, BRCA2, 5q31, ENM010, gwa*, PARK3.

[Bookmark this] [Upload your own data] [Hide banner] [Share these tracks] [Link to Image] [SNP genotype data] [High-res Image] [Help] [Reset]

Search

Help links:

- LD - - tagSNPs - - Phased Haplotype - - Genotype data - - Frequency data - - Symbols and colours used -

Landmark or Region: MTHFR Search

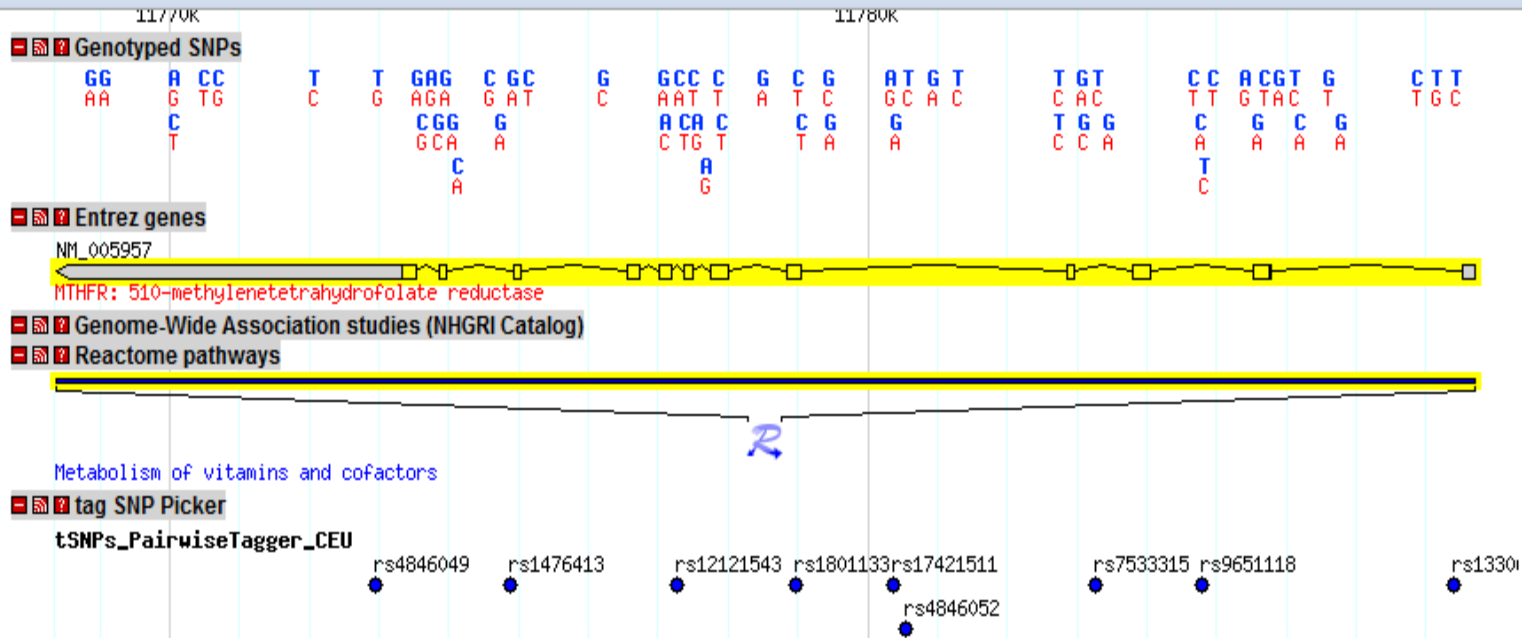
Data Source: HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126

Reports & Analysis: Annotate LD Plot Configure... Go

Scroll/Zoom: << < - Show 20.33 kbp + > >> Flip

Population descriptors: YRI: Yoruba in Ibadan, Nigeria JPT: Japanese in Tokyo, Japan CHB: Han Chinese in Beijing, China CEU: CEPH (Utah residents with ancestry

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Clear highlighting

Update Image

For performing in depth LD and Haplotype analysis of genotype data, install [Haploview](#) in your local machine. Haploview (ver 4.1) is currently available for download. This version does not handle hapmap3 samples. Please check the [Haploview website](#) for updates.

Tracks

Overview ☐ All on ☐ All off



Click on Download HapMap data to find LD scores for all SNPs, including rs1801133

HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126: chr1:11768374..11788702 - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Data Rel 24/phaseII Nov08, on ...

http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/

Project

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Showing 20.33 kbp from chr1, positions 11,768,374 to 11,788,702

Instructions

Searching: Search using a sequence name, gene name, locus, or other landmark. The wildc...

Navigation: Click one of the rulers to center on a location, or click and drag to select a region...

Examples : Chr20, Chr9:660,000..760,000, SNP:rs6870660, NM_153254, BRCA2, 5q31, EN...

[Bookmark this] [Upload your own data] [Hide banner] [Share these tracks] [Link to Im...

Search

Help links:

- LD - - tagSNPs - - Phased Haplotype - - Genotype data -

Landmark or Region : MTHFR Search

Data Source HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126

Population descriptors:YRI: Yoruba in Ibadan, Nigeria, JPT: Japanese in Tokyo, Japan, CHB: Han Chinese in Beijing, China, CEU: CEPH (Utah residents with ancestry from northern and western Europe)

Annotate LD Plot
Annotate Phased Haplotype Display
Annotate tag SNP Picker
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Download HapMap LD Data
Download Impute Data
Download Phased Haplotype Data
Download SNP Allele Frequency Data
Download SNP Genotype Frequency Data
Download SNP genotype data
Download tag SNP Data
Highlight SNP Properties
Annotate tag SNP Picker

Scroll/Zoom: <<< < < Show 20.33 kbp > >> >>> Flip

EN 11:35 PM 11/23/2011

results

Mozilla Firefox

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http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/#search

http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/#search

#Wed Nov 23 16:36:22 2011: HapMap LD data dump, 60 SNPs genotyped in population CEU on chr1:11768374..11788702

#pos1	pos2	pop	marker1	marker2	D'	r ²	LOD
11768839	11770023	CEU	rs4846048	rs2184226	1	0.2	4.76
11768839	11770036	CEU	rs4846048	rs3737967	0.015	0	0
11768839	11770448	CEU	rs4846048	rs1537516	1	0.056	1.53
11768839	11770655	CEU	rs4846048	rs1537514	1	0.063	1.61
11768839	11772952	CEU	rs4846048	rs4846049	0.573	0.283	5.72
11768839	11773514	CEU	rs4846048	rs2274976	1	0.032	0.4
11768839	11773590	CEU	rs4846048	rs3818762	0.538	0.247	4.85
11768839	11773906	CEU	rs4846048	rs2274974	1	0.004	0.18
11768839	11774697	CEU	rs4846048	rs13306556	0.641	0.026	0.52
11768839	11774887	CEU	rs4846048	rs1476413	0.453	0.182	3.5
11768839	11775103	CEU	rs4846048	rs17375901	1	0.027	1.11
11768839	11777063	CEU	rs4846048	rs1801131	0.52	0.241	4.82
11768839	11777258	CEU	rs4846048	rs12121543	0.478	0.166	3.05
11768839	11777342	CEU	rs4846048	rs1994798	0.881	0.456	8.6
11768839	11777483	CEU	rs4846048	rs2066462	1	0.067	1.62
11768839	11778454	CEU	rs4846048	rs6541003	0.95	0.53	13.2
11768839	11778965	CEU	rs4846048	rs1801133	1	0.16	4.16
11768839	11779434	CEU	rs4846048	rs17421462	1	0.2	4.16
11768839	11780298	CEU	rs4846048	rs1572151	1	0.2	4.76
11768839	11780375	CEU	rs4846048	rs17421511	1	0.524	12.86
11768839	11780538	CEU	rs4846048	rs4846052	0.95	0.53	13.2
11768839	11780911	CEU	rs4846048	rs17421560	0.641	0.026	0.52
11768839	11782707	CEU	rs4846048	rs11121832	0.866	0.663	10.66
11768839	11783045	CEU	rs4846048	rs2066471	1	0.476	10.77
11768839	11783052	CEU	rs4846048	rs13306567	0.208	0.001	0.02
11768839	11783270	CEU	rs4846048	rs7533315	1	0.891	24.23

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11/23/2011

LD results for rs1801133

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http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/#search

http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/#search

Google

11775103	11786736	CEU	rs17375901	rs7553194	1	0.432	6.36
11775103	11788129	CEU	rs17375901	rs3753582	1	0.434	7.09
11775103	11788391	CEU	rs17375901	rs13306561	1	0.239	3.88
11777063	11777258	CEU	rs1801131	rs12121543	1	0.671	18.76
11777063	11777342	CEU	rs1801131	rs1994798	0.947	0.619	13.31
11777063	11777483	CEU	rs1801131	rs2066462	1	0.226	6.23
11777063	11778454	CEU	rs1801131	rs6541003	1	0.675	19.42
11777063	11778965	CEU	rs1801131	rs1801133	1	0.189	5.61
11777063	11779434	CEU	rs1801131	rs17421462	1	0.048	1.25
11777063	11780298	CEU	rs1801131	rs1572151	1	0.048	1.25
11777063	11780375	CEU	rs1801131	rs17421511	0.865	0.363	7.96
11777063	11780538	CEU	rs1801131	rs4846052	1	0.675	19.42
11777063	11780911	CEU	rs1801131	rs17421560	1	0.222	6.39
11777063	11782707	CEU	rs1801131	rs11121832	0.498	0.147	2.07
11777063	11783045	CEU	rs1801131	rs2066471	0.838	0.306	6.24
11777063	11783052	CEU	rs1801131	rs13306567	1	0.072	1.64
11777063	11783270	CEU	rs1801131	rs7533315	0.517	0.204	4.03
11777063	11783430	CEU	rs1801131	rs17037390	1	0.345	9
11777063	11784634	CEU	rs1801131	rs17037396	1	0.222	6.39
11777063	11784750	CEU	rs1801131	rs17037397	1	0.096	2.77
11777063	11784801	CEU	rs1801131	rs9651118	1	0.198	6.8
11777063	11784919	CEU	rs1801131	rs7525338	1	0.016	0.45
11777063	11785365	CEU	rs1801131	rs17367504	1	0.345	9.6
11777063	11785644	CEU	rs1801131	rs2066470	1	0.204	5.54
11777063	11786736	CEU	rs1801131	rs7553194	0.872	0.171	4.19
11777063	11788129	CEU	rs1801131	rs3753582	0.888	0.191	4.97
11777063	11788391	CEU	rs1801131	rs13306561	0.753	0.252	4.36
11777258	11777342	CEU	rs12121543	rs1994798	1	0.434	9.93
11777258	11777483	CEU	rs12121543	rs2066462	1	0.244	8.27

EN ?

11:37 PM
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Back to MTHFR page, click Annotate LD plot

HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126: chr1:11768374..11788702 - Mozilla Firefox

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HapMap Data Rel 24/phaseII Nov08, on ...

http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/

Project

Home | About the Project | Data | Publications | Tutorial

Showing 20.33 kbp from chr1, positions 11,768,374 to 11,788,702

Instructions
Searching: Search using a sequence name, gene name, locus, or other landmark. The wildc
Navigation: Click one of the rulers to center on a location, or click and drag to select a region

Examples : Chr20, Chr9:660,000..760,000, SNP:rs6870660, NM_153254, BRCA2, 5q31, EN

[Bookmark this] [Upload your own data] [Hide banner] [Share these tracks] [Link to Im

Search
Help links:
- LD - - tagSNPs - - Phased Haplotype - - Genotype data -

Landmark or Region :
MTHFR Search

Data Source
HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126

Population descriptors: YRI: Yoruba in Ibadan, Nigeria, JPT: Japanese in Tokyo, Japan, CHB: Han Chinese in Beijing, China, CEU: CEPH (Utah residents with ancestry from northern and western Europe)

Annotate LD Plot
Annotate Phased Haplotype Display
Annotate tag SNP Picker
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Download HapMap GFF File
Download HapMap LD Data
Download Impute Data
Download Phased Haplotype Data
Download SNP Allele Frequency Data
Download SNP Genotype Frequency Data
Download SNP genotype data
Download tag SNP Data
Highlight SNP Properties
Download HapMap LD Data

Scroll/Zoom: <<< < < Show 20.33 kbp > >> >>> Flip

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Click configure

HapMap Data Rel 24/phaseII Nov08, on NCBI B36 assembly, dbSNP b126: chr1:11768374..11788702 - Mozilla Firefox

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HapMap Data Rel 24/phaseII Nov08, on ... +

http://hapmap.ncbi.nlm.nih.gov/cgi-perl/gbrowse/hapmap24_B36/#search

Google

Configure... LD Plot

Cancel Configure

Max. Segment Size 250Kb Max. # gt'd SNPs 200 Box Size Proportionate

LD Properties: lod greater than -1 and less than 250

Color: Pairwise plot red

Populations: CEU ☐ off ☒ on CHB ☒ off ☐ on JPT ☒ off ☐ on YRI ☒ off ☐ on

Orientation: normal invert normal invert

Cancel Configure

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

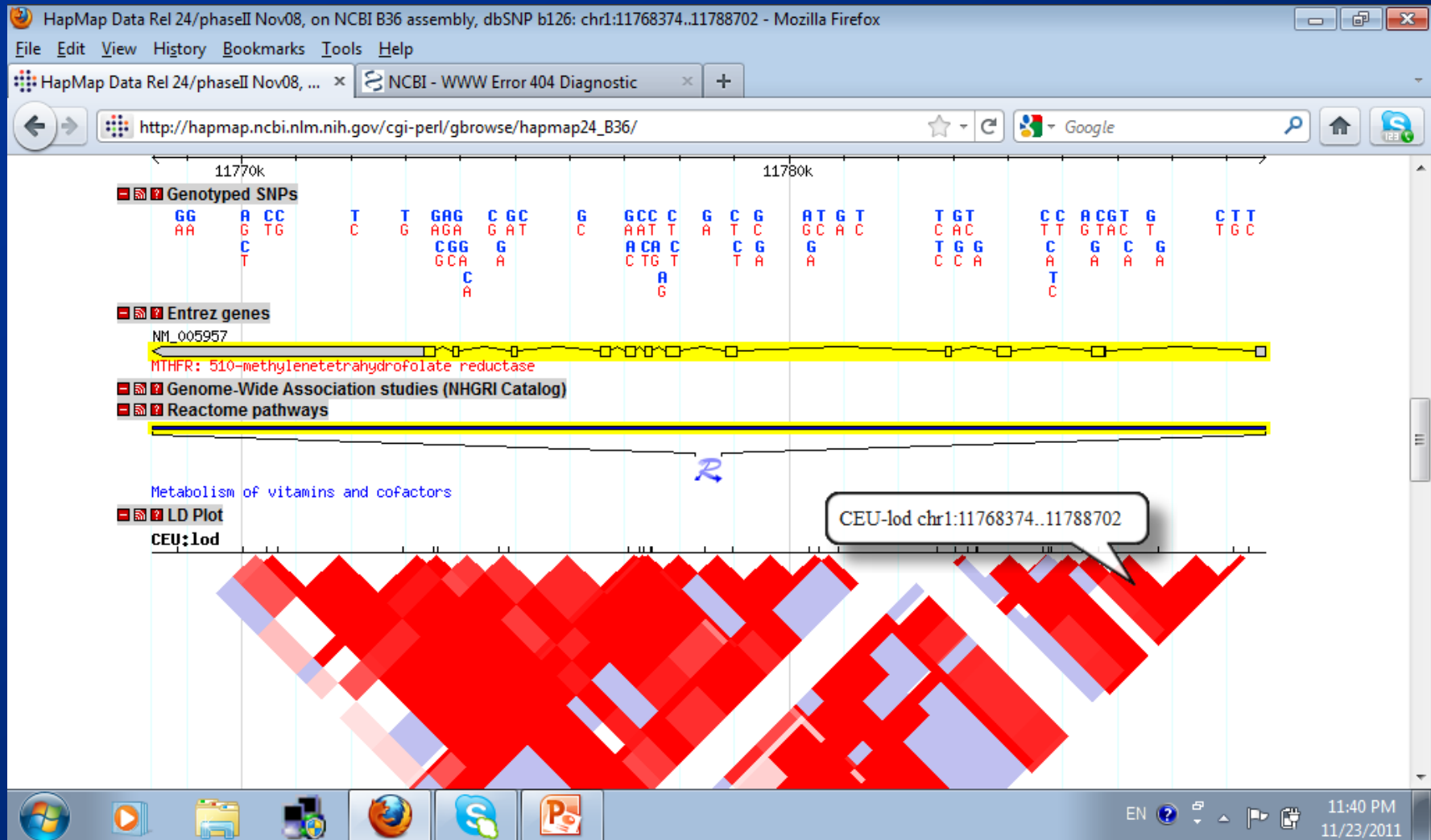
Please send questions and comments on website to hapmap-help@ncbi.nlm.nih.gov

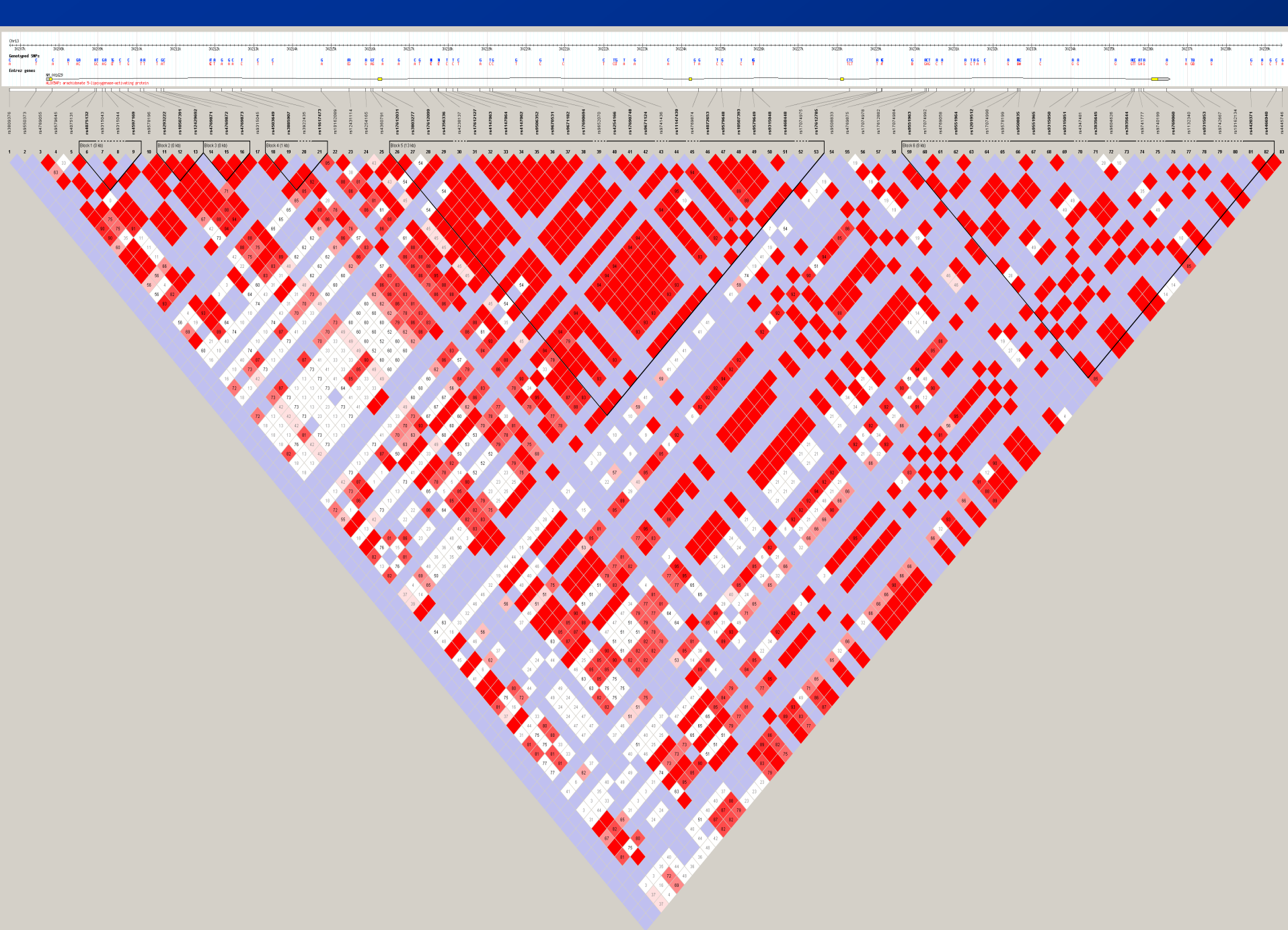
Note: This page uses cookies to save and restore preference information. No information is shared.

Generic genome browser version 0.1.1

EN 11:39 PM 11/23/2011

Red color shows regions of High LD





Search for the published literature of MTHFR gene. Go to HuGE Navigator (<http://www.hugenavigator.net/>), and click on HuGE literature finder

The screenshot shows a Mozilla Firefox browser window with the title "HuGENavigator - Mozilla Firefox". The address bar displays "http://www.hugenavigator.net/HuGENavigator/home.do". The page content includes the HuGE Navigator logo (a blue circle with "HN" in yellow) and the text "HuGE Navigator (version 2.0) An integrated, searchable knowledge base of genetic associations and human genome epidemiology." Below this is a navigation bar with links: Home, Download Center, Open Source Projects, and Contact. A section titled "Curator's recommended HuGE article for this week:" features a summary of a meta-analysis on FTO variants and obesity risk, with a "PubMed Link" provided. To the right, there is a description of HuGE Navigator as a continuously updated knowledge base, social media links for Facebook and Twitter, and the "Last database update: 23 Nov 2011". A site citation is listed at the bottom: "Site citation: W Yu, M Gwinn, M Clyne, A Yesupriya & M J Khoury. A Navigator for Human Genome Epidemiology. Nat Genet 2008 Feb;40(2): 124-5." The page also features several toolboxes with icons and descriptions: "Phenopedia" (genetic associations by disease), "Genopedia" (genetic associations by gene), "HuGE Literature Finder" (published articles), "Gene Prospector" (evaluating genes), "GWAS Integrator" (GWAS and relevant), and "Cancer GAMAdb" (cancer genetic associations). The Windows taskbar at the bottom shows various application icons and the system clock indicating 11:42 PM on 11/23/2011.

HuGENavigator - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Data Rel 24/phaseII Nov08, ... x HuGENavigator x +

http://www.hugenavigator.net/HuGENavigator/home.do

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Εγγραφείτε ή συνδεθείτε

6 Σχόλια »

97 Μέλη »

HuGE Navigator (version 2.0)
An integrated, searchable knowledge base of genetic associations and human genome epidemiology.

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Curator's recommended HuGE article for this week:

Physical Activity Attenuates the Influence of FTO Variants on Obesity Risk: A Meta-Analysis of 218,166 Adults and 19,268 Children. Kilpeläinen TO, Qi L, Brage S, Sharp SJ, Sonestedt E, et al. PLoS Med. 2011 Nov;8(11):e1001116.
[PubMed Link](#)

HuGE Navigator is a continuously updated knowledge base in human genome epidemiology, including population prevalence of genetic variants, genetic associations... [more](#)

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Last database update: **23 Nov 2011**

Site citation: W Yu, M Gwinn, M Clyne, A Yesupriya & M J Khoury. A Navigator for Human Genome Epidemiology. Nat Genet 2008 Feb;40(2): 124-5.

Phenopedia: Look up genetic associations and human genome epidemiology summaries by disease. ?

Genopedia: Look up genetic associations and human genome epidemiology summaries by gene. ?

HuGE Literature Finder: Find published articles in genetic associations and human genome epidemiology. ?

Gene Prospector: A gateway for evaluating genes in relation to disease and risk factors. ?

GWAS Integrator: Explore published GWAS and relevant ?

Cancer GAMAdb: Database of cancer genetic associations ?

EN ?

11:42 PM
11/23/2011

Search literature for MTHFR

HuGENavigator|HuGE Literature Finder|Search - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Data Rel 24/phaseII Nov08, ... x HuGENavigator|HuGE Literature Find... x +

http://www.hugenavigator.net/HuGENavigator/startPagePubLit.do

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HuGE Navigator (version 2.0)
An integrated, searchable knowledge base of genetic associations and human genome epidemiology.

HuGE Navigator > HuGE Literature Finder Last data upload: 23 Nov 2011. (Total 66054 articles)

HuGE Literature Finder

Data collected since 2001 [Home](#) | [About](#) | [Search Instructions](#) | [FAQs](#)

Search Literature ▾ for

Queries for the summary

- [Since last upload](#)
- [All publications](#)
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- [Meta-analysis](#)
- [HuGE review](#)

- Enter search terms into the text box.
- Search terms can include disease, exposure, gene, author, journal, etc.
- Simple Boolean operators are allowed, such as AND or OR.
- Use the Search dropdown list to switch to other HuGE Navigator applications.

HuGE Literature Finder is a search engine for finding published literature on genetic associations and other human genome epidemiology. The search query can include disease/outcome, environmental factors, genes

EN ? 11:43 PM 11/23/2011

2429 results found!!!

The screenshot shows a Mozilla Firefox browser window displaying the HuGE Navigator search results. The address bar shows the URL: <http://www.hugenavigator.net/HuGENavigator/searchSummary.do?firstQuery=MTHFR&publitSearchType>. The page header includes the HuGE Navigator logo and version 2.0, along with the description: "An integrated, searchable knowledge base of genetic associations and human genome epidemiology." The search criteria are displayed as "MTHFR[Text+MeSH]". The results show 2429 articles, with the first article listed as "Common variants of homocysteine metabolism pathway genes and risk of type 2 diabetes and related traits in indians." The page also includes navigation links like Home, About, Search Instructions, and FAQs, and a filter section for Disease, Gene, StudyType, Year, Author, Journal, and Country.

HuGENavigator|HuGE Literature Finder|Search - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Data Rel 24/phaseII Nov08, ... x HuGENavigator|HuGE Literature Find... x +

http://www.hugenavigator.net/HuGENavigator/searchSummary.do?firstQuery=MTHFR&publitSearchType ☆ ↻ Google

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HuGE Navigator > HuGE Literature Finder Last data upload: 23 Nov 2011. (Total 66054 articles)

HuGE Literature Finder

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Search Literature ▼ for MTHFR Go Clear

Search Criteria: MTHFR[Text+MeSH] [\[Query Detail\]](#) Download

Filtered By ? Disease ? Gene ? StudyType ? Year ? Author ? Journal ? Country

Articles 1 - 25 of 2429

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1. [Common variants of homocysteine metabolism pathway genes and risk of type 2 diabetes and related traits in indians.](#) [\[Detail\]](#)

Experimental diabetes research 2012 2012

EN ? ⬇ ⬆ ⬇ ⬇ 11:44 PM 11/23/2011

Limit by disease and choose Breast Neoplasms...and click continue

HuGENavigator|HuGE Literature Finder|Search - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Data Rel 24/phaseII Nov08, ... x HuGENavigator|HuGE Literature Find... x +

http://www.hugenavigator.net/HuGENavigator/searchSummary.do

<input type="checkbox"/> Birth Weight		7
<input type="checkbox"/> Blindness		1
<input type="checkbox"/> Blood Coagulation Disorders		7
<input type="checkbox"/> Blood Coagulation Disorders, Inherited		7
<input type="checkbox"/> Blood Platelet Disorders		1
<input type="checkbox"/> Body Weight		4
<input type="checkbox"/> Bone Marrow Diseases		1
<input type="checkbox"/> Bone Neoplasms		1
<input type="checkbox"/> Bone Resorption		1
<input type="checkbox"/> Brain Damage, Chronic		1
<input type="checkbox"/> Brain Diseases		1
<input type="checkbox"/> Brain Infarction		2
<input type="checkbox"/> Brain Injuries		2
<input type="checkbox"/> Brain Ischemia		49
<input type="checkbox"/> Brain Neoplasms		4
<input checked="" type="checkbox"/> Breast Neoplasms		71
<input type="checkbox"/> Bronchial Hyperreactivity		2
<input type="checkbox"/> Bronchopulmonary Dysplasia		1
<input type="checkbox"/> Burkitt Lymphoma		1

C [^Top](#)

EN ? 11:47 PM 11/23/2011

Limit by study type and choose meta-analysis

HuGENavigator|HuGE Literature Finder|Search - Mozilla Firefox

File Edit View History Bookmarks Tools Help

HapMap Data Rel 24/phaseII Nov08, ... x HuGENavigator|HuGE Literature Find... x +

http://www.hugenavigator.net/HuGENavigator/searchSummary.do

HuGE Navigator > HuGE Literature Finder Last data upload: 23 Nov 2011. (Total 66054 articles)

HuGE Literature Finder

Data collected since 2001

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Search for

Search Criteria: MTHFR[Text+MeSH]>>Mammary Neoplasms[Mesh]>>Meta-analysis[StudyType] [\[Query Detail\]](#)

Filtered By ☐ Disease ☐ Gene ☐ StudyType ☐ Year ☐ Author ☐ Journal ☐ Country

Articles 1 - 8 of 8

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- [Polymorphic variants of folate metabolizing genes \(C677T and A1298C MTHFR, C1420T SHMT1 and G1958A MTHFD\) are not associated with the risk of breast cancer in West Siberian Region of Russia.](#) [\[Detail\]](#)
Molekuliarnaia biologii 2010 44 (5): 5.
Authors are not available
- [Lack of association between methylenetetrahydrofolate reductase gene A1298C polymorphism and breast cancer susceptibility.](#) [\[Detail\]](#)
Molecular biology reports 2010 Nov .
Giu LY, Zhang J, Li WH, Zhang Q, Xu H, Wang BY, Wang LB, Wang JL, Wang HJ, Liu XL, Luo ZC, Wu YH

Click on the title to view the PubMed abstract

The screenshot shows a Mozilla Firefox browser window with the title bar "Methylenetetrahydrofolate reductase gene and susc... [Clin Genet. 2006] - PubMed - NCBI - Mozilla Firefox". The address bar displays the URL "http://www.ncbi.nlm.nih.gov/pubmed/16630166?dopt=Abstract". The browser's menu bar includes File, Edit, View, History, Bookmarks, Tools, and Help. The address bar also shows several open tabs: "HapMap Data Rel 24/phaseII Nov08, ...", "HuGENavigator|HuGE Literature Find...", and the current tab "Methylenetetrahydrofolate reductase ...".

The main content area of the browser displays the PubMed website. The top navigation bar includes the NCBI logo, "Resources" (checked), "How To" (checked), "My NCBI", and "Sign In". Below this is the "PubMed.gov" logo, the text "US National Library of Medicine National Institutes of Health", a search bar with "PubMed" selected, and a "Search" button. Links for "Limits" and "Advanced" are also present. On the right side of the search bar, there is a "Help" link.

The abstract page shows the title "Methylenetetrahydrofolate reductase gene and susceptibility to breast cancer: a meta-analysis." by Zintzaras E. The abstract text states: "The methylenetetrahydrofolate reductase (MTHFR) gene polymorphisms have been linked to the risk of developing breast cancer. A meta-analysis of 18 case-control studies investigating the association between the C677T and the A1298C polymorphisms of the MTHFR gene and breast cancer (BC) was carried out. The meta-analysis included genotype data on 5467/7336 and 3768/5276 cases/controls for C677T and A1298C, respectively. In the meta-analysis, the consistency of genetic effects across different ethnicities and the effect of menopausal status for various genetic contrasts were investigated. The overall analysis for investigating the association between the C677T allele T and the risk of developing BC showed significant heterogeneity ($p = 0.08$, $I^2 = 34\%$) and non-significant association [odds ratio (OR) 1.02; 95% confidence interval (0.95-1.10)]. The allele contrast was not significant in Caucasians (nine studies) and in East Asians (four studies) [OR 1.03 (0.93-1.14) and OR 0.96 (0.81-1.15), respectively] or in pre-menopausal (five studies) and post-menopausal (four studies) groups [OR 1.10 (0.94-1.29) and OR 1.06 (0.95-1.18), respectively]. The genotype contrast of the homozygotes (TT vs CC)

On the right side of the abstract, there is a section titled "Related citations" with a list of related articles: "C677T and A1298C methylenetetrahydrofolate reductase gene polymorph [Psychiatr Genet. 2006]", "Methylenetetrahydrofolate reductase polymorphisms a [Breast Cancer Res Treat. 2010]", "Methylenetetrahydrofolate reductase C677T and A1298C polymorphisms and [Arch Med Res. 2010]", "Review Peripheral arterial disease and methylenetetrahydrofolate redu [J Vasc Surg. 2009]", and "Review 5,10-Methylenetetrahydrofolate reductase polymo [Cancer Epidemiol Biomarkers Prev. 2006]". Below this list are links for "See reviews..." and "See all...".

At the bottom of the browser window, the taskbar shows several icons: Windows Start button, Internet Explorer, Firefox, Google Chrome, and several application icons. The system clock in the bottom right corner shows "11:50 PM 11/23/2011".

Back to HuGE Literature Finder, click on HuGE Reviews, to find state-of-the-art open-access reviews for gene-disease associations

HuGENavigator|HuGE Literature Finder|Search - Mozilla Firefox

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HapMap Data Rel 24/phaseII Nov08, ... x HuGENavigator|HuGE Literature Find... x Methylenetetrahydrofolate reductase ... x +

http://www.hugenavigator.net/HuGENavigator/startPagePubLit.do

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HuGE Navigator > HuGE Literature Finder Last data upload: 23 Nov 2011. (Total 66054 articles)

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Search Literature for MTHFR Go Clear

Queries for the summary

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- [Meta-analysis](#)
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- [Clinical trial](#)

- Enter search terms into the text box.
- Search terms can include disease, exposure, gene, author, journal, etc.
- Simple Boolean operators are allowed, such as AND or OR.
- Use the Search dropdown list to switch to other HuGE Navigator applications.

HuGE Literature Finder is a search engine for finding published literature on genetic associations and other human genome epidemiology. The search query can include disease/outcome, environmental factors, genes, author's name, affiliation, etc. The results can be further refined by the filtering feature. The list of selected articles can be redirected to the PubMed website to take advantage of the functionality it provides, such as

Last Update: 23 Nov 2011

EN ? 11:51 PM 11/23/2011

HuGENavigator|HuGE Literature Finder|Search - Mozilla Firefox

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HapMap Data Rel 24/phaseII Nov08, ... x HuGENavigator|HuGE Literature Find... x Methylenetetrahydrofolate reductase ... x +

http://www.hugenavigator.net/HuGENavigator/searchSummary.do?firstQuery=HuGE+review&publitSearc ☆ ↻ Google

HuGE Navigator > HuGE Literature Finder Last data upload: 23 Nov 2011. (Total 66054 articles)

HuGE Literature Finder

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Search Literature ▼ for HuGE review

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Search Criteria: HuGE Review[StudyType] [\[Query Detail\]](#) Download

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Articles 1 - 25 of 92

Export Display 25 ▼ on Page 1 of 4 >>

1.	-251 T/A polymorphism of the interleukin-8 gene and cancer risk: a HuGE review and meta-analysis based on 42 case-control studies. [Detail] Molecular biology reports 2011 Jun . Wang N, Zhou R, Wang C, Guo X, Chen Z, Yang S, Li Y
2.	Glutathione S-Transferase M1 (GSTM1) and Glutathione S-Transferase T1 (GSTT1) Null Polymorphisms, Smoking, and Their Interaction in Oral Cancer: A HuGE Review and Meta-Analysis. [Detail]

EN ? ▲ ▼ 11:52 PM 11/23/2011



Useful links

- <http://www.genecards.org/>

GeneCards is an integrated database of human genes that includes automatically-mined genomic, proteomic and transcriptomic information, as well as orthologies, disease relationships, SNPs, gene expression, gene function, and service links for ordering assays and antibodies.

GeneCards V3 - Human Genes | Gene Database | Gene Search - Mozilla Firefox

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http://www.genecards.org/

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neurotrophic tyrosine kinase, receptor, type 1

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- Field-specific input
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Version 3.07

EN ?

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11/23/2011

- <http://www.snps3d.org/>
- **SNPs3D** is a website which assigns molecular functional effects of non-synonymous SNPs based on structure and sequence analysis.
- Gene-gene interactions networks

(3949)LDLR : **Disease Gene**;click **Node** for detailed info.

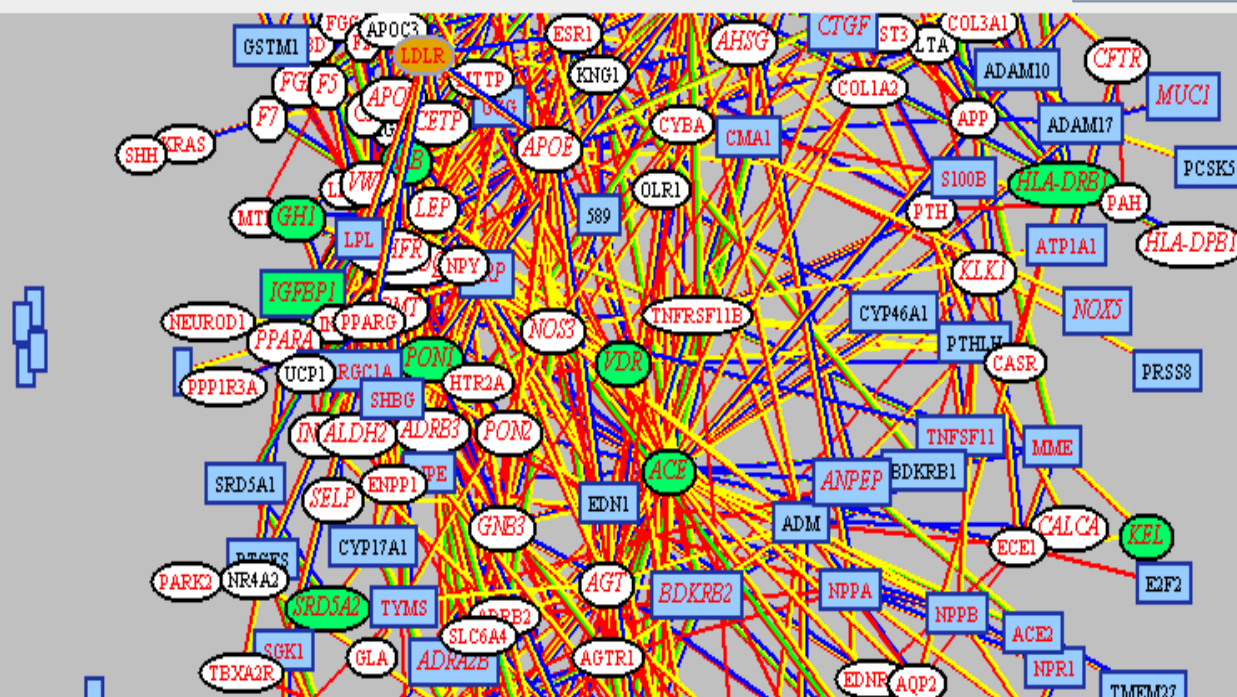
Mouse KnockOut: gene knockouts which are compatible with viability:cardiovascular system;

GO: plasma membrane; lipid metabolic process; steroid metabolic process; lipoprotein metabolic process; clathrin-coated endocytic vesicle membrane; very-low-density lipoprotein re

☒ Gene Symbol ☐ Keyword ☐ Gene IDs

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Genetic Association Database

- archive of human genetic association studies of complex diseases and disorders. The goal of this database is to allow the user to rapidly identify medically relevant polymorphism from the large volume of polymorphism and mutational data, in the context of standardized nomenclature.
- The data is from published scientific papers.
- additional molecular reference numbers and links.
- gene centered.
- <http://geneticassociationdb.nih.gov/>

Gene View

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http://www.snps...=ACE&type=gene

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http://geneticassociationdb.nih.gov/cgi-bin/tableview.cgi?table=geneview

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For a complete gene list, click [here](#).

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Advanced Search

Batch Search

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Gene View

Search for All

Re...

	Assoc? YorN	Gene Symbol	OMIM	Gene Expert	Gene Name	Unigene Cluster	Entrez GeneID	Chr	Ch-Band	DNA Start(bp)	DNA End(bp)	Rep seq	EG	GC	Ace View	BBID	e!	PUB MED
view		3.8-1.2						6	6p21.3					GC		BBID	e!	PM
view	Y	A2BP1	605104		ataxin 2-binding protein 1	Hs.459842	54715	16	16p13.3	6069131	7763340	R	EG	GC		BBID	e!	PM
view		A2BP1	605104		ataxin 2-binding protein 1			16	10q12	6366996	7763340			GC		BBID	e!	PM
view		A2BP1	605104		ataxin 2-binding protein 1	Hs.459842	54715	16	16p13.3	6069131	7763340	R	EG	GC		BBID	e!	PM
view		A2BP1	605104		ataxin 2-binding protein 1	Hs.459842	54715	16	16p13.3	6069131	7763340	R	EG	GC		BBID	e!	PM
view		A2BP1	605104		ataxin 2-binding protein 1			16	10q12	6366996	7763340			GC		BBID	e!	PM

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Genetic Association Database

P Q R S T U V W X Y Z

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Unigene Cluster	Entrez GeneID	Chr	Ch-Band	DNA Start(bp)	DNA End(bp)	Rep seq	EG	GC	Acc View	BBID	e!	PUB MED	P Value	Disease Class	Broad Phenotype (Disease)	Reference
		6	6p21.3					GC		BBID	e!	PM		UNKNOWN	Atherosclerosis Hyperlipidemia	Felekis T et al. 2010
Hs.459842	54715	16	16p13.3	6069131	7763340	R	EG	GC		BBID	e!	PM		METABOLIC	Osteoarthritis	G Zhai , et al. Journal of medic
		16	10q12	6366996	7763340			GC		BBID	e!	PM		METABOLIC	obesity	Ma L et al. 2010
Hs.459842	54715	16	16p13.3	6069131	7763340	R	EG	GC		BBID	e!	PM		PSYCH	ADHD attention-deficit hyper	J Elia , et al. Molecular psychia
Hs.459842	54715	16	16p13.3	6069131	7763340	R	EG	GC		BBID	e!	PM		CHEMDEPENDENCY	Tobacco Use Disorder	George R Uhl , et al. Archives o
		16	10q12	6366996	7763340			GC		BBID	e!	PM		METABOLIC	BILIARY CIRRHOSIS Liver Cirrho	Joshita S et al. 2010

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Date Added to Catalog (since 11/25/08)	First Author/Date/ Journal/Study	Disease/Trait	Initial Sample Size	Replication Sample Size	Region	Reported Gene(s)	Mapped Gene(s)	Strongest SNP-Risk Allele	Con
11/18/11	Frank J October 18, 2011 <i>Addict Biol</i> Genome-wide significant association between alcohol dependence and a variant in the ADH gene cluster.	Alcohol dependence	1,333 European ancestry cases, 2,168 European ancestry controls	NR	4q23 4p16.2 14q24.2 2q35 13q12.12 3p22.3	ADH1B, ADH1C NR NR NR NR GPD1L	ADH1B - ADH1C STX18 - MSX1 PCNX MREG - PECR SGCG RPSAP11 - CMTM8	rs1789891-? rs1000579-? rs2810114-? rs1344694-? rs4770403-? rs9825310-?	intergenic intergenic intron intergenic UTR-5 intergenic
11/19/11	Hollingsworth P October 18, 2011	Alzheimer's disease	1,299 European ancestry,	NR	4q12 2p16.3	AC110611.1 AC079250.1	NMU - EXOC1 KCNK12 -	rs753129-? rs2969775-?	intergenic intergenic



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GENETIC EPIDEMIOLOGY GLOSSARY

M.Tevfik DORAK

Accompanying [Genetic Epidemiology Lecture Note](#) & [Presentation](#)

ACCE project (analytic validity, clinical validity, clinical utility, ELSI): A CDC sponsored project for evaluating data on emerging genetic tests. It takes its name from the four components of evaluation: analytic validity, clinical validity, clinical utility and associated ethical, legal and social implications (ELSI). For details, see [ACCE Project website](#). See also [Grosse & Khoury, 2006](#) for the clinical utility of genetic testing, [Offit, 2008](#) for issues surrounding genomic disease profiling, and [Pharoah, 2008](#) for the possible utility of genomic profiling in breast cancer risk assessment.

Additive genetic model: In a disease association study, if the risk conferred by an allele is increased r -fold for heterozygotes and $2r$ -fold for homozygotes, this corresponds to additive model ([Lewis, 2002](#)). These data are best analyzed using Armitage trend test for genotype frequencies or by logistic regression in which the genotypes are represented as (-1) , 0 , $(+1)$. This genotype-based association test does not require the locus to be in **Hardy-Weinberg equilibrium**. In the case of an association with heterozygosity, the additive model test may be statistically non-significant despite the presence of an association. Thus, a non-significant additive model test does not rule out an association. It has been pointed out that "genes do not generally act in a simple additive manner but through complex networks involving gene-gene and gene-environment interactions" ([Colhoun, 2003](#)). The effect that cannot be explained by an additive (or heterogeneity / non-interactive) model in complex disease genetics is due to the dominance (epistatic / interactive) model. See also **multiplicative genetic model**. See [MODEL](#)-online tool for genetic association analysis for different models.

Additive variance: The component of genetic variance due to the additive effects of alleles segregating in the population. In evolutionary genetics, additive genetic variance is a measure for the potential amount of evolutionary change caused by natural selection. See [Genetic Calculation Applets](#):



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The
"PRIZE OF DISTINGUISHED RESEARCHER IN MEDICINE
AT
UNIVERSITY OF THESSALY"
was awarded to
Elias Zintzaras








Research Interests

- Mathematical modelling of genetic systems
- Genetic statistics and epidemiology
- Bioinformatics
- Meta-analysis
- Monte-Carlo modelling
- Bioequivalence testing



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