

# Navigating the Epidemiology of the Human Genome

CCGTCGACTGGAGTGTCTGTGAATTGACTTTTGTGCCAGTTGGCAGCGGCAGAAGCAGCAAAGCCC GGCCAACAGCAACAAGCTCCTGCCAGATCCCAAAGCAAACAGT

Marta Gwinn, MD, MPH  
National Office of Public Health Genomics, CDC

Personal Genomics Workshop  
Bethesda, MD – December 17, 2008



# Navigating the Epidemiology of the Human Genome

CCGTCGACTGGAGTGTCTGTGAATTGACTTTTTGTTGCCAGTTGGCAGCGGCAGAAGCAGCAAAGCCCGGCCAACAGCAACAAGCTCCTGCCAGATCCCAAAGCAAACACGT

## Size up the domain

- population-based research studies

## Map the topography

- population prevalence, associations, interactions, genetic tests

## Identify major features

- consistent findings, patterns, and gaps

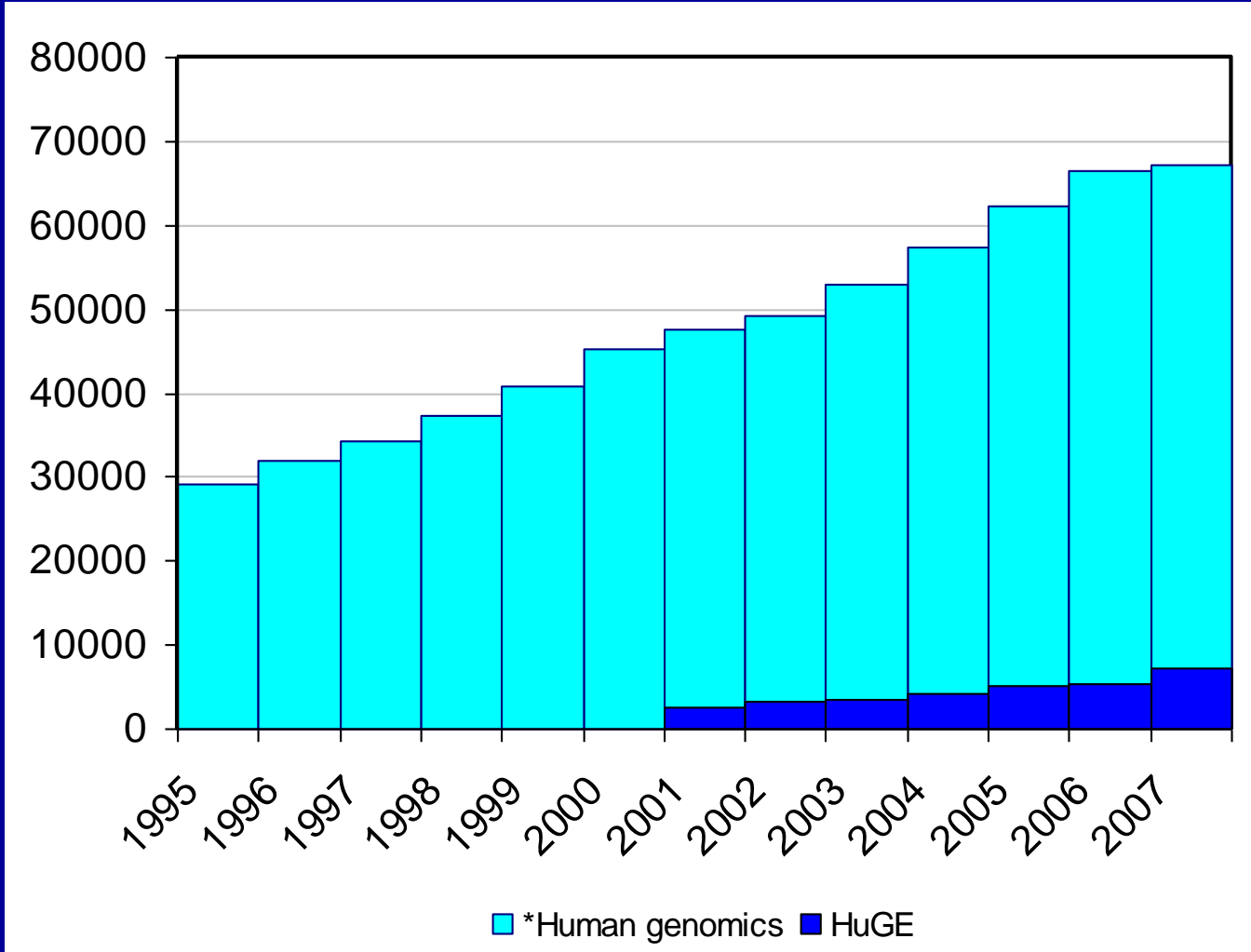
## Develop navigational tools for further exploration

- reliable knowledge base with applications



[www.hugenavigator.net](http://www.hugenavigator.net)

# PubMed citations on human genetics/genomics and human genome epidemiology (HuGE)



\*PubMed query: "gene or genetic or genome or genomic" / limits: human, year and HuGE Navigator, Oct 2008



# Size up the domain

(( (((((((((((((((((((genetic[All Fields] AND (((("disease"[MeSH Terms] OR ("disease susceptibility"[MeSH Terms] OR predisposition[Text Word]) OR disease[Text Word] OR defect[Text Word] OR susceptibility[Text Word]) OR ("counseling"[MeSH Terms] OR counseling[Text Word])) OR (("disease susceptibility"[MeSH Terms] OR susceptibility[Text Word] AND ("genes"[MeSH Terms] OR gene[Text Word] OR ("genes"[MeSH Terms] OR genes[Text Word])) OR (((("mutation"[MeSH Terms] OR mutation[Text Word] OR ("genes"[MeSH Terms] OR gene[Text Word] AND ("mutation"[MeSH Terms] OR mutation[Text Word])) OR (("mutation"[MeSH Terms] OR mutations[Text Word] AND ("genes"[MeSH Terms] OR gene[Text Word])) OR (("mutation"[MeSH Terms] OR mutations[Text Word] OR mutation[Text Word] AND ("genes"[MeSH Terms] OR gene[Text Word])) OR ("hereditary diseases"[MeSH Terms] OR genetic disorder[Text Word]) OR (genetic[All Fields] AND (((("TEST"[Substance Name] OR ("TEST"[Substance Name] OR test[Text Word])) OR ("research design"[MeSH Terms] OR testing[Text Word]) OR study[All Fields])) OR ("genetic screening"[MeSH Terms] OR genetic screening[Text Word]) OR (genetic[All Fields] AND ("risk"[MeSH Terms] OR risk[Text Word])) OR ("polymorphism (genetics)"[MeSH Terms] OR ("polymorphism (genetics)"[MeSH Terms] OR polymorphism[Text Word])) OR (((("genotype"[MeSH Terms] OR ("genotype"[MeSH Terms] OR genotype[Text Word]) OR genotyping[All Fields]) OR ("haplotypes"[MeSH Terms] OR haplotype[Text Word]) OR ("haplotypes"[MeSH Terms] OR haplotypes[Text Word])) OR (((("genome"[MeSH Terms] OR genome[Text Word] OR genomic[All Fields]) OR ("Genomics"[MeSH Terms] OR genomics[Text Word])) OR (((gene-environment) OR (gene AND environment)) AND interaction[Text Word]) OR (((genetic[Text Word] OR gene[Text Word] OR allelic[All Fields]) AND ((variant[All Fields] OR variants[All Fields]) OR ("epidemiology"[MeSH Subheading] OR "epidemiology"[MeSH Terms] OR frequency[Text Word])) OR ("alleles"[MeSH Terms] OR allele[Text Word] OR ("alleles"[MeSH Terms] OR alleles[Text Word])) OR ("heterozygote detection"[MeSH Terms] OR Heterozygote Detection[Text Word]) OR ((Neonatal[All Fields] OR ("infant, newborn"[MeSH Terms] OR newborn[Text Word]) AND ("diagnosis"[MeSH Subheading] OR "mass screening"[MeSH Terms] OR Screening[Text Word])) OR germline[All Fields] OR somatic[All Fields] OR ("human genome project"[MeSH Terms] OR human genome project[Text Word]) AND (((((((((((((((((((("epidemiology"[Subheading] OR "epidemiology"[MeSH Terms] OR epidemiology[Text Word] OR ("public health"[MeSH Terms] OR public health[Text Word]) OR (((("alleles"[MeSH Terms] OR allele[Text Word] OR allelic[All Fields]) AND (((("epidemiology"[MeSH Subheading] OR "epidemiology"[MeSH Terms] OR frequency[Text Word] OR frequencies[All Fields])) OR ("public policy"[MeSH Terms] OR policy[Text Word]) OR ("education"[Subheading] OR "education"[MeSH Terms] OR education[Text Word]) OR "prevalence"[MeSH Terms] OR prevalence[Text Word]) OR ("prevention and control"[Subheading] OR prevention[Text Word]) OR ("risk"[MeSH Terms] OR risk[Text Word]) OR (((((((population[Text Word] OR (a number of) OR genetic[All Fields]) OR comparative[All Fields]) OR prospective[All Fields] OR cohort[All Fields] OR cross-section[All Fields] OR cross-sectional[All Fields] OR case-control[All Fields] AND (studies OR study[All Fields])) OR (clinical trial[All Fields] OR randomized controlled trial[All Fields]) OR ("drug interactions"[MeSH Terms] OR interactions[Text Word]) OR ("interpersonal relations"[MeSH Terms] OR "drug interactions"[MeSH Terms] OR interaction[Text Word])) OR ("questionnaires"[MeSH Terms] OR questionnaire[Text Word]) OR ((("sensitivity and specificity"[MeSH Terms] OR sensitivity[Text Word] OR ("sensitivity and specificity"[MeSH Terms] OR specificity[Text Word])) OR (((case[All Fields] OR cases[All Fields]) OR ("patient"[MeSH Terms] OR patients[Text Word]) OR (study[All Fields] AND group[All Fields])) OR (((("prevention and control"[MeSH Subheading] OR control[Text Word] OR controls[All Fields] OR (healthy[All Fields] AND subjects[All Fields])) OR ("child"[MeSH Terms] OR children[Text Word]) OR ("adult"[MeSH Terms] OR adults[Text Word]) OR individuals[All Fields])) OR (((("association"[MeSH Terms] OR association[Text Word] OR ("association"[MeSH Terms] OR associations[Text Word]) OR ("disease"[MeSH Terms] OR disease[Text Word]) AND ("genes"[MeSH Terms] OR gene[Text Word] OR ("genes"[MeSH Terms] OR genes[Text Word])) OR oversight[All Fields] OR (((("genotype"[MeSH Terms] OR genotype[All Fields] OR allelic[All Fields] AND (disorder[Text Word] OR ("genotype"[MeSH Terms] OR genotype[Text Word] AND ("phenotype"[MeSH Terms] OR phenotype[Text Word]) OR genotype[Text Word] OR phenotype[All Fields] AND correlation[All Fields]) OR ((positive OR negative) AND predictive value)) OR (odds ratio)) OR ((("ethics"[MeSH Terms] OR ethics[Text Word] OR ethical[All Fields])) ) ) AND "2004/7/7 8.00"[MHDA]:"2004/7/14 8.00"[MHDA])



HuGE search query 2004



# Knowledge base for Human Genome Epidemiology

GTGCGACTGGAGTGTCTGTGAATTGACTTTTTGTTGCCAGTTGGCAGCGGCAGAAAGCAGCAAAGCCCCGGCCAACAGCAACAAGCTCCTGCCAGATCCCCAAAAGCAAACAGG

- prevalence, associations, interactions, genetic tests
- updated weekly from PubMed since 2001
- combination of human and machine curation processes



SVM screens PubMed



Curator selects, indexes



Auto-indexing (Entrez Gene, MeSH, UMLS)



Web applications search, sort, filter, display data

[www.hugenavigator.net](http://www.hugenavigator.net)

# HuGE Navigator: [www.hugenavigator.net](http://www.hugenavigator.net)



## HuGE Navigator (version 1.3)

An integrated, searchable knowledge base of genetic associations and human genome epidemiology.

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HuGE Navigator

**HuGEpedia** - an encyclopedia of human genetic variation in health and disease.



### [Phenopedia](#)

Look up gene-disease association summaries by disease.



### [Genopedia](#)

Look up gene-disease association summaries by gene.

**HuGETools** - searching and mining the literature in human genome epidemiology.



### [HuGE Literature Finder](#)

Find published articles in human genome epidemiology.



### [HuGE Investigator Browser](#)

Find investigators in a particular field of human genome epidemiology.



### [Gene Prospector](#)

## About the Navigator

HuGE Navigator provides access to a continuously updated knowledge base in human genome epidemiology, including information on population prevalence of genetic variants, gene-disease associations, gene-gene and gene-environment interactions, and evaluation of genetic tests ... [more](#)

## What's New

- New Publication, [Gene Prospector: An evidence gateway for evaluating potential susceptibility genes and interacting risk factors for human diseases](#). BMC Bioinformatics 2008, 9:528  
Wei Yu, Anja Wulf, Tiebin Liu, Muin J. Khoury and Marta Gwinn (12/10/2008)
- A new version of HuGE Navigator (1.3) has been launched. The new version contains:
  - a new application, [Variant Name Mapper](#), for

# HuGE Published Literature

GTGCGACTGGAGTGTCTGTGAATTGACTTTTTGTTGCCAGTTGGCAGCGGCAGAAAGCAGCAAAGCCCCGGCCAACAGCAACAAGCTCCTGCCAGATCCCCAAAAGCAAACAGG

As of December 11, 2008, the knowledge base contained:

34,208 genetic association studies

863 meta-analyses

243 GWAS

3,888 genes

1,958 MeSH disease terms

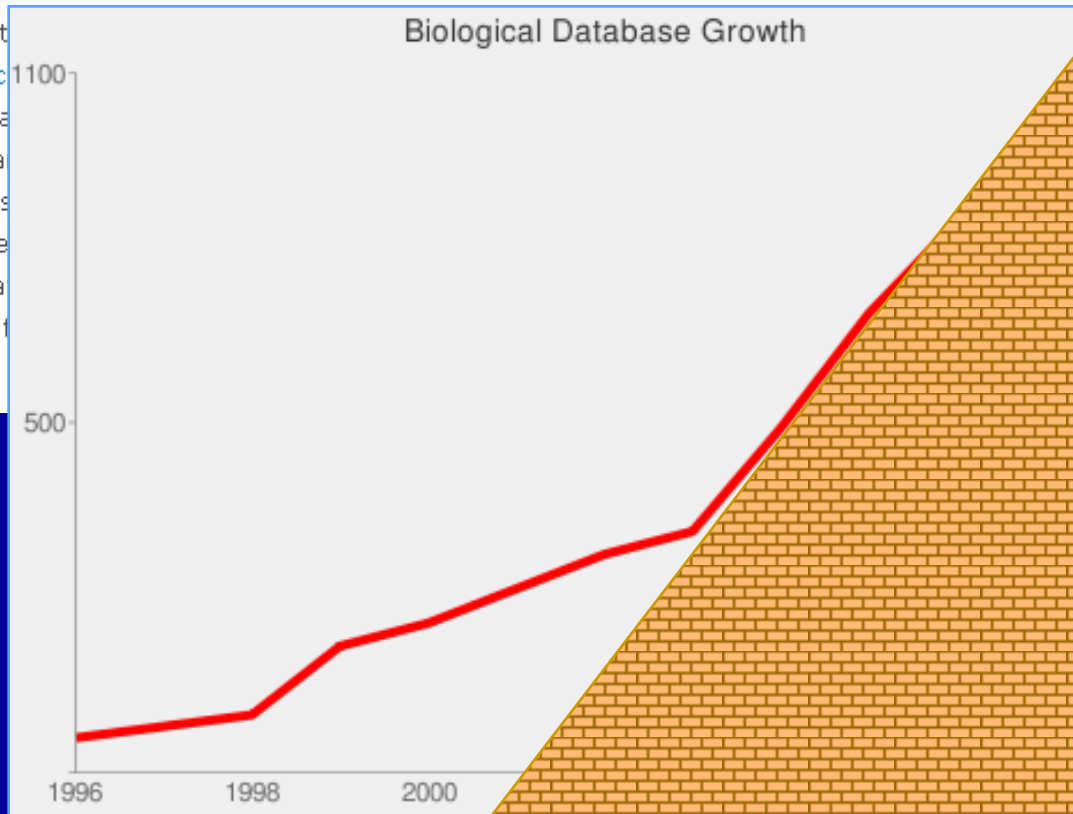
5,645 common variant names matched to rs numbers

*...but numbers alone mean little...*

## One Thousand Databases High (and rising)

Fri, 2008-01-18 21:08 — Duncan

Well it's that stamp collection also known as published databases list previous one thousand data proportion of be used?



data tombs

*Nucleic Acids Research, 2008*







### Flying Over the Columbia Hills of Mars

Animated Illustration Credit: [Doug Ellison](#), [Randolph Kirk \(USGS\)](#), [MSSS](#), [MER](#), [NASA](#)



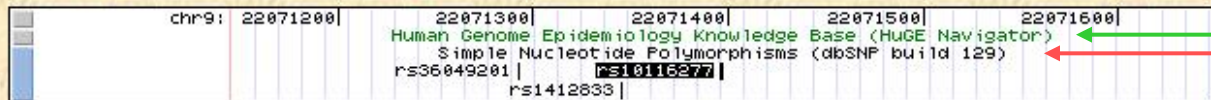
<http://apod.nasa.gov/apod/ap080519m.html>



# UCSC Genome Browser on Human Mar. 2006 Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

position/search chr9:22,071,147-22,071,647 jump clear size 501 bp. configure



move start < 2.0 > Click on a feature for details. Click on base position to zoom in around move end < 2.0 >  
cursor. Click gray/blue bars on left for track options and descriptions.  
default tracks hide all manage custom tracks configure reverse refresh

Use drop-down controls below and press refresh to alter tracks displayed.  
Tracks with lots of items will automatically be displayed in more compact modes.

## Custom Tracks refresh

HuGE  
full

## Mapping and Sequencing Tracks refresh

<u>Base Position</u> dense	<u>Chromosome Band</u> hide	<u>STS Markers</u> hide	<u>FISH Clones</u> hide	<u>Recomb Rate</u> hide
<u>Map Contigs</u> hide	<u>Assembly</u> hide	<u>Gap</u> hide	<u>Coverage</u> hide	<u>BAC End Pairs</u> hide
<u>Fosmid End Pairs</u> hide	<u>GC Percent</u> hide	<u>Short Match</u> hide	<u>Restr Enzymes</u> hide	

view for rs10116277 on UCSC Genome Browser

<http://genome.ucsc.edu/>

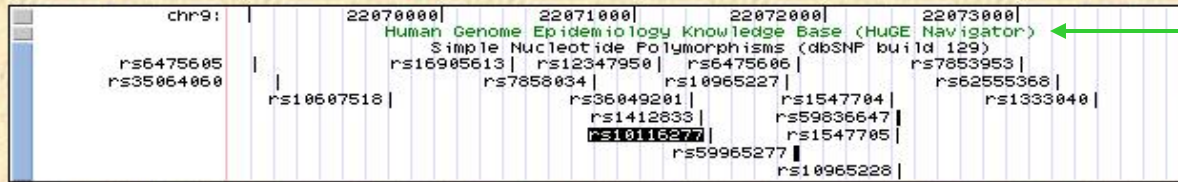




# UCSC Genome Browser on Human Mar. 2006 Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

position/search chr9:22,068,892-22,073,901 jump clear size 5,010 bp. configure



move start Click on a feature for details. Click on base position to zoom in around move end  
 < 2.0 > cursor. Click gray/blue bars on left for track options and descriptions. < 2.0 >  
 default tracks hide all manage custom tracks configure reverse refresh

Use drop-down controls below and press refresh to alter tracks displayed.  
 Tracks with lots of items will automatically be displayed in more compact modes.

**Custom Tracks** refresh

[HuGE](#)  
 full

**Mapping and Sequencing Tracks** refresh

<u>Base Position</u> dense	<u>Chromosome Band</u> hide	<u>STS Markers</u> hide	<u>FISH Clones</u> hide	<u>Recomb Rate</u> hide
<u>Map Contigs</u> hide	<u>Assembly</u> hide	<u>Gap</u> hide	<u>Coverage</u> hide	<u>BAC End Pairs</u> hide
<u>Fosmid End Pairs</u> hide	<u>GC Percent</u> hide	<u>Short Match</u> hide	<u>Restr Enzymes</u> hide	

10x view for rs10116277 on UCSC Genome Browser

# UCSC Genome Browser on Human Mar. 2006 Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

position/search chr9:22,046,347-22,096,446 jump clear size 50,100 bp. configure



100x

chr9:	22060000	22070000	22080000	22090000
	Human Genome Epidemiology Knowledge Base (HuGE Navigator)			
	Simple Nucleotide Polymorphisms (dbSNP build 129)			
rs7866783	rs58814679	rs34665955	rs7855660	rs1970112
rs10120688	rs8181050	rs4451405	rs6475605	rs5896963
rs13292618	rs8181047	rs35307545	rs35064060	rs5896964
rs10121501	rs10811647	rs4645630	rs10607510	rs7857345
rs58096158	rs1333038	rs9632884	rs16905613	rs10738606
rs10522721	rs4144664	rs9632885	rs7858034	rs10738607
rs56272822	rs1333039	rs10965226	rs12347950	rs56321905
rs13299593	rs10531705	rs1855185	rs36049201	rs3222818
rs7021816	rs4977755	rs7855162	rs6475606	rs12235973
rs10757268	rs59732106	rs56965302	rs1547704	rs10757273
rs2095144	rs5896962	rs57076710	rs1547705	rs10965230
rs2383205	rs1412831	rs1831733	rs7853953	rs9644859
rs2184061	rs4977756	rs1831734	rs1333040	rs9644860
rs1537378	rs60687076	rs59616750	rs34538587	rs7034707
rs4977754	rs35572758	rs10757271	rs1537370	rs9644861
rs1011970	rs34871414	rs10811652	rs10122192	rs7866503
rs10522515	rs59584319	rs1412832	rs60166428	rs56064546
rs10965221	rs60332557	rs35390589	rs3028398	rs4007642
	rs34863803	rs1412833	rs16905640	rs16905644
	rs36028879	<b>rs10116277</b>	rs13300638	rs35998780
	rs34660701	rs10965227	rs13284693	rs1333041
	rs28557075	rs59965277	rs7019916	rs11286208
	rs10965223	rs59836647	rs7020031	rs59642159
	rs62560776	rs10965228	rs34172764	rs2891168
	rs62560777	rs62555368	rs9644862	rs10965231
	rs10965224	rs58419961	rs9722878	rs12238050
	rs60537401	rs59552649	rs3955237	rs10965232
	rs10811648	rs10120722	rs4977757	rs10652638
	rs10811649	rs16905635	rs10738608	rs6475608
	rs10811650	rs10757272	rs35568006	
	rs10811651	rs56937835	rs11787814	
	rs16905597	rs55672229	rs35147044	
	rs55936329	rs10811653	rs60850854	
	rs16905599	rs12005039	rs11437218	
	rs7042970	rs7869527	rs34107457	
	rs57222759	rs35368490	rs13292938	
	rs57840055	rs34857236	rs10681138	
	rs10539741	rs60356909	rs34108326	

100x view for rs10116277 on UCSC Genome Browser



<http://genome.ucsc.edu/>





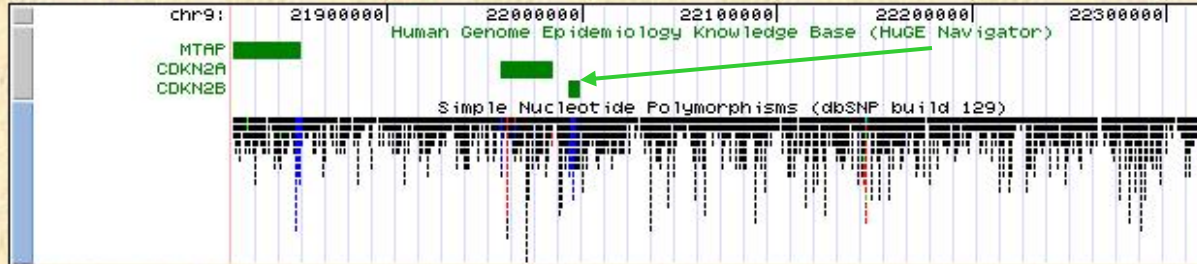
# UCSC Genome Browser on Human Mar. 2006 Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

position/search chr9:21,820,897-22,321,896 jump clear size 501,000 bp. configure



1000x



move start Click on a feature for details. Click on base position to zoom in around move end  
< 2.0 > cursor. Click gray/blue bars on left for track options and descriptions. < 2.0 >  
default tracks hide all manage custom tracks configure reverse refresh

Use drop-down controls below and press refresh to alter tracks displayed.  
Tracks with lots of items will automatically be displayed in more compact modes.

**Custom Tracks** refresh

HuGE  
full

**Mapping and Sequencing Tracks** refresh

Base Position Chromosome Band STS Markers FISH Clones Recomb Rate  
dense hide hide hide hide

1000x view for rs10116277 on UCSC Genome Browser

<http://genome.ucsc.edu/>





## Genopedia

Data collected since 2001

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Search

Genopedia



for

Enter a gene symbol (alias) or a protein name

Go

Clear

All

Download

**CDKN2B** SNP

- Related Disease Genes -

### Summary

- [Total Publications](#)  
45
- [Gene Prevalence](#)  
2
- [Meta-Analyses](#)  
5
- [MA Summary](#)  
NA
- [Disease](#)  
23
- [Investigator](#)  
82(F/L) / 534(All)
- [Trend](#)

22 disease terms (MeSH) have been reported with CDKN2B gene.

[Click to re-sort the table]

<a href="#">Disease Term (MeSH)</a>	Total <a href="#">?</a>	Meta <a href="#">?</a>	GWAS <a href="#">?</a>	Gene-Env <a href="#">?</a>	Trend <a href="#">?</a>
<a href="#">Diabetes Mellitus, Type 2</a>	17	4	3	1	
<a href="#">Melanoma</a>	3	0	0	0	
<a href="#">Breast Neoplasms</a>	2	0	0	0	
<a href="#">Insulin Resistance</a>	2	1	1	0	
<a href="#">Coronary Arteriosclerosis</a>	2	0	2	0	
<a href="#">Coronary Disease</a>	2	0	1	0	
<a href="#">Ovarian Neoplasms</a>	2	0	0	0	
<a href="#">Skin Neoplasms</a>	1	0	0	0	
<a href="#">Thyroid Neoplasms</a>	1	0	0	0	
<a href="#">Uveal Neoplasms</a>	1	0	0	0	
<a href="#">Diabetes Mellitus</a>	1	0	0	0	
<a href="#">Diabetes Mellitus, Type 1</a>	1	1	0	0	
<a href="#">Glucose Intolerance</a>	1	0	0	0	
<a href="#">Hyperglycemia</a>	1	0	0	0	
<a href="#">Hyperlipoproteinemia Type II</a>	1	0	0	0	

Links:

- [Entrez Gene](#)
- [GeneCard](#)
- [PharmGKB](#)
- [CIP](#)





## HuGE Literature Finder

Data collected since 2001

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Search  for

**Search Criteria:** Coronary Arteriosclerosis and gwas and CDKN2B[Text+MeSH] [\[Query Detail\]](#)

Filtered By  Disease  Gene  Category  StudyType  Year  Author  Journal  Country

Articles 1 - 2 of 2

[Export](#) Display  on Page  of 1

[A common allele on chromosome 9 associated with coronary heart disease. \[Detail\]](#)

**GWAS**

1. Science (New York, N.Y.) 2007 Jun 316 (5830): 1488-91.  
McPherson R, Pertsemidis A, Kavaslar N, Stewart A, Roberts R, Cox DR, Hinds DA, Pennacchio LA, Tybjaerg-Hansen A, Folsom AR, Boerwinkle E, Hobbs HH, Cohen JC

[A common variant on chromosome 9p21 affects the risk of myocardial infarction. \[Detail\]](#)

**GWAS**

2. Science (New York, N.Y.) 2007 Jun 316 (5830): 1491-3.  
Helgadóttir A, Thorleifsson G, Manolescu A, Gretarsdóttir S, Blondal T, Jonasdóttir A, Jonasdóttir A, Sigurdsson A, Baker A, Palsson A, Masson G, Gudbjartsson DF, Magnusson KP, Andersen K, Levey AI, Backman VM, Matthiasdóttir S, Jonsdóttir T, Palsson S, Einarsdóttir H, Gunnarsdóttir S, Gylfason A, Vaccarino V, Hooper WC, Reilly MP, Granger CB, Austin H, Rader DJ, Shah SH, Quyyumi AA, Gulcher JR, Thorgeirsson G, Thorsteinsdóttir U, Kong A, Stefansson K

[Export](#) Display 25 on page 1 of 1

Articles 1 - 2 of 2




**CDKN2B** <sup>SNP</sup>

## - Related Disease Genes -


**Summary**
















































22 disease terms (MeSH) have been reported with CDKN2B gene.

- ② Total Publications [45](#)
- ② Gene Prevalence [2](#)
- ② Meta-Analyses [5](#)
- ② MA Summary NA
- ② Disease 23
- ② Investigator [82\(F/L\)/\\_534\(All\)](#)
- ② Trend 

## Links:

- [Entrez Gene](#)
- [GeneCard](#)
- [PharmGKB](#)
- [GHR](#)
- [OMIM](#)
- [dbSNP](#)
- [more ...](#)

[Click  to re-sort the table]

 ② Disease Term (MeSH)	 Total ②	 Meta ②	GWAS ②	Gene-Env ②	Trend ②
<a href="#">Diabetes Mellitus, Type 2</a> 	<a href="#">17</a>	<a href="#">4</a>	<a href="#">3</a>	<a href="#">1</a>	
<a href="#">Melanoma</a> 	<a href="#">3</a>	0	0	0	
<a href="#">Breast Neoplasms</a> 	<a href="#">2</a>	0	0	0	
<a href="#">Insulin Resistance</a> 	<a href="#">2</a>	<a href="#">1</a>	<a href="#">1</a>	0	
<a href="#">Coronary Arteriosclerosis</a> 	<a href="#">2</a>	0	<a href="#">2</a>	0	
<a href="#">Coronary Disease</a> 	<a href="#">2</a>	0	<a href="#">1</a>	0	
<a href="#">Ovarian Neoplasms</a> 	<a href="#">2</a>	0	0	0	
<a href="#">Skin Neoplasms</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Thyroid Neoplasms</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Uveal Neoplasms</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Diabetes Mellitus</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Diabetes Mellitus, Type 1</a> 	<a href="#">1</a>	<a href="#">1</a>	0	0	
<a href="#">Glucose Intolerance</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Hyperglycemia</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Hyperlipoproteinemia Type II</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Leukemia, Lymphocytic, Acute, L1</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Leukemia, Pre-B-Cell</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Lymphoma, Non-Hodgkin</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Carcinoma, Medullary</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Colorectal Neoplasms</a> 	<a href="#">1</a>	0	0	0	
<a href="#">Myocardial Infarction</a> 	<a href="#">1</a>	0	<a href="#">1</a>	0	
<a href="#">Obesity</a> 	<a href="#">1</a>	0	0	0	

Note: The number of publications displayed in this table will differ from the number displayed in the HuGE Literature Finder as the number in GenoPedia reflects only the indexed disease term without children terms, but the number in the HuGE Literature Finder reflects all text searches of the disease term including the indexed term and corresponding children terms.







## Phenopedia

Data collected since 2001

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Search  for

## Myocardial Infarction

- Related Diseases -

### Summary

367 genes have been reported with Myocardial Infarction

[Total Publications](#)  
[1012](#)

[Meta-Analyses](#)  
[30](#)

[MA Summary](#)

[Genes](#)  
318

[GWAS Publications](#)  
[11](#)

[Investigators](#)  
[1315\(F/L\)](#)  
[4753\(All\)](#)

[Trend](#)

Last Update: 11 Dec 2008

[Field Synopsis](#)

NA

[Click to re-sort the table]

Group genes by [KEGG](#)

Gene <a href="#">?</a>	Total <a href="#">?</a>	Meta <a href="#">?</a>	GWAS <a href="#">?</a>	Gene-Env <a href="#">?</a>	Trend <a href="#">?</a>
<a href="#">ACE</a> <small>SNP</small>	<a href="#">80</a>	<a href="#">4</a>	0	<a href="#">17</a>	
<a href="#">APOE</a> <small>SNP</small>	<a href="#">49</a>	<a href="#">1</a>	0	<a href="#">9</a>	
<a href="#">MTHFR</a> <small>SNP</small>	<a href="#">45</a>	<a href="#">3</a>	0	<a href="#">5</a>	
<a href="#">NOS3</a> <small>SNP</small>	<a href="#">42</a>	<a href="#">1</a>	0	<a href="#">7</a>	
<a href="#">F5</a> <small>SNP</small>	<a href="#">41</a>	<a href="#">3</a>	0	<a href="#">4</a>	
<a href="#">SERPINE1</a> <small>SNP</small>	<a href="#">35</a>	<a href="#">1</a>	0	<a href="#">5</a>	
<a href="#">ITGB3</a> <small>SNP</small>	<a href="#">34</a>	<a href="#">3</a>	0	<a href="#">4</a>	
<a href="#">AGTR1</a> <small>SNP</small>	<a href="#">31</a>	<a href="#">1</a>	0	<a href="#">4</a>	
<a href="#">F2</a> <small>SNP</small>	<a href="#">28</a>	<a href="#">2</a>	0	<a href="#">3</a>	
<a href="#">IL6</a> <small>SNP</small>	<a href="#">26</a>	0	0	<a href="#">1</a>	
<a href="#">LTA</a> <small>SNP</small>	<a href="#">25</a>	<a href="#">1</a>	<a href="#">1</a>	<a href="#">2</a>	
<a href="#">FGB</a> <small>SNP</small>	<a href="#">25</a>	0	0	<a href="#">2</a>	
<a href="#">CETP</a> <small>SNP</small>	<a href="#">24</a>	0	0	<a href="#">5</a>	
<a href="#">AGT</a> <small>SNP</small>	<a href="#">22</a>	<a href="#">2</a>	0	<a href="#">4</a>	



1: [Clin Chem Lab Med](#). 2006;44(3):274-81.

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**Associations of apolipoprotein E exon 4 and lipoprotein lipase S447X polymorphisms with acute ischemic stroke and myocardial infarction.**

[Baum L](#), [Ng HK](#), [Wong KS](#), [Tomlinson B](#), [Rainer TH](#), [Chen X](#), [Cheung WS](#), [Tang J](#), [Tam WW](#), [Goggins W](#), [Tong CS](#), [Chan DK](#), [Thomas GN](#), [Chook P](#), [Woo KS](#).

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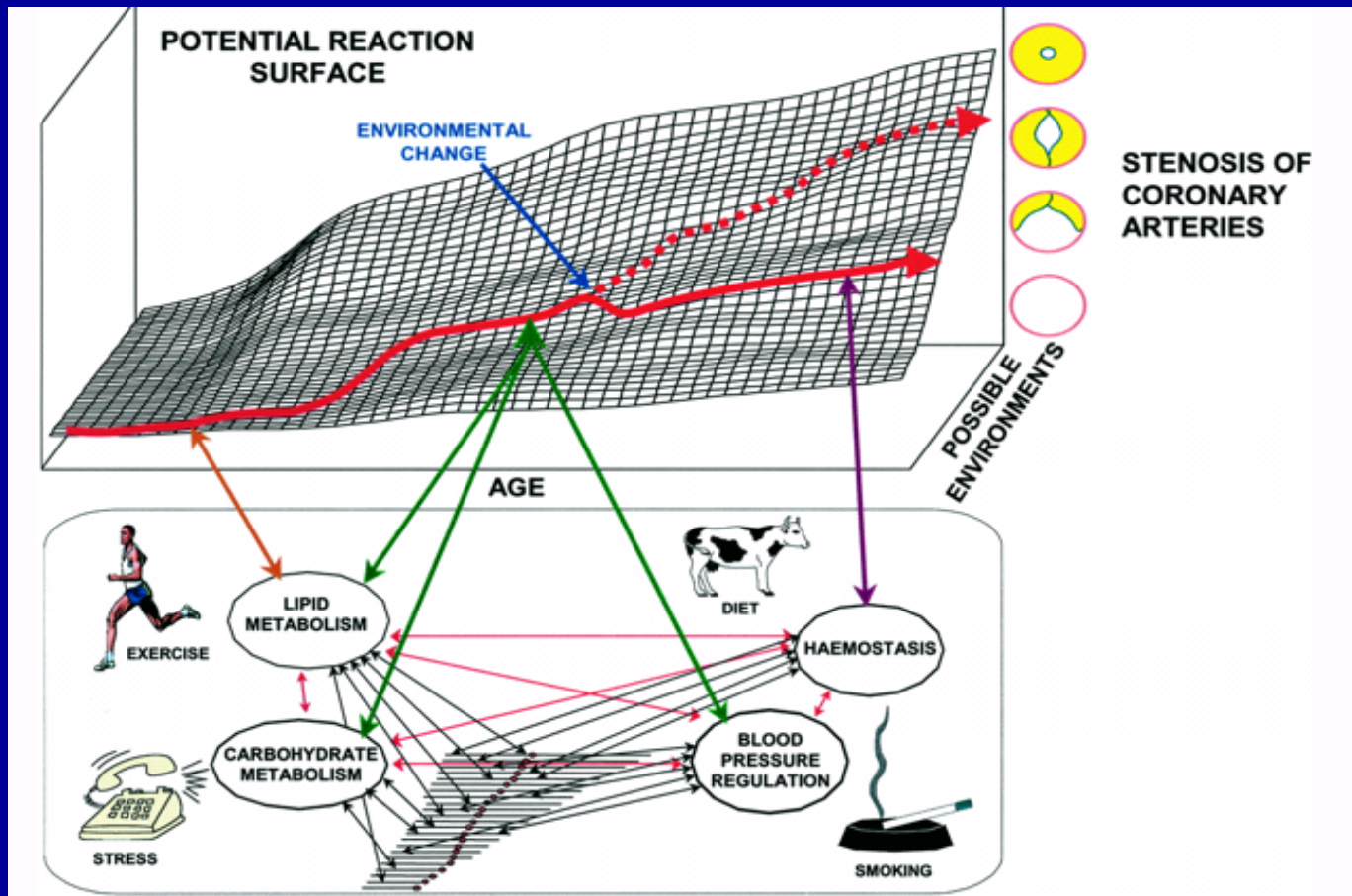
Only one meta-analysis has examined APOE alleles in association with myocardial infarction, concluding:  
“For subjects with either APOE epsilon2 or epsilon4 alleles, LPL X alleles were increased in vascular disease (OR = 2.2, p = 0.01). LPL X alleles displayed opposite tendencies toward association with disease when subjects were divided by sex, smoking, or APOE genotype. Meta-analysis and regression analysis of previous studies supported the sex and smoking dichotomies....”

with each other and with  
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patients, 234 acute  
RESULTS:APOE  
infarction the epsilon4  
06). For subjects with  
01). LPL X alleles  
r APOE genotype.  
LUSION: This is the  
es. Therefore, APOE  
on and ischemic stroke

Publication Types:  
• [Research Support, Non-U.S. Gov't](#)

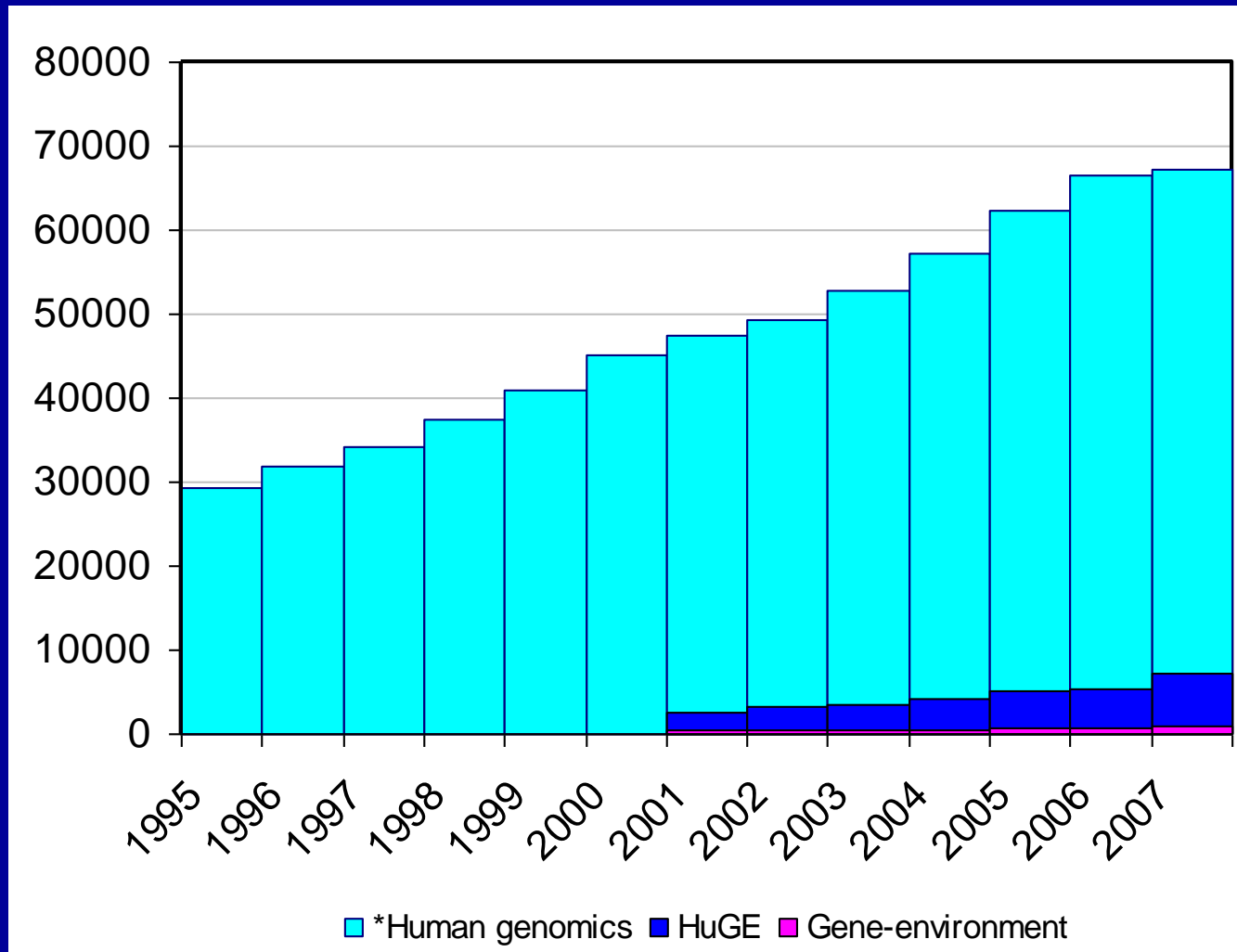
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# Gene-environment interaction in atherosclerosis



Sing CF, Stengard JH, Kardia SL. Genes, environment, and cardiovascular disease. *Arterioscler Thromb Vasc Biol* 2003;23:1190-6.

# PubMed citations on human genetics/genomics and human genome epidemiology (HuGE)



\*PubMed query: "gene or genetic or genome or genomic" / limits: human, year and HuGE Navigator, Oct 2008

# Navigating the Epidemiology of the Human Genome

CCGTCGACTGGAGTGTCTGTGAATTGACTTTTTGTTGCCAGTTGGCAGCGGCAGAAGCAGCAAAGCCCGGCCAACAGCAACAAGCTCCTGCCAGATCCCAAAGCAAACACGGT

## Size up the domain

- population-based research studies
- document data sources, define populations, measure exposures

## Map the topography

- population prevalence, associations, interactions, genetic tests
- use data repositories, controlled vocabularies, standard reporting

## Identify major features

- consistent findings, patterns, and gaps
- promote knowledge synthesis

## Develop navigational tools for further exploration

- reliable knowledge base with applications
- Web 2.0: interconnected, interactive Web-delivered information







# Acknowledgements

CCGTCGACTGGAGTGCTGTGAATTGACTTTTGTGCCAGTTGGCAGCGGCAGGAAGCAGCAAAGCCC GGCCAACAGCAACAAGCTCCTGCCAGATCCCAAAGCAAACACGT

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- Mindy Clyne, curator since 2001

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- University of Ioannina
- University of Cambridge

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